Potential Futures:
An Ethnography of a Familial Cancer Counselling
and Genetic Testing Unit

Amaya Gilson

School of Psychology and School of Population Health
Faculty of Health Sciences
and
Discipline of Anthropology
Faculty of Humanities and Social Sciences
The University of Adelaide

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Declaration

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Abstract

In current Western biomedicine, an accepted and shared belief is that human beings are made up of ‘genes’ and ‘genetic mutations’ which people can inherit from one’s biological parents at the point of conception. Various environmental conditions and factors can also alter genes – thus, genes also mutate over time. With particular genetic mutations identified and considered responsible for illness and disease, inherited mutations are increasingly being used to provide explanations for disease causation and as motivation to reduce or prevent potential associated illness.

In an Australian familial cancer and genetic testing unit, ethnographic fieldwork was conducted over twelve months between 2010 and 2011. In this unit, twelve members of staff worked with approximately nine hundred clients a year referred because of a suspected inherited familial cancer risk. There were four main clues that suggested cancers could be familial, and so warranted referral to the Unit: cancers involving several family members, early age-of-onset of cancer (less than fifty years), multiple cancers in one family or “unusual cancers” (e.g. male breast cancer). Such experiences indicated that cancer was not due to “chance” or “similar environmental upbringing” (like the majority of cancers), but rather due to the genetic inheritance of a “mutation” that was sometimes identifiable by a blood test. The clinical geneticists and genetic counsellors in the Unit were powerful gatekeepers of information. Knowledge and power were exercised in various ways: deciding who became a client, and involving the clients in providing various information including medical histories, genetic material and sometimes the contact details of particular biogenetic relations. Regardless of whether genetic testing was offered to clients, clients received information about their familial cancer risk that
sometimes included information about the precautionary risk-reduction measures they could undertake. These included undertaking regular surveillance screening, the surgical removal of body parts (e.g. prophylactic breast surgery) and family planning measures. In the majority of cases, clinicians offered genetic testing to clients who met their criteria and in whom the clinicians predicted a mutation could be found. Even when results identified a familial cancer mutation, there was no way of telling if, when, and which individuals would develop familial cancer. Familial cancer risk was predictive and not prescriptive, an ambivalent diagnosis that caused and supported the ongoing engagement by the clinicians and the clients around what the diagnosis could potentially mean.

The thesis demonstrates the uncertainties and potentialities surrounding the work of familial cancer risk. It critically examines how the clinicians (in particular) performed and propagated particular ideas and information. There were various complexities present in the work of the Unit including tensions, contradictions and paradoxes, which were a direct result of the uncertainties surrounding familial cancer risk. I demonstrate and analyse the particular ways in which the complexities and uncertainties were experienced and expressed by the clinicians, clients and visitors of the Unit. This thesis adds to medical anthropological knowledge by examining the complexities and uncertainties that surround genetic information and material as both shared and separable; the clinical emphasis of “genetic counselling”; genetic and social families; a client’s health status when considered at risk of familial cancer; the Unit’s formal practice of risk notification; the right to be informed and not informed; and staff competition surrounding mutation identification which reflected engagement with uncertainty and was linked with personal and broader potentialities.
Important Notes

The abbreviation “Unit” is used to refer to the familial cancer counselling and genetic testing unit throughout the thesis for easier readability.

The names of places, organisations, participants and other identifying factors are replaced by pseudonyms in order to protect the anonymity of all participants. The removal and modification of identifying information in order to conceal the identities of all the participants was an ongoing process. Confidentiality issues were discussed with the staff of the Unit both prior to undertaking the research and subsequently discussed as necessary. To further help protect the identity of the participants, a one-year embargo has been placed on this thesis.

At the time of writing (and its subsequent readings), the ethnographic findings, occurrences and descriptions have occurred in the past. Thus, this thesis is written in the past tense rather than an ethnographic present. Writing in the present tense carries more authority than the past tense can evoke, but it implies a view of the community, and the events, ideas and behaviours as frozen in time, predictable, rule-determined and unchanging (Murchison 2010, p. 207; O’Reilly 2005, p. 137; Sanjek 1991, p. 612). By writing in the past tense, I instead emphasize the historical nature of the findings and therefore the potential for change (Murchison 2010, p. 207).
Chapter 1. Situating the Field: Introduction and Background

“It’s all in the Genes”…. or not?

It is a bit like flipping heads and tails, so half of your genetic material came from mum, half from dad, and we have two copies of every gene so really we would be looking at this gene. It is a case of whether or not when your mum made eggs that became you, did you get her good copy or that bad copy, and that is where that fifty-fifty chance comes in, and it’s just like tossing a coin. (An extract from a genetic counselling clinic appointment)

The quote above provides an example of the belief system held and shared by the clinicians of the Unit in which I conducted my research. Recorded during a genetic counselling clinic appointment (commonly referred to as “clinic”), the conversation involved a genetic counsellor informing their client about their chance of having inherited a familial cancer mutation that had already been identified in the client’s mother. The description the genetic counsellor provided to the client demonstrated a belief in the failure of minute biological material (in the form of genetic material) and hereditary factors as aetiological explanations for illness. The quote also demonstrates the underlying notion that people are created from both “good” and “bad” genetic material and that half of a person’s genetic material comes from their mother and half from their father at the time of conception. These notions were not unique to the Unit. Gibbon’s work (2007, p. 53) also described “good” and “bad” terminology used by a practitioner during a breast cancer risk assessment clinic in Britain. The notion that each parent provides one half of a child’s composition reflects the classic Western biomedical understanding of the “blood” relative, where relatives share “common biogenetic heredity” (Schneider 1980, pp. 25, 26).
Humans have, across time, constructed diverse aetiological theories, or belief systems, around the cause of illness to explain their suffering. Some of these theories include: witchcraft, invasion by spirits, the punishment by a deity, and negative spatial environments (Finkler 2001, p. 259; Foster 1976, p. 775; Hahn 1995, p. 5; Kleinman 1988, p. 25). The belief system shared by the genetic counsellor above reflected and reproduced common cultural understandings in Western biomedicine. Western biomedical aetiological explanations usually revolve around the failure of biological material, invasion by pathogens, contamination, trauma, stress, aging, and hereditary factors (Finkler 2001, p. 259; Kleinman 1988, p. 9).

In the Unit where I commenced fieldwork in 2010, the concept of genes and the notion that human beings are made up of things called genes was widely ingrained and accepted, which reflected and contributed to notions held by the discipline of genetics, Western biomedicine and popular culture. I gained knowledge about what genes and mutations were and why they were important to the clinicians, mainly through the observation of the clinicians’ work, and their interchanges with fellow staff, their clients and visitors to the Unit.

During an interview with John, the Head of the Unit and a clinical geneticist, I was informed that each person is made up of “approximately 20,000 genes”. There are two copies of each gene, one from a biological mother and one from a biological father. Each gene has a “life story”, starting “somewhere”, coming from a myriad of ancestors who come together in one particular person at one particular time. Genes not only go back in time, they also project out into the future as any children born will start the “unravelling” of the gene. I learnt that each gene could change as it moves through each
person, resulting in inherited mutations. Every cell is said to have its own mutations that accumulate, and when they eventually all “fray”, it results in the death of the person.

I was also informed that in the majority of human beings, there are cancer-preventing genes. In individuals and families that experience cases of familial cancer, there is, instead, an error, called a mutation in a cancer-preventing gene. The mutation occurs when a gene is either missing or is in the wrong place causing the gene to not work properly. The mutation was already present in the egg or the sperm at conception, and copied into every cell of the person’s body, which meant that the cells in the body were considered to be “a step closer”, to becoming cancer in comparison with people without the mutation. The key point that was emphasised to me, and also to clients during their clinic appointment, was that the genetic mutation by itself was not enough to start cancer, but it meant that the person with the mutation was more likely than others the same age to have certain types of cancer.

_Cancer and Familial Cancer_

According to material produced by the Unit, cancer occurs when there is unregulated cell growth that interferes with the function of the gene. Of all cancers that occur, 90-95% can be termed “sporadic” and are said to be due to external factors (e.g. climate patterns, toxic environments, infections, human behaviour and lifestyles). The remaining cancers, between 5% and 10%, are classified as familial, and said to be due to an inherited mutation in the genes that normally protect it from cancer. The majority of familial cancers (60-70%) result in breast, bowel, and endometrial cancer. Where a mutation is identified, there is no way to tell if, or when, a cancer will occur and, depending on the mutation, the suspected risk of going on to have cancer is anywhere between 1% and
85%. This huge variation reflects the limitations to genetic knowledge in not knowing why some mutations result in cancer and others do not – what is the cause or the “switch”.

Genetic knowledge is complex and because much remains uncertain, research in the area is constantly changing. On the one hand, people born with familial cancer mutations have an increased chance of cancer because “it’s all in the genes”. Developments in the field of epigenetics, however, consider that whether a person with a familial cancer mutation will experience cancer, or not, as not “all” related to the inheritance of a mutation. Instead of the opinion that the mutation in the gene results in cancer, epigenetics argues that the various environmental conditions are the “switch”, or the cause of cancer. Such environmental conditions include both those external to the body (those already mentioned) and in the internal “cellular environment” (so called “junk DNA” of proteins, methylaion and bacteria) (Hubbard & Wald 1999, p. 9; Strohman 2001, p. 8). Social researchers are also arguing that multiple variables are involved. The complexity, uncertainty and “nonlinear multidirectional biological pathways” of epigenetics are explicitly recognised and highlight the “inseparable entanglement of the material body with environmental, socioeconomic, political, and cultural variables” (Lock 2012, p. 130). The uncertainty and variation surrounding the knowledge and practice in this area, demonstrates some of the complexity faced by the clinicians involved in the prediction, judgement and explanation of clients’ familial cancer risk, and what it could possibly mean for the client and their relations.

In order to situate the following chapters and arguments, this first chapter has begun by introducing aspects of the familial cancer explanatory model the staff members of the
Unit valued and practised. The belief systems practised by the Unit developed from, and contributed to, the belief systems in biomedicine. The term biomedicine refers to a primary focus on human biology, in particular physiology and pathophysiology (Hahn & Kleinman 1983, p. 306). In order to understand where such beliefs and practices stemmed from, a brief history of biomedicine, the new genetics, preventative medicine and risk, is also provided. The final section provides description of each chapter and overview of the main arguments and contributions of the thesis.

Biomedicine and the Hospital
Prior to the 18th century, hospitals were institutions of charity and welfare, where physicians trained on the poor, using them as “objects of instruction” (Foucault 1973, p. 84). Biomedicine became the dominant medical theory and form of health care that emerged in Europe during the 19th century and spread throughout the world along with the hegemonic rule of Western society (Hahn & Kleinman 1983, p. 305; Van Der Geest & Finkler 2004, pp. 1996, 1997). The hospital became a professional institution where medical practitioners generated medical knowledge through the observation, categorisation, division and organisation of human bodies according to their illness (Starr 1982, p. 148). While diseases were classified and treatments introduced, the modern hospital evolved from taking care of people to treating people (Foucault 1973, p. 169; Singer & Baer 2007, p. 141). At this time, the human body became the focus of medical attention, known as the “medical gaze” which produced knowledge about what could be considered normal and abnormal (Foucault 1973, pp. 9, 14). Further to how patients were classified and treated, Hahn and Kleinman (1983, p. 311) define biomedicine as a socio-cultural system with five features; 1) a distinctive domain and system of ideas that make
up medicine, 2) a division of labour (for example, doctors, nurses, public servants), 3) corresponding roles, 4) rules of practice and interaction, and 5) institutionalised settings.

The majority of my fieldwork was undertaken in the Unit, which resided in Fairbank Children’s Hospital. In a State capital city in Australia, Fairbank Children’s Hospital was the State’s only hospital that exclusively provided obstetric services and the health care of children and adolescents. The Hospital was an amalgamation of buildings, built up over many years. It consisted of many divisions and units including paediatric medicine, surgery, obstetrics and gynaecology, neonatology, medical imaging, mental health and allied health. Fairbank Hospital contained all five features of the socio-cultural system mentioned above. Apart from the large number of people in Fairbank hospital that either accompanied children or who were pregnant, other things made it apparent that the hospital specialised in obstetrics and the health care of children. Bright, colourful walls and pictures drawn by children decorated the otherwise plain hallways and also areas undergoing refurbishment (see Figure 1, next page). It was obvious to visitors that they were in a hospital set up for obstetrics and the health care of children. There was a large playground in the centre of the hospital (see Figure 2, next page), a gift shop selling baby paraphernalia, and it was common to see clowns walking the halls, there to entertain and lift the spirits of sick children. However, all of these signs that indicated the specialisation of women and children’s health in the hospital did not extend to the Unit in which my research was conducted, because the Unit did not specialise in the healthcare of pregnant women or children.

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1 As stated in Important Notes, this name, and all names of places and persons, are pseudonyms.
2 The clowns, who made jokes and performed tricks, were doctors who volunteered their time to “spread cheer” to sick children throughout the hospital.
Figure 1. An area undergoing refurbishment decorated by children staying in the hospital.

Figure 2. A large outside playground in the centre of the hospital.
The patients of Fairbank were different from the service users of the Unit and this was immediately evident in the terminology used to describe them. The children and women who attended the hospital were considered to be sick, or in need of particular forms of medical observation and/or intervention (e.g. to monitor pregnancy). Classified as either “inpatients”, or “outpatients”, the former needed to be admitted and stay overnight for observation and/or treatment, while the latter described those who attended appointments and received diagnosis and/or medical treatment without the need for admission to the hospital.

To be explained in Chapter 4, the people who accessed the services of the Unit were different from the patients mentioned above. Although they accessed genetic counselling provided by the Unit in an outpatient area of Fairbank Hospital (and other related hospitals), the majority were not considered as patients but were termed “clients”. Being termed a “client” reflected that the majority of the Unit’s referrals were of individuals not seen in relation to the diagnosis or treatment of a current illness; rather they sought to explain past experiences of cancer and to obtain information about future familial cancer risk and management.

One of the first ethnographies on a biomedical hospital conducted in 1962 considered going to hospital like going to an exotic island (Coser 1962, p. 3). Filled with liminal spaces, the hospital is where patients, cut off from the outside “normal” world, undergo another regime, by dressing differently, inhabiting other roles and forging new identities (Coser 1962, p. 3; Long, Hunter & Van Der Geest 2008, p. 73). The patients of Fairbank Hospital all underwent transformations of some kind during their stay – a pregnant woman left as a mother, or a child who had entered with a broken arm left with a cast.
For clients of the Unit, transformations were also apparent because in learning of their risk of familial cancer, and/or their mutation status, there were impacts on how they viewed themselves and their relations (see Chapter 5 & 6).

A more recent and dominant view of hospitals considers them as shaped by rules and ideas from outside and, as such, are places where the core values and beliefs of a culture are both reflected and reinforced through the practice of biomedicine (Long, Hunter & Geest 2008, p. 72; Van Der Geest & Finkler 2004, p. 1996; Zaman 2005, p. 2026). Long, Hunter and Geest (2008, p. 73) consider both perspectives, “hospital-as-island” and “hospital-as-culturally-embedded”, as correct. They state that in order to understand the complexity of hospital systems and practices and allow for correct portrayal, ethnographers should account for such ambiguity of the hospital.

In my fieldsite, there were various ambiguities that support the perspectives by Long, Hunter and Geest; complexities which will be discussed throughout the thesis. The reason for such tensions and contradictions stem from biomedicine’s theoretical and clinical efforts that are constructed in dichotomies (Dossey 1984, p. 15). Having also been described as dualisms in ways of knowing, the main dichotomous relationships found in biomedicine include: the classic Cartesian dualism of body and mind, which involves the distancing of the body from the person; ideas of the passive patient and expert physician; health and illness and rationality and irrationality (concerning patients adhering to particular guidelines given by medical professionals); nature versus nurture; subjective awareness versus direct observation; and male versus female (Kirmayer 1988, p. 59; Lock 2012, p. 129). A number of these dichotomies will become relevant throughout this thesis.
In addition to such classical understandings, anthropologists and sociologists have uncovered and discussed dichotomies, tensions and contradictions present across various practices of biomedicine. For example, Joanna Latimer demonstrated the ambiguity of genetic medicine, arguing that because genetic science is a new frontier where so much is not yet known, or not yet standardised, undecidability, uncertainty and instability of a genetic diagnosis were prominent features of clinical practices in dysmorphology. However, the ambiguity was evident because of the firm commitment to a future of diagnostic certainty (Latimer 2013a, p. 202), an argument also presented in this thesis. Latimer described and analysed the moments when a genetic diagnosis was accomplished and when it was not accomplished (2013a, p. 103). Her work demonstrated how two contradictory notions surrounding bodies and persons play out in the genetic clinic where diagnosis is often elusive and where clinical practices are undecidable and involve explicit deferral (Latimer 2013a, pp. 134, 165).

**The New Genetics and Preventative Medicine**

The speciality area of genetics, which is part of biomedicine, has a long history. There is considered to be a marked separation between the older practice of genetics, and the current practice which has been re-defined by some as the “new genetics”, consisting of measures taken to prevent diseases rather than treating their symptoms (for example, to prevent cancer from occurring) (Finkler 2001, p. 235; Latimer 2007, p. 99; Palsson 2007, pp. 6–14). The new genetics arrived at the beginning of the 21st century with the undertaking of the Human Genome Project (HGP), which was hoped to lead to cures for all diseases (Palsson 2007, pp. 6–14). This demonstrates how the potential of the HGP to dramatically improve health was linked with the potential of genes (Taussig, Hoeyer & Helmreich 2013, p. 8).
Beginning in 1990 and completed in 2003, the HGP was an international research effort with contributors working from universities and laboratories, throughout the United States, United Kingdom, France, Germany, Japan and China, to determine the DNA sequence of the whole human genome. With a number of objectives to achieve, the first draft of the human genome was published in 2001, with the full sequence said to be completed and published in 2003 (National Human Genome Research Institute 2012). When the majority of the human genome was published in 2001, Francis Collins the director of the National Human Genome Research Institute (NHGRI) noted:

the genome could be thought of in terms of a book with multiple uses: It’s a history book – a narrative of the journey of our species through time. It’s a shop manual, with an incredibly detailed blueprint for building every human cell. And it’s a transformative textbook of medicine, with insights that will give health care providers immense new powers to treat, prevent and cure disease. (Collins in NHGRI 2012)

One of the achievements of the HGP was the discovery that there are around 30,000 human genes. This, along with other discoveries, contributed to a resource of detailed information about the structure, organization and function of the human genome and other functional elements found in DNA (NHGRI 2012). The scientists involved with mapping the human genome at the time labelled 98 per cent of the DNA identified as “junk”, because they had an assumption of how life was assumed to work and because this “junk” did not conform, it was not included. Since the completion of the project, it has been made apparent by studies in epigenetics, that what was previously labelled as “junk” is considered far from “junk”, having functional significance by acting as a genetic “switch” to regulate the expression of genes (Lock 2012, p. 134).

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3 A genome is said to be a set of DNA instructions where the DNA contained within each cell carries instructions needed to build and maintain the many different types of cells that make a human (NHGRI 2012).

4 Where page numbers are absent from the in-text references in this thesis, they have not been provided by the online source.
Such developments and arguments demonstrate how social, cultural, political and economic activities are entangled with the ever-transforming “molecularized body” so that human behaviour is modified by biology and environmental biology (Lock & Nguyen 2010, p. 317).

The attempt to predict health and illness, which is the work of the new genetics more generally and specifically in the Unit, is not new. There are countless historical and current practices of predicting health and illness across various parts of the world. Just some include the consulting of shamans, priests or oracles, with diagnostic tools including astrology, tea reading and dream interpretation. The present scientific and genetic projects that are hunting for eternal youth and longevity are not too different from the “mythological fiction”, the practice of magic that dominated in the past (Boia 2004, pp. 9, 10, 104). Despite the cultural variations, the goal of such practices is the desire to understand and explain the present and/or to foresee the future (Hubbard & Wald 1999, p. 13).

The HGP forecasted great advances in identifying genetic factors that contribute to (and thus predict) common and complex diseases such as cancer and diabetes, and then in finding cures for these diseases through drug therapy and other forms of treatment and prevention (Daiger 2005, p. 362). Moving from a reactive to a preventative practice, the HGP aimed to identify, remove or manipulate a “defective” gene before its negative effects manifested, thus ultimately prolonging human life (Finkler 2000, p. 49). Although the removal or manipulation of genes has not come to fruition, other forms of preventative medicine have been practised as a result of the HGP, such as new technologies, medical surveillance or surgical removal of body parts, lifestyle changes
and reproductive decisions. The social and political factors implicated in disease occurrence are argued by some (Lippman 1998, p. 73; Lock & Nguyen 2010, pp. 75, 77, 99, 314; Petersen 1998, pp. 64–66) as being deflected by the bioethical attitudes regarding the “right to know”, “informed choice,” and the assessment of risks and benefits associated with various medical interventions.

For the clinicians of the Unit, they attempted to explain past occurrences of familial cancers and to predict future cancer experiences. Gross and Shuval argue, “defining an individual’s present health status based on the evaluation and calculation of probable futures … favours prognostic data and predictive knowledge over the more classical pair of diagnosis-therapy” (2008, p. 551). The tools the clinicians used to predict familial cancer risk included the information received about the client and their biogenetic relations, knowledge held about human genes, and sometimes the results received from genetic testing and/or Next Generation Sequencing (NGS) machines. NGS machines are considered as resulting in and enabling major advancements in mutation identification (Marx 2013, p. 263).

A presentation in September 2014 by a group of leading professors and clinical geneticists shared and demonstrated further potentialities involving NGS machines to the State’s medical specialists.⁵ It was explained that with NGS machines clinicians would no longer need to order individual mutations to be looked for and when they were not found to keep ordering different tests until something was found. As Gibbon et al. (2014, p. 2) have very recently predicted, with NGS machines becoming more efficient and cost

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⁵ Although fieldwork in the Unit occurred between 2010 and 2011, I collected relevant data on changes to genetic knowledge or practices up to the point of thesis submission. In this case it involved attendance at a presentation held in 2014.
effective, it is now cheaper for the client’s whole genome to be searched for mutations rather than just looking for specific mutations. One of the speakers at the presentation mentioned above, highlighted how the use of NGS machines will result in “diagnostic categories” being provided rather than one diagnosis. Not only would the massive quantity of data produced require storage and raise questions about data interpretation, but the use of NGS machines was resulting in two changes to the future practice of genetic testing. The first was that consent forms were being reviewed and would need to ask the client if they wanted to know about the “incidental findings” (risks for illnesses the client did not expect would be found). The second was that with the NGS machines scanning the whole genome, medical specialists (such as heart surgeons for cardiovascular disease, or oncologists for familial cancer) on suspicion of a familial disease would be able to “bypass” the genetic counselling services to order and interpret the results of the “diagnostic categories”. More questions were raised by the presenters than answers were provided, demonstrating how the clinicians were required to catch up with the technological advancement of the NGS and predict the possible positive and negative implications. In relation to the study of new genetic technologies, Jackson (2002, p. 333) suggests that rather than debate how new technologies are good or bad for society, rather the focus should be on revealing the attitudes towards them, how the effects of new technologies are evaluated and managed, and how all those involved experience and interact with the technology.

**Implications of the New Genetics**

As a result of the HGP, new technologies, institutions, resources, diagnostics and treatments, immunotherapy techniques, power relations, practices and ideologies have been built around the concept of “genes”. This is evident in the quote: “We used to think
our fate was in the stars. Now we know, in large measure, our fate is in our genes” (Watson 1989 cited in Jaroff 2001). The new genetics has been noted for its ability to redraw boundaries around what constitutes sickness and kinship. Genetic evidence about who is related to whom adds an entirely new set of “experts”, such as geneticists and bio-ethicists, to already longstanding debates by archivists, historians and anthropologists. Brodwin (2002, p. 325) predicts that the evidence produced by these “new experts” may not make the argument about who is related to whom any easier but rather might make it harder.

In the work “Future Imaginaries”, Joan Fujimura describes both laboratory experimentation and imagination as regular practices in which scientists engage, stating: “imagination is a social practice deployed in the production of science and technology” (2003, p. 176). Through the biological gaze and imagination, the new genetics is radically transforming social and cultural understandings and imaginations of what it means to be human, along with understandings and imaginations of kinship, property, health, illness, relatedness and personhood (Palsson 2007, pp. 13, 212). An analysis of the new genetics underscores what Foucault (1988, p. 18) called the “technologies of the self”, practices undertaken by individuals to improve themselves, within institutional frameworks of power. Such notions and practices, involving the improvement of people’s health through genetics, are regularly likened to the eugenics movement linked with Nazi Germany in the early twentieth century, which intended to improve “genetic legacy” (Boia 2004, p. 128; Lock 2002, p. 249). A tension between “both the perils and the possibilities that spring from genetic technologies” has also been highlighted and termed as “flexible

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6 James Watson (and Francis Crick) won the Nobel Prize for solving the complexity of DNA structure.
7 Although eugenics became renowned after its use by Nazi Germany, Francis Galton founded the term and work (in works produced in 1869 & 1889), which sought to remedy the “degeneration” of Western civilisation (Boia 2004, p. 128).
“eugenics” (Taussig, Rapp & Heath 2003, pp. 60, 61). The current preventative practices are said to be overtly and subtly contributing to “the new eugenics” (Hubbard & Wald 1999, p. 23) and a “utopian eugenics” (Kitcher 1996, p. 203, 204). Unlike past eugenic practices, the new genetics delivers the message of population control not through population coercion but rather through emphasising individual choice through genetic counselling (Petersen 1998, pp. 63, 64).

**Being at Risk and Embodying Risk**

The hallmark of the new genetics is based upon the calculation of current and future health risks (Everett 2007, p. 377; Finkler 2003, p. 51; Konrad 2003, p. 27). It has been argued that as a result of the HGP, boundaries have been redrawn in relation to what constitutes sickness, potentially making all people possible carriers of genes that could malfunction and cause illness at any time (Finkler 2003, p. 57; Finkler, Skrzynia & Evans 2003, p. 408). Lock argues that as a result of the HGP a new space has emerged where there is anxiety about the future with respect to disease (2012, p. 135). The effect of risk is its ability to hold people accountable (Douglas 1990, p. 1), a new form of self-surveillance that results in chronic anxiety and “hyper-rationalism” (Castel 1991, p. 289). Self-surveillance is evident when a person uses the results of a genetic test to make lifestyle changes (such as through taking prophylactic surgery measures and making reproductive decisions) (Lock 2012, p. 136).

The increase of genetic testing in research and clinical practice is raising questions about “concomitant transformations in kinships, human affiliation, including biosociality, and new forms of citizen”: topics that are increasingly being recognised by medical anthropologists (Lock 2012, p. 130). The idea of the genetic basis of disease has been
argued persuasively against and has constituted what Goodman, Heath and Lindee (2003, p. 2) term a “techno cultural revolution”, Lippman (1992, p. 1470) as “geneticization”, Rabinow (1996a, p. 99) as “biosociality” and Hubbard and Wald (1999, p. 3) as genetic “reductionism”. Concerns surround the possibility of discrimination and reinforcement of racism and social inequalities grounded in DNA differences (Lock 2012, p. 137). Concerns which contribute to the reluctance of people when considering genetic testing can include the potential for discrimination, costs, confidentiality of genetic information, insurance cover and employment difficulties (Peterson et al. 2002, p. 79). Latimer however, considers the alignment between the new genetics and the clinic as offering different ways of viewing the body, which may extend the “possibilities for the performance of medicine” (2013b, pp. 24, 25). The Unit in which I did ethnographic fieldwork lay at the intersection of these debates and practices surrounding the new genetics.

Overview of the Thesis: Knowledge, Uncertainty and Potentiality
This thesis is about the various complexities, uncertainties and potentialities surrounding the practice of familial cancer, which became evident through the forms of tensions, ambiguities and contradictions. I have chosen to weave examples of these throughout the nine chapters because this was how they presented and emerged in the field, not as straightforward occurrences but complexities malleable and interconnected with the everyday practices of staff from the Unit.

During my time in the Unit, I gained a plethora of knowledge around familial cancer. Rather than my understanding becoming clearer with the more I learnt, instead my understanding of familial cancer risk became further complicated and uncertain.
Throughout my thesis, I demonstrate how knowledge surrounding familial cancer resulted in uncertainty, not only for me, but for clients of the Unit, visitors to the Unit and the staff of the Unit. This discovery is central to my argument because it demonstrates the complex and ambiguous nature of the topic. This first chapter, “Situating the Field: Introduction and Background”, has introduced aspects of the familial cancer explanatory model as practised by the staff members of the Unit. In order to understand where such beliefs and practices stemmed from, this chapter has also provided a brief history of biomedicine, the new genetics, preventative medicine and risk.

While conducting ethnographic fieldwork, various complexities, tensions and uncertainties surrounded my role. A discussion of these is provided early in the thesis (Chapter 2), because how participants from the Unit perceived me, and how I experienced the field, affected and illuminated the focus of this thesis. Titled, “Situating the Field: Fieldwork Conditions and Relations”, I first provide a historical overview of the establishment of the Unit, detailing how the Unit came to be, what it set out to achieve, and how it evolved. I then detail the early processes that I was required to undertake and negotiate in order to gain entry into the Unit. The chapter also highlights ongoing consent and rapport issues due to a paradigm clash between me as an anthropologist and my participants in the Unit, and the reality of “procedural ethics” versus “ethics in practice” (as used by Guillemin & Gillam 2004, pp. 263–264). The “procedural ethics” guidelines which established what, where and how I was to do things (which came from the research proposal, ethics applications and further documentation) did not influence what occurred in the field in the way they were intended. The “procedural ethics” were not conducive or reflective of the “ethics in practice” of ethnographic research which: “unfolds according to its own temporality and logic: that is, following the contours of social life as these are
revealed by the persons with whom one engages in the field” (Guillemin & Gillam 2004, pp. 263–264; Simpson 2011, p. 381). In practice, what I could observe was negotiated by the clinicians, those with the most powerful positions in the Unit. These processes illustrate some of the complexities and uncertainties involved with undertaking fieldwork in a biomedical environment and how earning the right to know certain things, was entangled with, and reflected, the complexities surrounding working with familial cancer risk.

The third chapter “Staff Roles and the Working Environment”, provides a snapshot of the staff’s work roles and the working environment in the Unit. Apart from situating the Unit, the chapter demonstrates how different roles affected practices of familial cancer work, in specific ways. These include an emphasis on clinical work, group hierarchy, team building, work satisfaction, and the use of humour and gossip through informal and formal debriefing. The Unit’s role was to provide genetic counselling and genetic testing to people considered at high genetic risk of developing familial cancer. A General Practitioner (GP) or a medical specialist (such as a surgeon) referred clients to the Unit (see Chapter 4 for detail). There were four main clues that suggested that cancers could be familial and so warranted referral to the Unit: one, if several family members either had cancer at the time of referral or had cancer previously; two, if there had been an early age-of-onset of cancer in a family member (usually under fifty years of age); three, if there had been multiple cancers in one family member; and four, if there was an “unusual cancer” (e.g. male breast cancer). It was said that if an individual or family had any, or, all of these clues, they might be considered to be at increased risk of familial cancer. Most of the Unit’s clients were aged over eighteen because the majority of the familial
cancers have an adult onset. The majority of the service’s clients were female and considered to have an Anglo-Saxon background with very few considered as culturally and linguistically diverse (see Chapter 4).

In Chapter 4, “The Construction of Clients, Families and Familial Cancer Risk”, I examine the process involved in becoming a client of the Unit. The chapter describes the intake, division, diagnostic and classification processes relating to clients. Such practices resulted in particular reproductions around what a client, a family, and familial cancer risk estimates were (and were not). The majority of the Unit’s clients were not considered to be “sick” with familial cancer, but instead considered to be at risk. The clients of the Unit were “enrolled” in the process, providing various forms of information in the search for expert genetic explanation about past experiences of cancer, and/or to obtain information and possible genetic testing about future familial cancer risk.

The clinicians were in charge of deciding which referrals suggested a possible increased risk of familial cancer. As discussed in detail in Chapter 5, “Genetic Counselling, Knowledge and Power in the Clinic”, after receiving a referral, the clinicians reviewed the information received about the history of cancer in the individual client and genetic relatives. Clinical judgement and discretion was then used to decide if the client had an increased risk of familial cancer by distinguishing between familial, and non-familial, experiences of cancer. If the client’s familial medical information indicated a high risk of familial cancer they became a client of the Unit. With the majority of the clients not suffering from cancer, the clinicians did not undertake any biomedical treatments.

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8 Some conditions have an early age onset (e.g. familial bowel conditions), and so medical surveillance or surgery was recommended before the age of sixteen to remove the early onset of tiny growths called “polyps” from which most bowel cancers develop.
Instead their role primarily involved offering and undertaking “genetic counselling” and sometimes genetic testing. As long as the clients met the Unit’s criteria, they were exempt from paying for the services provided by the Unit.

Genetic counselling appointments, termed the “clinic”, was where a client’s familial history of cancer was reviewed and where clients were provided with up to date information about familial cancer, including their possible risk of developing familial cancer in the future and the risk for certain biogenetic relations.\(^9\) Largely a medical practice rather than a psychosocial practice, the clinicians provided their clients not with biomedical treatments, rather, with information about reducing familial cancer risk by undertaking surveillance measures considered relevant to the client. Thus, the information was provided to the clients, and then the clients were responsible to make various decisions around their own (or their family’s) health care (see Chapters 5 & 6).

Chapter five also reveals what occurred in clinic. Information provision in the clinic involved identification, communication of the prognoses (the prediction of familial cancer risk), and the possible implications of being at increased risk of familial cancer. Clients typically sought confirmation and certainty about their risk, which the clinicians were not able to provide. This chapter explores how the clinicians emphasised the positives of knowing one’s increased risk, which reflected clinicians’ belief in the power of the information they provided to impact positively on their clients. It also substantiated their reason for being and practising as a service. Clients were expected to use the information provided to take responsibility and make decisions concerning if, and to what

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\(^9\) To avoid confusion and to clarify meaning, rather than use the service’s general term of “family member” and/or “families”, I use the term “biogenetic relation”, “biogenetic relationship” or “biogenetically related” to describe the relationship.
extent, they would try to minimise or prevent a potential experience of familial cancer. Regardless of the risk category provided to clients, there were always degrees of unknowns and ambiguity as knowing of an increased risk was considered “both a threat and an opportunity”. Although all of the information was uncertain as the clinicians could not predict if, who, and when familial cancer could occur (due to the very nature of risk), the clinicians distinguished between sharing some of the known unknowns, those that were “good” for the client to know, and not the unknowns that were considered not necessary to share. The clinicians and the clients revealed various extents of knowing, of wanting and being able to know certain things. The power to include and exclude information surrounding the client’s familial cancer knowledge, their risk and possible implications, revealed and continued a tension between doubt and certainty, and medical discretion and judgement.

In some circumstances during the clinic, the clinicians offered genetic testing to identify the “genetic abnormality”, most commonly referred to as a “mutation”, to clients who met the criteria for referral, and/or in whom the clinicians thought a mutation was likely to be identified. In clients offered genetic testing, there was only a one in five chance of finding a mutation. The clinicians wanted their clients to be identified with mutations for various reasons. As explored further in Chapter 6, “Post Clinic and Beyond: Mutation Identification”, the clinicians believed that mutation identification resulted in knowledge that helped their client avoid and/or minimise their, or their biogenetic relatives, experience of familial cancer. Chapter six also describes what happened after a client’s clinic appointment, how genetic testing was undertaken and the provision of genetic testing results. It highlights some of the different ways in which clinicians provided, and clients learnt and responded to, the uncertain information surrounding genetic testing for
familial cancer mutations. The chapter demonstrates and argues how the reality of being informed about the existence of a familial cancer mutation resulted not only in contradictory emotions – but also new concerns and questions over the protection of genetic information and future discrimination. Regardless of the uncertainties, the potential value of mutation identification was emphasised and practised by the clinicians. This notion demonstrates another way in which the work of familial cancer counselling and genetic testing, undertaken by the clinicians, was about possibilities and potentialities. Other sections of the chapter also provide and reflect on the actual usages and knowledges surrounding the term “mutation”, and about the emphasis placed on the biogenetic family.

Certain biogenetic relations of a client considered at high risk of familial cancer were also offered genetic counselling and genetic testing from the Unit. Chapter 7, “The Practice of Risk Notification: Considerations and Implications”, concerns the Unit’s practice of “risk notification” where individuals considered at high-risk of familial cancer, either due to a family history of cancer or an identified genetic mutation, were asked for the contact details of certain biogenetic relations. The Unit facilitated the sharing of the information with at risk biogenetic relations and their medical professionals due to a perceived duty of care to provide potential lifesaving information. It was a process whereby permission was obtained from the proband client (the name describing the first/initial client) for the service to send a letter to the proband’s biogenetic relations considered could to be potentially at high risk of familial cancer. The letter informed the biogenetic relation that a mutation had been found in a “family member” and that genetic counselling and possible testing was available to them to clarify their own risk. The practice resulted in a
slight increase in the numbers of people attending the Unit compared to occasions in which clients were left to inform or pass on letters directly to biogenetic relations.

The formal practice of risk notification required the clinicians to enrol clients in providing consent and provision of contact details, in order for the practice to be considered as adhering to autonomy and confidentially. Both suspecting and unsuspecting relations were contacted by the service and informed about a possible increased genetic risk of developing familial cancer. The clinicians had a great deal of knowledge about biogenetic connections between people, which they termed “families”, and they owned and protected this knowledge. The chapter, about the right to know and not to know, focuses on a number of key, interrelated issues related to the social, ethical and practical implications of risk notification, including implications for clinicians, the public, clients, and consumers. In it, I argue that people’s right to know was considered as greater than their right not to know. On the one hand, the clinicians of the Unit argued that risk notification was a pro-active strategy based on a duty of care, benefiting individuals, families and the community. On the other hand, it can be argued that it was contrary to biomedical ethical principles including privacy and confidentiality.

In the case that the client had a mutation identified, staff appeared energised and engaged, and there was staff comradery and enthusiasm. Conversely, the clinicians expressed disappointment when a mutation was not identified. As detailed in Chapter 8, “Scientific Competition, Discovery and Progress”, the clinicians demonstrated the desire, and need, for mutations to be identified. These were displayed in various ways: from holding competitions involving chocolate frogs as a reward for accurate prediction, to displays of excitement, through to charting the annual number of mutations found.
The prediction, betting about, and finding of mutations excited the clinicians for a number of reasons. With only a one in five chance of finding a mutation, even in carefully selected clients, a clinical geneticist once said: “we don’t expect to find a mutation in our families, we expect not to find a mutation in our families”. Finding a mutation also provided recognition that the clinician involved was “selecting people properly”. It confirmed that they knew what they were doing and confirmed that their ideology was “truth”. By discovering mutations, including mutations considered rare or new, the clinicians were at the forefront of scientific discovery and they used this knowledge, along with their failures at prediction, to progress genetic understanding. This knowledge not only contributed to the individual client’s case and family but it also contributed to scientific discoveries and progressed genetic knowledge, thus increasing the power of the clinicians. As described in chapter eight, competing in biomedicine and amongst the genetics community was not unique to the Unit, with bets previously placed during the HGP over the number of genes in the human genome, resulting in monetary and social rewards for the declared winners. The chapter discusses the local and global competitions and practices of competing in the broader context of scientific discovery and progress objectification and commodification of genetic material. On a local level, excitement over the finding and identification of mutations, and competition through the practice of betting, was regularly displayed by the clinicians of the Unit and expressed in numerous ways that were sometimes explicit and other times implied. The ownership, objectification and commodification of genetic material and knowledge, which had been physically separated from the individual, was largely expressed through competition and betting which, whether local or global, was both collegial and competitive, serving numerous purposes. The betting practices demonstrate one way in which uncertainty and contingency was performed and engaged with (Malaby 1999, p. 158). This proves the
knowledge is not certain despite claims to certainty and value of the Unit. In the concluding ninth chapter, “Conclusions: Potentialities of Familial Cancer Risk”, the key findings and arguments are summarised and future considerations and implications drawn from the ethnography are considered.

**Locating the Ethnography**

This ethnographic thesis, which involved conducting ethnographic research over a year in a biomedical Unit located in my home town, uncovers and makes apparent experiences and findings that would otherwise not have been possible. This was precisely because what, where, when and how the work of the Unit was undertaken, was not visible or open to the public or outsiders. The thesis contributes insights and knowledge specifically about organising, conducting and negotiating an ethnographic study conducted at home and in a biomedical Unit, therefore building upon previous research by medical anthropologists and sociologists on these topics (these include, Murchison, O’Reilly, Parker, Punch, Sluka, Watts & Wind). The major contribution of this ethnography, however, is its work towards an anthropology of contemporary genetic knowledge and practice. Adding to the work by major contributors in the field such as Finkler, Fujimura, Konrad, Latimer, Lock, Lupton, Palsson, Petersen, Rabinow, Rapp, Strathern and Taussig, this thesis shares, situates and analyses knowledge and practices around identities, kinship, risk of illness, property, potentialities and genetics itself, primarily in the local context of an Australian familial cancer counselling and genetic testing unit.

Biomedicine, genes, bodies, populations, social policy, technologies and research undertaken in the clinic and the lab, are “imbued with potential” (Taussig, Hoeyer & Helmreich 2013, pp. 3, 4). Rose (2007, p. 19) has commented on the ability of genetics to
bring the health of an individual’s and family’s “potential futures into the present”.
To be demonstrated and argued in the thesis, the identification of an increased familial cancer risk, the use of risk models and the decision to offer genetic testing constituted a complex process, requiring ongoing negotiation by the clinicians. The ongoing negotiation reflected the complexities of potentially identifying those at high risk of familial cancer – a process entrenched with uncertainties. The negotiation and uncertainties reflected how “the presence of specific genes informs only about potentiality and nothing more” (Lock 2012, p. 158).
Chapter 2. Situating the Field: Fieldwork Conditions and Relations

Taking things for granted is said to be a common problem when conducting fieldwork “at home” (Laavy 2011). Some of the challenges I faced in the field, as detailed below, were directly related to the taken for granted expectations I had about what I would access and experience during fieldwork, and assumptions about my relationships with participants. What I had exposure to in the field in reality, was very different from what I had assumed prior to undertaking fieldwork “at home”, in the same city where I lived. Sometimes it was clear to me why I was allowed to be a part of something, or not, but at other times, the reasons seemed unclear or inconsistent, often contradicting earlier rules and decisions. I negotiated a number of challenges in the field, challenges that I had not predicted prior to entry, because they could not have been predicted.

The main focus of this thesis is about revealing the complexities and uncertainties surrounding practices, knowledges and implications of familial cancer risk, as they unfolded in the Unit. My fieldwork experience was one of an ongoing ambiguous process, an experience that was separate from, but informed by, the complexities and uncertainties surrounding the work of familial cancer. This chapter initially provides a historical overview of the establishment of the Unit which details how the Unit came to be, what it set out to achieve, and how it had evolved. The remainder of the chapter details some of the fieldwork conditions I experienced and the impact they had on my data collection, research focus and findings. With topics including discussions of revelations, fieldwork at home, fieldwork methods, and procedural ethics versus ethics in
practice, this chapter demonstrates some of the complexities encountered while arranging and doing my ethnographic research.

Revelations at Home

So why did I accept you here? Because you are different… (Expressed by John, in a recorded interview)

My point of entry into the Unit occurred through John, the clinical geneticist and Head of the Unit who legitimated my entry. John’s education, position and knowledge about genetics made him a powerful figure in the Unit and wider genetics community. Towards the end of my fieldwork, during a pre-arranged and recorded interview, John provided me with a number of explanations as to why he wanted an anthropologist to conduct ethnographic research in the Unit. As a result of this interview, the expectations and motivations behind inviting and allowing me to conduct research in the Unit became clearer. These expectations influenced and shaped my experiences working in the field, the data I collected and the findings. Although I was there to fulfil my agenda of producing an ethnography, John and the staff of the Unit had more power in our relationship, which they exercised by deciding what I could (and could not) access in the field.

The opening quote from John expressed how it was that I came to conduct ethnographic research in the Unit: to him, I was “different”. During the recorded interview, John explained that my research methods were considered different and it was because of this difference that I was viewed as being of some value. The underlying reason for having an anthropologist in the Unit, which had not been articulated prior to this interview, became apparent. Although John emphasized at that time that he had no expectations of me, what
he chose to emphasize during the interview was in contradiction to this, and was, therefore, very insightful. I was informed at that time that my findings were of interest, but that I was not there to change anything. In the same interview, John stated to me that he wanted all different types of researchers to come to the Unit, explaining:

We haven’t had a poet in space, we haven’t had an artist on the moon. We’ve had engineers and geologists, and I can understand why they need to be the first people on the moon, but we are more than engineers and geologists on this planet.

Through the articulation of this moon analogy, it became apparent that John viewed the Unit like the moon. The Unit was a place removed and in need of exploration, first by those traditionally commonplace and equipped for the role (such as medical professionals like themselves). Then John considered it important that the Unit be explored by those traditionally not associated with the place (such as myself as an anthropologist), because we represented the “other” and contributed something different.

Not only was I considered to be different as I was an anthropologist, but John also considered the Unit as different because the services they provided were unlike other clinical units (discussed further in Chapter 6). This notion of difference was sometimes acknowledged by staff members of the Unit and other health professionals external to the Unit. At various times, staff members stated to me that they had no idea why I was studying them and what I would write, yet in contradiction, they often proudly remarked to visiting medical and genetic counselling students, “she is studying us because we are interesting” or “because we are doing something different”. These claims also demonstrated how the opportunity to be studied was flattering because it gave, or confirmed, a sense of uniqueness. Based on Pope’s experience conducting ethnography in medical settings, he reported that the positive reception he received from all those
involved in his research “partly reflects the novelty of having an outsider profess fascination with the minutiae of your everyday work” (2005, p. 1180). The importance placed on being different or unique affected various aspects of the clinicians’ perceptions of themselves and the services they provided. As discussed in Chapter 8, such a drive to be authentically different (within appropriate boundaries) in an arena of similarities, reflected and propagated the biomedical and scientific competition, where there is a desire to be distinctive as it drives scientific discovery and notions of progress. However, although being considered different was what enabled my entry into the Unit, concurrently being considered different created challenges which are discussed below.

**Coming to Be: Historical Background and Present Practices of the Unit**

The proposed establishment of the State-wide Unit occurred in 1996 by a committee comprised of representatives from clinical genetics, laboratories, teaching hospitals, the Royal Colleges of Medicine and the State’s Department of Health. The main reasons outlined for establishing such a Unit were to address both the growing demand for familial cancer genetic counselling and testing, and to incorporate genetics into mainstream health care delivery. Throughout the thesis, I provide examples of, and emphasise, how the staff members of the Unit considered their work as important and valid in preventing and/or minimising the impact of familial cancer. The development of, and ongoing need for, familial cancer genetic counselling and genetic testing by the Unit, is part of a dominant focus on preventative health and medicine. As described below, the preventative health focus stems from, displays, and contributes to, relationships between the new genetics, biomedicine and the HGP.
Significant Events Fuelling the Unit’s Establishment

Prior to the establishment of the Unit, familial cancer counselling and genetic testing was undertaken as part of a general genetics unit, which worked on all familial disorders affecting human development. According to Unit documents, the decision to create a specific familial cancer service was fuelled by three main events in the early 1990’s. The first event was the identification of four genes: Adenomatous Polyposis Coli (APC) and Hereditary Non-Polyposis Colorectal Cancer (HNPCC), which it was said can account for the early onset of some bowel cancers; and Breast Cancer 1 (BRCA1) and Breast Cancer 2 (BRCA2), said to be responsible for some breast and ovarian cancers. The second event was the widespread publicity about the identification of the two “breast cancer genes” (named above), and the third was the establishment of the National Breast Cancer Centre by the Australian Federal Government.¹⁰

At the time the Unit was established in 1996, the State’s health department agreed to fund the State-wide familial cancer Unit from 1998 to 2003 as it was (wrongly) anticipated that the backlog of cases (for individuals who were eligible to be seen) would be addressed within five years. The service continued to receive further funding beyond this time, as the demand for its services continued to exceed its resources. Originally, the Unit began with four members of staff, but with ongoing increases of client referrals, staff numbers also increased, with twelve members of staff employed at the time of my fieldwork.

¹⁰ Established in 1995, in 2001 its name was changed to the National Breast and Ovarian Cancer Centre to recognise its ongoing work in both diseases (NBOCC 2010).
The Location of the Unit

It was not until I began my fieldwork that I became aware that clients did not enter the physical space where the Unit operated. Services were provided at four metropolitan public hospitals in the State (one in which they were based) and two State rural hospitals (every six months). Four divisions were established at the State’s Fairbank Hospital: (1) the Unit which provided the genetic counselling; (2) genetic laboratories which carried out some genetic testing; (3) a “registry” which provided follow-up services; and (4) an education officer from the State’s leading non-government cancer organisation.11

The physical location of the Unit was in the back section in a worn-down area of the hospital. Entering through Fairbank Hospital’s main entrance, the Unit was difficult to find. Reflecting the fact that no clients entered the Unit, it was not mentioned on the hospital’s large directory maps or on signs placed throughout the hospital. To reach the Unit, I had to go up some stairs, take various left and right turns, and take a lift, with soiled carpets and small graffiti tags of people’s names, up to the seventh floor. Stepping out from the lift, a small plain, stale smelling corridor area, with no signs, no windows and various wooden doors dotted up a hallway, made it obvious that it was not a usable public space. Closest to the lift, the Unit was behind a large wooden door. There was an intercom by the door, and on the door a peephole and a lock that required a code to open (see Figure 3, next page). Attached to the door, in a clear plastic sleeve, was an A4 sized poster with a picture of a DNA spiral and the Unit’s name. Such signage made the Unit’s location appear temporary and lacking in funds and value, when compared to the central location and professional signage used in the general genetics unit and throughout the hospital.

11 The four divisions of the Unit began and remained at four separate spaces within Fairbank Hospital.
Figure 3. Photograph of the entry door to the Unit.

The presence, aesthetics and location of the Unit are significant because they were indicative of its role, value and the variations in medical views and practice. Although biomedical hospitals appear to be deceptively familiar throughout society, it is increasingly being recognised that the medical views of people working within hospitals and related technical facilities often vary, reflecting and influencing differing diagnostic and medical treatments (Van Der Geest & Finkler 2004, pp. 1996, 1998). There were differing reasons provided as to why the Unit, which practised a specific type of preventative medicine particularly relating to familial cancer, was positioned in a hospital that largely provided obstetric services and health care to children. Overall the Unit’s presence in the hospital indicated the existence of an important relationship with what was largely being practised in the hospital: reproductive and paediatric medicine and human development. The clinical geneticists of the Unit, as medical professionals, worked in close partnership with other medical professionals from the hospital’s general genetics unit and laboratories. The general genetics unit and laboratories dealt with certain conditions in women and children in the hospital and those suffering from genetic conditions (unrelated or related to familial cancer). By positioning the Unit in the
hospital, the clinical geneticists were in close proximity to other medical professionals allowing the provision and receiving of medical information, and timely collaboration on cases.

The Unit was unique in its practice of predictive diagnostic-medicine, where clients received a diagnosis of familial cancer risk, in a hospital where classic diagnostic-medicine dominated. Although the presence of the Unit within Fairbank Hospital indicated that the services the Unit provided were of value, the physical aesthetics and location indicated that they were not considered as important as the other dominant diagnostic medical units.

**Important Predictions**

At the time of the Unit’s establishment, it was promoted as being a major development as there was said to be no “comparable service” in Australia. According to Unit documents, it was also expected that the service would be the forerunner of other similar services “when” (not “if”) the genes responsible for dementia, diabetes, and hypertension were identified. These predictions provide an example of how assumptions around genes and genetics extended beyond familial cancer. Staff of the Unit displayed, in various ways and contexts, confidence in the ability of genetic testing to predict illness and to modify outcomes, if not at the time, then in the future (discussed further in subsequent sections of this thesis). This confidence revealed a belief that the Unit was progressive in nature and given time, all diseases (and even certain human behaviours) would be explained and accounted for by inherited mutations in genes. The desire to be the first of its kind and progressive was not unique to the Unit. The greater the theoretical and technical expertise and advancements, the more scientific value and authority one receives (Hong 2008, p.
544). Bourdieu states that scientific authority is a particular kind of cultural and symbolic capital that can be “accumulated, transmitted, and even reconverted into other kinds of capital under certain conditions” (1975, p. 25). In the field of the new genetics, the race to be the first to find, discover, uncover and reveal has been widely documented by scientific, academic and social commentators (for more information see Chapter 8).

**Funding and “Cost-Savings” of the Unit**

Ownership of the Unit was adopted by Pathology Partners, the State’s leading not-for-profit network of laboratories. Funding to operate the Unit came from the health department, by the institutions hosting the laboratories, fee-paying referral of test samples to the laboratories from outside the State and country, and annual grants from the State’s leading cancer organisation. The provision and funding of such services was said to enable the move of genetics into the mainstream of healthcare delivery, while also addressing cancer, said to be “one of the largest causes of morbidity, mortality, and health-related costs in the community” (Unit document 2009).

The structure of the Unit, its reporting processes and outcome measures, were not specified initially in the proposal for the Unit’s establishment, however estimates of both the potential savings the Unit could generate and its demand were provided. These estimates were based on the prediction of possible future use of the Unit’s services by clients, the accumulation of clients waiting to be seen, and the developments in the technology of genetic testing. The emphasis was placed on the “cost-savings” – the financial benefits associated with the identification of both those termed “unaffected non-

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12 Funding by this organisation ceased while I was undertaking fieldwork in 2010, along with the removal of the funding for the education officer.
carriers” and “unaffected carriers”. In those individuals deemed to be an “unaffected non-carrier” of a known genetic mutation, the avoidance of unnecessary cancer surveillance in them, and subsequently their genetic relatives, was estimated (at the time) to save $13,000 in cancer surveillance per person over their lifetime.  

The savings associated with those identified as being “unaffected carriers” of mutations, learning of their status was predicted to lie in the reduced incidence or severity of cancer. This was because those identified were expected to undertake early or more effective cancer surveillance and possibly undertake more serious cancer reduction strategies. These included having surgery to remove a part of the body (for example, their breasts and/or their ovaries) that was associated with an increased risk of familial cancer.

It was predicted that the costs of the Unit would be offset by these two factors and would consequently result in a reduction in total cancer care costs. Predictions of improving health and lowering costs are part of normal practice in modern healthcare. In Chapter 8, I discuss the impact and concern of economics and the commodification of the body. It is common practice in medicine that funding by both private and public sources is dependent on a partnership where certain agendas, found in mission statements, are shown, or can be shown to be addressed. Such agendas require the pursuit of knowledge that will be useful in improving health. Latimer argued:

> they need to show how they help reveal correlations between biological processes and the development of disease or the sustaining of health, together with the direction of possible interventions. (2013a, p. 41)

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13 These estimates were acknowledged to be “at best, approximate and at times no more than guesses” (Unit document 2009).
Fieldwork Methods

Although I began in the Unit with the understanding that I would focus my research on the staff members and their clients, my day-to-day time was spent in the Unit observing the staff members without clients present. In the field, I spent between two and eight hours per day observing staff of the Unit in their office setting. I employed various ethnographic research techniques and strategies throughout my time in the field. Participant observation is the primary research method used by anthropologists and refers to a process of learning through the observation of, and involvement in, the routine activities of the researched group (Schensul, S, Schensul, J & Lecompte 1999, p. 91). The majority of the time I observed staff and only sometimes participated in the routines of the staff within the office space (such as helping to prepare food or rooms for meetings). The majority of the data I gathered around staff knowledge and practice was collected while observing staff in their work roles, and in their engagement with each other and external health professionals. To a lesser extent, their knowledge was directly explained to me or observed with clients.

Even though I was generally not able to participate in the daily routines of staff members, I employed other ethnographic research techniques and strategies to learn about what they did and how they did it. The techniques included conducting both formal and informal interviews with staff and some of their clients. I collected written and visual familial cancer related material including that produced by the Unit. I also attended some genetic counselling “clinic” appointments, attended and audio recorded meetings, and photographed and mapped various environments.
A Desk to Sit and Observe From

On my “official” first day of fieldwork in the Unit, I was greeted by Ella, an administrative officer, whom I followed down a corridor to a back room. In this back room I was shown a desk area that I was informed had been “prepared for me to sit at” (see Figure 4).

Figure 4. Photograph of my assigned seat.

I sat down at the desk that faced a brick wall. On the desk were signed consent forms (for this ethnographic study) from all of the staff, which Ella took to make photocopies of in the hallway. Turning around I could see three desks, one to which Ella returned. As a novice anthropologist, I was feeling very nervous and I got out my field diary and began writing. I heard talking in the background and the “jingling” of teaspoons in cups from the kitchen. I sat thinking about whether I should wait for someone to approach me or whether I should, or could, leave the desk and explore my surroundings.

The location of the desk at which I was placed – situated in the administration room and facing a brick wall, with the busyness of the staff behind me – turned out to be significant. Where I sat, what I could observe, and what I could not observe, highlighted
the inner workings of the Unit and appeared to reflect understandings about why I was there. I sat at my assigned desk and listened intently to conversations being held around me. I looked forward to sitting with staff members at lunchtime, and during their routine Monday clinical meeting, as well as accompanying the clinicians to a weekly held journal club held in the general genetics unit.\(^{14}\)

The reality of conducting ethnographic research in an office environment proved difficult, resulting in a number of obstacles to be faced. I only felt comfortable going into areas that I had reasons to be in. For me, these were the kitchen, the meeting room, the hallway that led outside the Unit and the external bathroom. To increase my presence, and the chance of an interaction with staff of the Unit, I made trips to the kitchen to get a glass of water, or I walked through the hall out the front door to the toilet, grabbed books from a bookshelf in the hallway, or sat at the meeting room table writing notes. Although small, these strategies allowed me to observe staff members in their roles in their offices and sometimes resulting in interactions.

To a lesser extent, my fieldsite also extended outside the walls of the Unit. Away from the Unit, I collected fieldnotes and photographs of various things. These included: meeting the genetic counsellors at other local and rural hospital locations to observe genetic counselling clinics, attending an interstate conference, and accompanying John on a rural school visit. I also attended various staff gatherings (work and non-work related) that were held in eateries, and on one occasion I attended a BBQ that took place in a staff member’s home. I largely documented my observations and experiences in short hand.

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\(^{14}\) The journal club involved clinicians from the Unit and from the general genetics unit. Involving approximately 10-20 members, the clinicians took turns presenting either new research or a case study for discussion by the group.
form in a field-diary. These notes were then written up, usually on the same day, which made occurrences and details, including my emotional reactions, easier to remember and reflect upon.

At times it was confusing to know the type, and extent, of conversations and experiences that I should have, and should not have, documented in my fieldnotes. It has been stated by Thorne (2004, p. 166) that most ethnographers assume that if they are not told to leave, if their presence is tolerated, then consent has been granted. To accommodate this experience, I attended and documented what was going on, except or until I was told not to include what had taken place. Sometimes I was directly told not to include something that was said, or I was told that it was “off the record”. Other times, it was not so straightforward and, in some circumstances, when I expected to be told not to include something, and I was not, or when I was explicitly told to “write that down”, I became confused as to whether the staff did not mind if I wrote about what had taken place, whether staff had forgotten my presence, or, in the case of being told to write something down, if I was considered as a vehicle to express the normally inexpressible.

It is a common occurrence of ethnographic field research for participants to forget that they are being observed (Fine 1993, p. 283; Thorne 2004, p. 167). Punch (1993, p. 190) argues that a feature of regular presence and the use of non-obtrusive methods by the researcher can result in them becoming “as familiar as a piece of furniture”. Although “not being noticed” had significant benefits for my data collection because it meant that participants were not consciously modifying their behaviour in the presence of the observer, concerns have been raised about the ethics of this (Watts 2010, p. 3). In consideration of this ethical dilemma, to minimise the occurrence, and to remind
the staff that I was always there to document their behaviour, I carried and openly recorded notes in brightly coloured field diaries.

All of the methods and strategies used not only resulted in the accumulation of various forms of oral, written and visual data, but at times they helped me negotiate the fieldwork challenges encountered. I argue below that the fieldwork challenges I experienced were due to two main factors that influenced each other: the first, a difficulty establishing rapport and obtaining membership with my participants due to a paradigm clash; and the second, a distinction between “procedural ethics” and “ethics in practice” (Guillemin & Gillam 2004, pp. 263–264). These challenges are crucial to describe and explore because they not only reflect the uncertainty of the fieldsite but they shaped my fieldwork experience, the data I collected and the focus of my research. They also highlight the multiple power bases and relationships that existed within the field.

Fieldwork at Home

Traditionally, anthropologists travel to remote or foreign places to study the cultures of “other” people. Historically, anthropology has been based upon “a spatial distinction between a home base and an exterior place of discovery” where travel and dwelling in a place other than home have been seen as central to the experience of fieldwork (Clifford 1997, p. 186). Caputo (2000, p. 27) states that conducting fieldwork in one’s own culture can be harder than conducting fieldwork in foreign places where differences appear more obvious. When conducting fieldwork at home, travelling to and from the field still occurs but involves a conscious and challenging journey because it is not defined by physical distance or travel (Knowles 2000, p. 55).
Before entering the field, I had felt prepared as much as possible for “reversing the lens” and analysing the beliefs and practices around what it meant to be at increased risk of familial cancer. This was primarily because these ideas were unfamiliar to me. As detailed below, the experiences of other conditions, such as the dual demands of my professional and personal life, and the ongoing difficulty establishing rapport and obtaining membership due to a paradigm clash, had not been predicted and proved difficult to negotiate.

*Dual Demands of Being Both an “Insider” and an “Outsider”*

Some of the challenges that I encountered while conducting fieldwork at home related to the dual roles and demands of my professional and personal life. Although I entered and exited the main fieldsite (the Unit) on a daily and weekly basis, I still remained partly in the field at all times because my fieldsite, and the university office I also worked from were in close proximity to each other, in the city in which I lived. My position was as both an “insider” and “outsider”. Having no medical background, particularly in the area of genetics or cancer, I was an “outsider” to my participants. However, I lived in the same city and I was still very much part of a culture that has deeply ingrained biomedical beliefs and practices, giving me some “insider” status. Fieldwork at home demands a “degree of self-consciousness” that can be extremely challenging because there are “role conflicts” that appear between the already assumed role in one’s own society and the multiple roles of the ethnographer (Caputo 2000, p. 26).

Caputo describes fieldwork at home as “a juggling act” requiring continuous shifting according to whether one is at home, at work, or in the field. It involves adding another dimension to a researcher’s established social relationships, everyday situations, routines
and commitments once fieldwork begins (Caputo 2000, p. 27). This statement reflected my experience. The same amount of work was required whilst also juggling normal home, family and social responsibilities. On a couple of occasions, personal and field appointments clashed with each other when I was given late notice to attend events that were held outside of the Unit’s “normal” operating hours. In one instance, I was invited to a staff member’s farewell dinner occurring that evening, while another time I was asked if I wanted to attend a very significant interstate conference, to be held within that fortnight. In these instances, I was able to put off personal commitments and attend the fieldwork events because I believed they were crucial to attend. These are examples of some of the dual demands I experienced because I was conducting fieldwork at home. They also demonstrate how staff held the power to decide whether, and when, to invite me to events.

There were certainly positive aspects of conducting fieldwork at home. Surroundings were familiar, I did not have to leave family and friends for long periods, the financial expenses were minimal, and I still had the availability of my supervisors to discuss fieldwork challenges.

**Field Relations – Access Not Membership**

I was provided access (with restrictions) to conduct ethnographic research in the Unit but I was not provided with, nor did I earn, full membership. There are varying degrees of membership. Anthropologists have produced various terms to describe the range in the degree of participant observation as linked with the membership roles that researchers may take. The first, considered external membership, involves observing activities from outside the research setting. With peripheral membership, the researcher is considered a
“non-participant” or “passive participant” and has a bystander role, observing in the setting but not participating in activities. Active membership roles signify the researcher’s participation in certain, or nearly all, activities. The final membership, known as full membership, sees complete participation in the setting. The degree to which the researcher participates in membership roles is said to be determined by both the researcher and the setting (Adler, P & Adler A 1987, p. 8; DeWalt, K.M, DeWalt, B.R & Wayland 1998, pp. 259–300; Spradley 1980, pp. 58, 62).

Of the membership types mentioned above, my experience could be considered as peripheral or passive participation. The negative aspect of this membership is that it limits the ability to immerse oneself in the field and establish rapport (Adler, P & Adler A 1987, pp. 36–46; Spradley 1980, pp. 58, 62). Limited immersion in my fieldsite shaped my fieldwork experience and impacted on the research project in terms of the focus, how I collected data, and the type of information I gathered.

The inability to participate when conducting fieldwork in hospitals is not uncommon. After conducting fieldwork in a hospital setting, Wind (2008, p. 81) was instrumental in questioning whether an anthropologist could ever truly participate in a hospital setting because it is not possible to be the patient, visitor, nurse or doctor. Further to this, Wind (2008, p. 85) questioned the limitations of participant observation, and ethnographers naïveté in claiming to understand their participants better because they had participated. A similar notion is shared by Kleinman (2006, pp. 161, 235), who after experiencing pain in his life, expressed remorse over previous claims of understanding the pain and suffering of patients. The underlying message is that ethnographers in hospital settings should be cautious and critical towards the achievements of participant observation.
It has also been stated that researchers who have been in the field for some time should be accepted to some degree (Schensul, S, Schensul, J & Lecompte 1999, p. 18). Unfortunately for Unit staff and myself, confusion as a result of not knowing what I should, or could do and see, or what I was there for, increased instead of decreasing. Once entry had been gained, instead of knocking down barriers and gaining a sense of more freedom and access, my experience was of limited access as my fieldwork progressed. Playing a fundamental role in shaping my research, these developments are important to consider as part of my ethnography. For the duration of my fieldwork I believe I remained at the same level of membership for two main reasons. As discussed below, there was confusion over what I could do practically (in the field) versus in theory (on paper), and there was a lack of rapport due to a paradigm clash between our methods of investigation. By not being able to question, participate in and learn some of the everyday skills and practices of the Unit, I was denied the possibility of observing certain things, but on the other hand, other areas were illuminated.

The Ambiguity of Rapport and Acceptance

The last several decades have seen an increase in writing about the ambiguity of boundaries between researchers and those researched, particularly regarding indigenous ethnographers (these include Karim 1993, p. 248; Messerschmidt 1981, p. 198). The greatest challenge I faced in the field related to the ability to interact with staff and gather certain information. I came to learn that there was no guarantee of rapport or acceptance just because I was doing fieldwork at home. “Good” rapport in the classic sense is when the anthropologist is adopted, or accepted, by the community. However, in the eyes of those being researched, anthropologists must assume a role that is both believable and non-threatening in order to achieve acceptance (Sluka 2012, p. 138).
Tied with building rapport, confidentiality is also a part of the reciprocal trust that needs to be established with participants. Participants need to be assured that they can share personal information without their identity being exposed to others (Kawulich 2005, p. 39). In theory, this was achievable in my fieldsite, through the agreed use of pseudonyms, but in practice, the staff raised concerns that they, and others whom they had close collaborations with (such as the general genetics unit, and other familial cancer services), would be able to identify the Unit and particular staff members in my writing.

As I had initially met with, informed and negotiated with all of the staff about my research interests and the ethnographic methods, I naively expected that data collection would be straightforward because I assumed that staff were aware of, and in agreement about, what I would do. However, due to a lack of rapport, I experienced what felt like subtle resistance and exclusion from staff in different forms. It has been argued that researchers should expect to experience exclusion at points during the research process and particularly in the beginning (Schensul, S Schensul, J & Lecompte 1999, p. 18). A number of factors can affect whether the researcher is accepted. These include their appearance, ethnicity, age, gender, education, class, a lack of trust, discomfort with having an outsider there and potential danger to either the community or the researcher (Plankey-Videla 2012, pp. 3, 19; Schensul, S Schensul, J & Lecompte 1999, pp. 27–28; Voloder 2008, p. 29). Such exclusion can take the form of the participants using an unfamiliar language in front of, or with, the researcher, the changing of conversation when a researcher approaches, the refusal to answer certain questions, the moving away from the researcher to talk, or by not inviting the researcher to events (Schensul, S, Schensul J & Lecompte 1999, p. 17). I experienced all of the above forms of exclusion. However, it is not possible to know to what extent, if any, each factor mentioned above
impacted on the type and amount of access I was given.

In my case, exclusion mainly came in the form of difficulties in eliciting information about what the staff were doing and why. At times, such questions of staff were met with reticence. I was successful in undertaking pre-arranged interviews, but I struggled to spend time sitting with the staff one-on-one to observe and/or talk to them about what they were doing. As I will discuss later, I had been allocated to a staff member who was to help me in the field. Such expectations were not to be realised because a lack of rapport with her resulted in cordial, but minimal interaction.

Episodes of exclusion and resistance impacted on the research process but they also highlighted a lack of rapport and confusion over methods. One of the reasons for the ongoing lack of rapport was because I was largely considered an outsider in the Unit. As I was not a member of a similar biomedical profession and my ethnographic methods were unfamiliar, I was excluded from certain knowledge and from the hierarchical group. I argue that the difficulties I had in implementing ethnographic methods emerged because the methods were very different from the scientific methodologies with which the staff were familiar. I was the researcher and they were the researched. Staff members were unsure about what I was doing and why I was studying them, which demonstrated a misunderstanding about ethnography and a lack of rapport and acceptance. The misunderstanding about ethnographic research related to and highlighted an ongoing paradigm clash.
Paradigm Clash

Evidence that there was an ongoing paradigm clash came in various forms. On a daily basis I provided justifications of my ethnographic methods because staff held different perceptions than those I tried to convey. I felt the need to explain my methods to answer both common comments made to me in regards to what I could be writing about staff of the Unit, and, at other times, one-off remarks (such as how I was “hanging around the corridor like a fly”). These repetitive questions and remarks made it clear that not only did the staff have different perceptions of what I did and what I should do, but that the explanations that I was providing to them, in the form of daily conversations and formal presentations (described later), were not able to change their perceptions. These are examples of a disjunctures that highlighted a paradigm clash.

In attempts to build relationships and gain rapport to increase my presence in the Unit, I implemented some strategies in the field with different outcomes. After noticing that there was a strong role for, and appreciation of, sweets (in particular homemade sweets), on a few occasions I made or bought cakes and cookies to share at their Monday clinical meetings where all staff attended (see Chapter 4 for discussion of these meetings). With praise and appreciation shown by staff towards my gesture, such attempts did appear to build rapport but did not resolve the paradigm clash.

Representation

The second major factor that influenced what I was allowed to observe and be informed of related to how staff believed I was going to present them as individuals, as a group and an organisation. The reason for their interest, other than being curious, reflected their need for clues as to what I was collecting and, therefore, how they would be represented.
Although it was agreed that members of staff and locations would be provided with pseudonyms and have identifying characteristics removed when publishing my research, staff still held concern over what might be written. Staff concern over what could be represented largely impacted on what was shown to me and explained when it came to information regarding their clients. This demonstrates ways in which control was attempted. As explained later, without access to the clients of the Unit in the way that was anticipated prior to undertaking fieldwork, ultimately the staff, and in particular the clinicians, became the focus.

Schwartzman (1993, p. 49) stated: “It is also important to recognize that while researchers are watching their informants, informants are also watching researchers”. This quote reflects my experience of being an object of attention as the staff watched and analysed me at the same time that I watched and analysed the staff.

*Justification*

Evidence of a paradigm clash also came through the ongoing written and verbal forms of justification for my research, which I needed to provide to staff. Although I had gained ethical approval and entry into the Unit, what I did, and did not do, including the justifications I made, affected how the staff viewed the importance of my research which impacted on what they did, and did not, show me. These barriers demonstrate that although I was able to gain entry into the fieldsite relatively easily, maintaining a presence was ongoing and complex. Foucault (1973, pp. 15, 17, 18) has been fundamental in describing how hospitals and clinics, as highly structured, protected and exclusive institutional spaces, are not easily accessible to ethnographic enquiry. It has also been noted that because such barriers arise when an anthropologist accesses a
hospital or clinic space, access cannot be taken for granted (Long, Hunter & Van Der Geest 2008, p. 71).

My inability to be more than an anthropologist observing from a desk, displayed the lack of power I had in the Unit while highlighting who had both the power and the certain types of knowledge that were favoured. Various statements and questions raised by staff displayed a concern about how they might be represented in my research. As mentioned earlier, I believe that such justifications were required because the staff did not understand ethnography. As discussed in the beginning of the chapter, the reasons for allowing me into the Unit as an anthropologist were because I represented and would contribute something different. However, what I was doing was different from what was expected, or wanted, resulting in staff determining what I was shown and what I accessed, influencing what I could report.

On two occasions I was asked to present formally to the group using PowerPoint. For both presentations I was provided with guidelines as to what the staff wanted to know. The decision to present some of my research in the way they had asked, and were used to, seemed the best way to dispel misunderstandings and encourage discussion. Both presentations resulted in different outcomes and played an important part in my fieldwork experience. The first presentation given in the third week of fieldwork was well received and opened access for me to attend an ongoing weekly journal club held with the general genetics unit. It was the reactions received from staff to my second presentation, given in the seventh month, which were illuminating and resulted in a watershed moment.
For the second presentation, due to concerns about the possible effects presenting my findings thus far could have on further data collection, I decided to highlight particular aspects of my findings (which heavily revolved around the role of food in the Unit). The expression of my agency lay in concluding the session by asking the staff to “show me more”. After the fifteen minute presentation I was asked various questions by the clinicians, relating to how the Unit differed from other similar services and what my analysis was at that point. I attempted to justify my methodology, explaining that I was not in the position to compare the Unit with other services and that I was not able to provide them with analysis while undertaking fieldwork because of the ongoing nature of data collection. It was at this point that I was informed that my presentation was “frustrating and not useful” because I had not shown them “anything”. While some staff members remained silent throughout the discussion, others explained that they were used to research that asked a question and provided an answer – which mine did not.

In the next chapter, I discuss the hierarchal structure of the staff. Relating to this hierarchy, I received both methodological and personal character critiques which were confronting at the time. Concerns also were raised in the form of suggestions. Staff tried to guide my methodology and wanted my data to show them how they compared and what made them different to other similar units. As discussed in Chapter 8, such a drive to be different (within appropriate boundaries) in an arena of similarities, reflects and propagates the biomedical/scientific competition where there is a desire to be distinctive. I was also provided with advice from staff as to how I could get more data from them. Although it is not in the scope of the thesis to describe in detail what these were, it is important to mention that the suggestions that were provided were subsequently attempted, but were not successful in improving access and rapport.
It became apparent from the clinicians’ comments and questions that the ethnographic methodology that I was using, and which had been agreed upon prior to my entry, became a source of frustration to staff because of my inability to provide immediate feedback of the kind that they were used to and that they could use in their work. These responses highlight the difference in the methods of our chosen fields. The kind of research that dominates biomedical teaching and practice is considered as empirical and measurable, first formulating a hypothesis and then testing and modifying it. Such research is said to result in measurable and objective results (Popper 1959, pp. 3, 4). I attempted to enter this “scientific” world by providing the PowerPoint presentations in a manner not typical of anthropologists; using a method of presentation the clinicians had asked for because it was a method they used regularly and with which they were familiar. When these attempts did not elicit the response intended, it did precipitate further discussion, which illuminated the disjuncture between biomedical, scientific research methods and anthropological research methods.

My participants shaped the data I collected but I too had agency. In providing the presentation, my authority and power lay in my choice to not provide information about what had been found up to that point, but instead to emphasise why I was there. My expression of power also lay in the ability to maintain my identity as an anthropologist and remain true to the methods of ethnography. Active reflexivity and thinking of informed consent as an on-going process helped me to navigate the shifting of the power dynamics.

These occurrences reminded me that not only did the staff have different perceptions of what I was trying to convey, but that the justifications that I was giving them, in the form
of formal presentations and in daily conversations, about what I was doing and why, did not appear to change their perception. The theory of practice, a social theory developed by Pierre Bourdieu is useful to consider here (1990, p. 15). The theory insists that in order to understand interactions among groups of people, including their decision-making processes, the degree to which people are guided by subjective structures (such as rules, laws, and the historical trajectory of an individual’s life experience), has to be acknowledged, while not disregarding the extent to which the structures are moulded and transformed in creative ways by human agency (Bourdieu 1990, p. 60). Although the processes that the staff and I were part of affected and influenced my project, the possibility for the actions of all involved in changing the outcomes should not be underestimated. The challenges that I faced in maintaining access in the Unit evolved while I was there and related to the ever-evolving nature of consent and staff relationships, which staff members tried to control through what they chose to allow me access to, and to what I was denied access.

“Procedural Ethics” Versus “Ethics in Practice”
Guillemin and Gillam (2004, pp. 263–264) describe two main dimensions to ethics in research, the first involves the formal process of gaining approval, termed “procedural ethics”, and the second dimension involve the “everyday ethical issues” that occur when doing research, termed “ethics in practice”. The proposed research I was given ethical approval for on paper prior to entering the field (what I will also refer to as “procedural ethics”) was different from the reality of what I was able to do once I was in the field (referred to as “ethics in practice”) (Guillemin & Gillam 2004, pp. 263–264). In practice, the research ethics regarding what I could do in the Unit, ethically and confidentially, were negotiated by the clinicians, those with the most powerful positions in the Unit.
My experiences of a disjuncture between “procedural ethics” and “ethics in practice” in the Unit revealed a number of complexities (Guillemin & Gillam 2004, pp. 263–264). These experiences shaped my research and highlighted the important ways in which the staff of the Unit (in particular the clinicians) had the power and knowledge to determine my ability to know and not know certain things.

I argue below that the disjuncture between the “procedural ethics” and “ethics in practice”, was influenced by two main factors (Guillemin & Gillam 2004, pp. 263–264). The first (which both informed and was influenced by the second) was due to a lack of rapport relating to the paradigm clash (as discussed in the previous section). The second related to ethical confusion around informed consent. The distinction between “procedural ethics” and “ethics in practice”, in the Unit brought with it many challenges and, as argued later, influenced the degree to which my participants and I were able to establish and build rapport (Guillemin & Gillam 2004, pp. 263–264). Below I describe the ethical process I undertook prior to entry followed by analysis of its impact once in the field. The ongoing consent issues that were experienced once in the field were absent in the initial research proposals and consent forms because (prior to undertaking fieldwork) they were not possible to predict and factor in.

**The Negotiation of Field Access**

In order to enter and have a presence as an anthropologist in the Unit, I needed to earn the right to have access to know what was going on, through the ongoing negotiation of a variety of processes. Initially I considered the process of seeking permission and ethical approval for research as something occurring prior to and separate from my fieldwork. Instead, it has been stated by Caplan (2003, p. 27) that ethical consideration occurs
through anthropological epistemology, fieldwork and writing. Caplan’s statements were indicative of my experience – as a continual process, ethical consideration and approval was a rich source of data, valuable in its own way and revealing aspects about my local fieldsite and that of the biomedical culture.

The work of anthropologist George Marcus also influenced what I considered as my fieldsite and what I included in my fieldwork notes about my fieldwork experience. In particular, Marcus stated that “multi-sited ethnography” is integral because fieldsites do not exist on their own and, therefore, should not be studied in isolation. Multi-sited ethnography does not necessarily mean including a number of fieldsites, but rather, considering how larger forces impact on local fieldsites and give a voice to all of those involved in a fieldsite (1998, p. 3). In consideration of this, the purpose of this section is twofold: to discuss the initial events, “the larger forces”, which impacted on how I conducted my research: and secondly, to demonstrate how these events reflected particular relationships and power between institutions, hospitals, governments, participants and myself. I argue that the power to allow, sign off and sometimes negotiate what I was able to do was held by particular members from the university, hospital research committees and those with leadership positions within the broader Pathology Partners organisation.

“Procedural Ethics” – Pre Fieldwork

Before I could begin fieldwork in the Unit, I was required to undertake a number of processes in order to provide justification of my study’s worth and obtain ethical approval for my research. Initially the “larger forces” that impacted on how I conducted research in my fieldsite related to the processes that came under the larger bureaucratic banner
known as “ethics”. Where research involves human or animal participants there is the expectation that all researchers submit proposals for consideration to ethics committees. In Australia, research must comply with the National Statement on Ethical Conduct in Human Research which comprised a series of guidelines (NHMRC 2007). In my case, the “procedural ethics” processes involved the University (in which I was enrolled), the State Government, Fairbank Hospital, the Unit, Pathology Partners (the organisation that owned the Unit), the participants and myself (Guillemin & Gillam 2004, pp. 263–264). It involved justifying, consulting and negotiating with these entities, both directly and indirectly to obtain either funding or ethical approval for my research.

All researchers need to consider ethics because all research is affected by ethics and the process of gaining ethical approval and answering to a research ethics committee is vital because it helps reduce harm and protects the anonymity and confidentiality of research participants (Chenhall, Senior & Belton 2011, p. 13; Guillemin & Gillam 2004, p. 277). The procedural requirement that all research be subject to review by ethics committees, has been set up for the protection of participants. The Nuremberg Code, consisting of ten principles, was the first international guidelines made on medical research in 1946 and was in direct reaction to the medical atrocities committed by some doctors during the Nazi era (Hope 2004, p. 99). These guidelines developed into the Declaration of Helsinki in 1964, an apolitical association made up of delegates throughout the world who discuss and recommend guidelines related to medical ethics, medical education and socio-medical affairs (Hope 2004, p. 100). Ethical approval to conduct research is strictly controlled and regulated because of this history and it has been related to a rise in the preoccupation of accountability, transparency, audit and funding in research (Parker 2007, p. 2252; Strathern 2000, p. 294).
Initially written justification to the State’s health department was required to obtain funding for the project. Details of the aims, objectives, background research, methodology, and a timeline were all required. I was also required to provide a statement about how my proposed research contributed towards meeting three of the State’s strategic health plan objectives of improving wellbeing (through preventive health), healthy life expectancy and psychological wellbeing. My predicted contribution for meeting the three objectives were by: understanding the role of genetic counselling on population health and individual health; maximising the effectiveness of existing services to increase life expectancy and prognosis after the identification of risk of potential chronic diseases; and exploring both the positive and negative impacts (on the individual and family) of learning that one is (or is not) a carrier of a genetic predisposition for cancer.

The required prediction of my contribution demonstrates how I was made part of an alliance that medicine has with government, through its relationship of health and wealth, an argument shared by Latimer (2013a, p. 40). This relationship sees private and public funding dependent on the partnership where agendas are, or can be shown to be addressed. Such agendas require the pursuit of knowledge that will be useful in improving health outcomes (Latimer 2013a, p. 41). As mentioned earlier, the establishment and funding of the Unit was dependent on the potential cost savings. Likewise, the justification of my contribution to the specific objectives set out by the State in exchange for a scholarship and approval to conduct my research, was an extension of this common practice and alliance.
Anticipatory Research and the Negotiation of Research

Neyland (2008, p. 62) stated, “It is in negotiating access that a great deal about the organization (and the ethnographer’s engagement with it) comes to light”. Once I was awarded the scholarship from the State’s health department, meetings and emails with my future hosts took place, in particular with John, in order to discuss and consult on details about the project. During the first meeting, John assigned one of the genetic counsellors (who had been there since the Unit had formed) to be my fieldsite contact. In anthropology, it is considered important that the ethnographer has an inside contact or “gatekeeper”, who occupies a position that allows them to control access to key resources either directly or indirectly (Neyland 2008, p. 83). It is important because there are seen to be two types of gatekeepers; whereas the right one assists the research, the wrong gatekeeper can hinder the research (Gallinaro 2009). I naively assumed that being assigned an inside contact would ease my fieldsite entry and access to resources but as discussed, I had difficulty accessing resources.

When I first met with my hosts numerous things were discussed including: the types of events that I could attend, how clients would be recruited, and how staff and clients’ confidentiality could be maintained (including in circumstances where people who had not consented were present). The verbal agreements made on that day regarding the maintenance of confidentiality, my access and methods of recruitment were then recorded in a research proposal for the University along with two ethics applications, one for the hospital and one for the University. Each proposal required typical justification of research aims, purpose and background, safety and ecological considerations, ethical considerations, confidentiality and data security. Other aspects of the research were also controlled by Pathology Partners, the organization which owned the Unit. They required
proof of my student enrolment, research indemnification insurance, a criminal history check, and a signed confidentiality agreement for the hospital, the department of genetics, and Pathology Partners.

As discussed, once I was in the field, a shift in the focus and direction of my research subsequently followed, due to the impact of power and knowledge held by my hosts. In the proposals written prior to entering the site, my intention was to provide an ethnographic account of the Unit and its staff, visitors and clients (and their family members where possible). At that time, the main focus was proposed to better understand the impact of familial counselling on kinship networks and emotions. It had initially been agreed, prior to recruitment, that the aim was to follow a diversity of clients visiting the Unit (to capture the many realities of familial cancer). As stated above, specific in depth details regarding who and how participants would be initially recruited were documented, along with details regarding how their ongoing experiences were to be captured through speaking with clients both prior to genetic counselling and post genetic counselling. Once I began fieldwork, most of the verbal and written agreements made prior to my entry in regards to what and how I could do things changed. No written documentation prepared in advance could have covered the ethical and consent issues that I experienced because factors I encountered were not possible to predict.

As stated above, prior to submitting the first proposal to the hospital’s Human Research Ethics Committee (HREC), John vetted the document, providing details of exactly how I was to recruit clients to interview (due to his authority, and knowledge and experience of clients in the Unit). The process of consulting and negotiating with the Unit – to reach an agreement as to how I was to conduct my research – was problematic because it was
impossible to establish, prior to completing the research, what could be negotiated. Parker (2007, p. 2253) has stated that the concept of “negotiation” as a solution to the need for anticipatory research and ethics proposals for anthropologists is problematic for a few reasons. One of these reasons is because “it is not easy to see how the ethnographer (or the member of the community hosting the research) can be confident about just what it is that is under negotiation”. I faced difficulties in negotiating access while I was in the fieldsite and, likewise, the staff struggled to understand what I was meant to be doing and how I was to do it. This reflected conceptual problems with viewing my research proposals as a framework for my research.

Hoeyer, Dahlager and Lynöe (2005) consider the central ethical issues in the two different research ethics traditions of medical research ethics and research ethics in anthropology (and its related disciplines), as the cause of “clashes” between researchers and medical staff. These issues are said to occur because medical research ethics is inconsistent with participant observation and the anthropological understanding of the research process. The need to obtain “informed consent” from research participants prior to entering the fieldsite is an example of this problematic clash in research paradigms (Parker 2007, p. 2252). Aagaard-Hansen and Johansen (2008, pp. 15–19) describe that a different set of ethical issues arise for qualitative researches in medical research. For Chenhall, Senior and Belton (2011, p. 13) the differences are often not “adequately recognized or catered for by research ethics committees”. As discussed later, the need for anticipatory informed consent, like the anticipatory research and ethics proposal, had ongoing implications for my fieldwork.
Although I very much anticipated and considered the questions about my research that were asked in the ethics proposals, the ethics process did not create a space for ethnographic methods. Simpson (2011, p. 378) has stated that because ethnography does not fit the “human subject model”, unintended consequences of ethical review occurs, particularly when it comes to research that is linked to health and medicine. Such consequences, for me, related to the written justifications needed in order pass through both the university and hospital ethics committees, and to the constricting effects related to a predefined bias towards what appropriate data were and how data should be collected and validated.

As part of any application specific information was required about who participants were, how they were to be recruited, selected and excluded, how many participants, the participants’ age range, and what exact questions I was going to ask, what I would find, and what important contributions my research would have. With limited space allowed for explanation of methodology, the references that I made to participant observation, photographs and interviews (both formal and informal), rendered questions wanting specifics about “where”, “how long”, “with whom” and “what for”. The re-review of my application and the responses indicated that the ethics committee members were accustomed to quantitative research and certain types of qualitative research, and so it was a challenge to obtain consent for ethnographic methods that occur through induction, trial and error, problem solving and chance (Silverman 2013, p. 49).

Practising Ethics in the Field

Pope states that negotiating access and consent is always a process (2005, p. 1181). My staff participants signed written consent forms providing informed consent but it
became clear they did not actually understand it – highlighting issues surrounding “informed” consent. Ethical consideration and issues are considered as magnified in medical environments, and particularly so with the issue of informed consent (Pope 2005, p. 1182). The implications of ethical requirements both on what data I collected in the Unit, and how, were directly related to the idea of informed consent, an idea that has become a central part of all research ethics (O’Neill 2003, p. 4).

Drawing on the principles of autonomy, beneficence and justice, informed consent is required to consider disclosure, decision-making capacity and voluntariness of the participant (Plankey-Videla 2012, p. 4; Rosenthal 2011, p. 1). Informed consent can be justified in two main ways. One model justifies it on the grounds that it is a key part of valuing individual autonomy, while the other model justifies it on the grounds of harm avoidance (Boddington 2012, p. 83; Pelias 2006, p. 75). Regardless of the model, such perspectives pose significant challenges for the practice of informed consent, not just when individuals are concerned but groups and relationships. Such challenges relate to who holds the power in the relationship and the inability to anticipate details of the encounters at the outset; informed consent given at one point cannot reflect the changing “kaleidoscope of relations that characterizes social reality” (Simpson 2011, pp. 387, 388).15

The Unit was not only a formal medical institution but it was also part of Pathology Partners, an organization with its own rules which governed membership and actions within. Although literature on the issues of ethics in ethnography and medical

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15 Simpson suggests that engaging with those working on ethics committees and educating them as to what an ethics of the “social subject” might entail (2011, pp. 387–388).
establishments exists, literature about informed consent in organizational ethnographies is sparse, with discussions about ethics in terms of the processes involved in obtaining informed consent from participants for access to an organization as a research site by anthropologists (Plankey-Videla 2012, p. 1). As my main fieldsite was both a medical establishment and an organisation (with commercial interests), it shaped my experiences because both required different types of informed consent for different reasons.

Below, I provide examples of some of the unanticipated occurrences I experienced, arguing that my research became increasingly shaped and affected by confusion surrounding informed consent. These difficulties related to confusion between staff members as to how I could be integrated into the workplace, as well as confusion regarding which staff members had the power to give consent for my attendance, and in which circumstances. They are examples of how power lay outside of my role as a researcher and impacted on what I could observe and know. It is commonplace to experience difficulties when conducting participant observation in organisations and medical establishments; the specific difficulties experienced are discussed with the purpose of analysing their meaning.

**Informed Consent in Practice**

Although the protection of human participants is always a concern, researchers have questioned the idea of informed consent and how it impacts on what can be realised in practice, in particular when it comes to participant observation (Gerrish 1997, p. 27; Moore 2002, p. 58; Murphy & Dingwall 2001, pp. 339–351; Punch 1994, pp. 90, 91). Informed consent in practice, namely, what was considered acceptable and not acceptable within the group, was challenging both for me as an anthropologist and for the staff.
As an anthropologist obtaining and maintaining informed consent was complicated when securing permission to do research through John, the organization’s main gatekeeper, for a number of reasons. Obtaining signatures from participants in this way, through a gatekeeper, is considered to be problematic by some researchers, mainly due to the belief that it changes the relationship dynamics, for all involved, affects the quality of data and can make people unnecessarily apprehensive about what they may be agreeing to. The process of obtaining informed consent of groups though the guidance of an individual is said to obscure the context in which research takes place (on the group) and it affects the ability of all staff involved to provide consent (Plankey-Videla 2012, p. 4; Simpson 2011, pp. 387–388).

Plankey-Videla, who conducted ethnographic research in a Mexican garment firm, problematized the practice of obtaining informed consent in organizations, and outlined ethical dilemmas commonly encountered. Those included: to whom the researcher is accountable when the gatekeeper or management provides permission to do research, how the access through the gatekeeper shapes the ability of the others in the group to interact with each other and the researcher, and what informed consent means given shifting power configurations that occur in fieldwork (Plankey-Videla 2012, p. 2). These unanticipated issues described above were also evident in my field. In certain situations it was clear who and which groups had more power in the relationship but unclear as to the extent to which each staff member’s individual consent, which made up the group, was legitimate (or if it was given because of a need, or sense to oblige with authority). The challenges I faced in the field were influenced by the confusion surrounding who decided whether, and what, was appropriate for me to attend, and also how I was to recruit participants. I have provided three examples of how
confusion surrounding informed consent impacted on my research in practice regarding
the discussion of clients, the recruitment of clients and the permission and power to
consent.

The Discussion of Clients

The first example which demonstrated a difference between what I was procedurally able
to do (ethically) and what I was socially expected to do, related to discussions held about
the Unit’s clients. In Chapter 3, I discuss how it was common practice for the clinicians to
share information, via formal and informal debriefs and gossip, about their clients with
each other. Such information included medical information (such as why their client had
been referred and their risk), and it also extended to other remarks regarding their client’s
personality, dress sense or the condition of their homes. In the circumstances where it was
my cue to respond socially and engage in such discussions, because I had spoken with the
client being discussed and there was interest in what I knew, I was not able to engage in
such conversations because of the importance of respecting the written agreement of all
my participants (in this case, the clients). This demonstrated the reality and difficulty of
maintaining confidentiality for those who had consented, in a space where such
discussions, which occurred daily, were not only taken for granted but were an important
part of group dynamics. This practice impeded rapport by reminding staff, and me, that I
was not a group member and so I was denied of relationship constructed through talk and
gossip.

The Recruitment of Clients

The main methods to recruit clients as participants, which were decided upon prior to
fieldwork and involved the genetic counsellors selecting and sending out a letter to
clients, resulted in a lack of response. In keeping with ethical protocol, the only subsequent method for the recruitment of clients – for clinical staff to ask their clients to be participants in my research prior or during their clinic appointment – was considered inappropriate by some staff. With the decision made not to compromise on the ethical approval I had obtained for my research or on the decisions made by some staff, the focus of my research, therefore, shifted to the staff of the Unit and their work. This demonstrated how the ethical procedures were not able to be put into practice in the field and highlighted the power that the clinical staff had in negotiating and determining my access to their clients.

Permission and Power to Consent

There were instances where it was challenging for me to distinguish if I needed to ask permission and to seek verbal consent, in order to attend certain events and/or take notes, and, if so, who was to grant such permission. Below I provide examples to demonstrate how some staff had the power to decide when, how and by whom I was informed, but that they too were confused at times.

Although it was agreed prior to my entry, as part of my proposal, that I would attend information sessions held for some of their clients, once I was in the field there was not agreement between clinicians as to whether this was acceptable. One of the concerns related to the clients not being aware of my presence and so not having consented prior to the event. With no personal information being discussed at the information sessions and the agreement that I be introduced prior to the session, my attendance was ultimately accepted. This example demonstrated how there was a distinction between what was ethical in theory (indicated by what was included in the ethics application) and what was
ethical in practice. In practice, what was ethical in ethnographic research was sometimes negotiated, contextual and fluid. This occurrence also demonstrated the power to decide if I was able to ethically attend the sessions depended on which staff members were present at the time and how the staff’s concern over the informed consent of their clients was sometimes negotiated (within the realm of adhering to ethical protocol).

There was confusion amongst staff about who had the authority and ability to tell me what I could, and could not do. In these cases, it was not possible to predict each situation, and so decisions regarding informed consent were made on a case-by-case basis. Whether it was documenting fieldnotes, attending a meeting, a social gathering, or looking at a consenting client’s folder, the decision regarding what was acceptable for me to do differed depending on who I was with and the context. When I was given permission to attend or observe something, I took the opportunity and went along with whoever was providing the permission at the time (only in circumstances where I thought it was ethical to do so). This decision was not always without ramifications; sometimes frustration and concern was expressed about my presence and on a few occasions I found out after an event that my attendance or observation was problematic because not all of the medical professionals present knew of, or had consented to, my role. These examples illustrate how it was not apparent to me whose responsibility it was to determine what I could and could not do, and it was also not apparent to staff.

Conclusions
This chapter has detailed the various ways in which my ambiguous position in the Unit as a visiting anthropologist shaped my experiences of the Unit and my research. I argue that between distinguishing between “procedural ethics” and “ethics in practice” is crucial in
understanding the unfolding research process and the data it yielded. The following chapter provides an overview of the teams that made up the Unit, followed by descriptions of the working environment and individual staff roles in order to provide a grounding for the chapters that follow.
Chapter 3. Staff Roles and the Working Environment

At the time of my fieldwork there were twelve members of staff in the Unit, the majority of whom worked part-time hours by choice. These included: two clinical geneticists, five genetic counsellors, two research nurses and three administrative staff. By the end of my fieldwork the positions of one genetic counsellor and one research nurse had ended and their workloads and duties shifted to the other relevant staff members, thus increasing their workloads. All of the workloads of staff increased each year as the demand for the Unit’s services increased, with approximately 900 clients attending annually in the year of fieldwork. Along with the increase in client numbers came an increase in the complexity of client cases – cases that required more time be spent because of the uncertainties that surrounded diagnosis and recommendations.

The twelve members of staff worked both individually and as members of small groups to take in, define and look after client referrals. The power and ability that each individual had to influence and make decisions within the Unit, was determined by their position, duties, qualifications, experience, and years worked in the Unit. The clinical geneticists had the greatest amount of certified medical knowledge and training (both holding a medical degree), and were at the top of the hierarchy. This resulted in them having the greatest amount of power to make decisions. The genetic counsellors held degrees in nursing, social work or health sciences and had completed postgraduate training in genetic counselling. The genetic counsellors required specific training in genetics and familial cancer and so they had more authority than the research nurses who had nursing
degrees. Those working in administrative positions were at the bottom of the hierarchy because the administrative staff did not have medical education or training.

Such divisions of labour were not unique to the Unit, but correspond to the distinctive area and epistemology of biomedicine, and are the product of an ongoing politics of legitimization (Hahn & Kleinman 1983, pp. 314, 315). As well as reflecting common practices in biomedicine, the division of each group on the hierarchal scale depended on, and reflected, the amount of certified education the individuals within each group had around familial cancer. Increased education equalled increased knowledge and the power to make decisions in relation to the management of clients and the service. Greater financial salaries and other symbolic rewards (increased status) were also directly related to the hierarchy, a practice that generally occurs in biomedicine and elsewhere (Hahn & Kleinman 1983, pp. 314, 315).

This chapter describes the working environment of the Unit providing a snapshot of the staff and work roles. Divided into three main sections, the first section introduces the work environment and roles of staff while emphasising a clinical approach. In the Unit, the emphasis by clinicians (the genetic counsellors and the clinical geneticists) in providing their clients with education, rather than the provision of psychological care, was reflected in clinic appointments (see Chapter 6), in the rooms of the Unit, and demonstrated by staff in their roles.

Throughout the thesis there is emphasis placed on the work of the clinicians, which reflects their group hierarchy due to the clinical work that only they performed with clients. In lieu of this, their roles are only briefly summarized in this chapter when
compared to the research nurses and the staff in administrative roles, who do not feature as widely throughout. Rather than just being descriptive, the purpose of this chapter is to reveal how their duties were much more than mundane administrative practices. In the formation of “research families”, to the creation of pedigrees and data reports, these duties were essential to the daily running of the Unit and contributed to the clinical work undertaken by the clinicians.

The second main section of the chapter describes how the satisfaction and socialisation of staff members in particular ways impacted on how the in-house preparation for clinical work was undertaken. Particular attention is paid to the role of food, humour and gossip. The remainder of the chapter explicates the meeting room, a significant space where both the socialisation of staff and the very important Monday clinical meetings occurred, where individual job roles, teamwork and hierarchy were displayed and reproduced.

The Working Environment of the Unit
The front wooden door was the only way to enter and exit the Unit. Upon entering the door there was a hallway with rooms on either side until the hall ended at a kitchen. Office furniture, equipment and noises common in Australian 21st century office settings made up the Unit. A bookcase in the hallway prominently displayed various medical and genetic reference books and journals (see Figure 5, next page).
These resources consisted of clinical texts written by medical doctors and clinical geneticists about familial cancer, or other familial conditions. As clinical texts the resources contained information about familial conditions, genetic testing, risk of occurrence and management. In most cases the reader required a high level of medical education and knowledge to be able to understand the terminology used. It was common to open a page and see a table, graph or a photograph. The photographs included genes under a microscope, the deformed face of a person as a result of a familial condition, and a person’s chest after a double mastectomy.16

With knowledge, practices and recommendations regarding genetics rapidly changing, it was important that the resources on the bookshelf were readily accessible and kept up to date. Even with access to the Internet and online familial databases, the clinical geneticists often grabbed an item from the shelf to consult. The genetic counsellors did not use the resources from that shelf. Although as clinicians they too were required to

16 A double mastectomy refers to the surgical removal of both breasts.
keep up to date with practices, they received selected resources via email from the clinical geneticists. It was viewed that the clinical geneticists had a much more challenging role in keeping up with the clinical literature because they were ultimately responsible for the clinical decisions.

Resources involving the psychological or social care of individuals and families affected by familial cancer existed, but these were displayed in one of the genetic counsellor’s offices. These resources differed greatly in scale, type and content. On a much smaller scale, their range was made up of multiple issues of “The Journal for Genetic Counselling” (the only journal dedicated to the subject), a few autobiographic accounts of familial breast cancer (in book and DVD form), and half a dozen reference books. Unlike those in the hall, these books did not have tables, graphs or photographs but largely focussed on helping the client to comprehend the medical facts to make appropriate decisions related to their individual, and familial cancer risk. In Chapter 6, I argue that there was a distinction between clinical work involving information provision and psychological therapy in the genetic counselling “clinic” appointments, with an emphasis placed on the former. A number of academics have commented that genetic counselling uses a teaching model, over a counselling model which involves informing the client of the relevant facts and enabling the client to decide (Biesecker 2003, p. 213; Kessler 1998, p. 263; Rapp 1999, p. 57). A teaching model was originally developed from the medical model where the transmission of information is highly valued and results in less attention paid in genetic counselling to the psychological and social needs of clients. The difference in scale and content of the bookshelf resources demonstrated one way in which there was an emphasis on the provision of familial cancer medical information rather than psychological counselling.
Staff Roles

Administrative Staff

The room in which each staff member sat depended upon their role, position and room available. Ella, Anne and Dean worked in administrative roles in the largest room at the back of the Unit. Although at the back of the Unit, the room was the visual and auditory centre, or heart of the Unit, because it was the room where the most consistent work activity occurred. With no door it was also the only room in the Unit that remained open and accessible at all times.

As introduced in Chapter 2, I was placed at a desk situated with the administrative team. Where I sat reflected how my role was viewed. I was not only an outsider, but I was also an anthropologist. As an anthropologist with no clinical medical background, I was seen to fit best with the administration staff, who had the least certified medical knowledge in the building. Sitting in this room it was common to hear the sound of the printer, typing on keyboards, clicking of computer mouses, the telephone ringing and conversations held in the kitchen and along the hallway.

Being the heart of the Unit, at least one of the three administrative staff members was always present or nearby because it was important that they were available to answer incoming calls. As the first point of call, they had a purely verbal relationship with clients, vetting calls and taking messages. Although they had minimal interactions with clients (occurring over the phone), when compared to their work colleagues, they were more directly involved in building the clients’ physical and electronic files and entering the clients’ individual and familial information. The ongoing presence of Ella, Anne and Dean in the Unit was different from that of the clinicians and, to a lesser extent, the
research nurses who flowed in and out of the Unit and hospital, seeing clients and attending meetings.

With twenty-five years of experience in other medical administrative roles, Ella’s work in the Unit over the two years prior to my fieldwork was described as the most interesting yet challenging work she had undertaken. Employed as the secretary, her tasks included creating client card files, scheduling client appointments, printing of information sheets, mail duties, billing of clients, making coffee, typing out dictated letters to clients and the use of a tailor made electronic database “FamilyTrace” (discussed below).

Dean’s job was that of “registry manager” and he was responsible for the effective running of “FamilyTrace” and the organization of reports (explained further in). FamilyTrace was a familial management database developed by the Unit and initially financed by Fairbank Hospital to help address the “unique requirements” around the information management of clients and their relatives. FamilyTrace was considered as the “repository” of information, allowing simultaneous sharing of information about clients between the laboratory and the general genetics unit involved in their clients’ care. The database was also the source of income for the Unit and Fairbank Hospital who received royalties on all sales of the software locally, interstate and internationally. The capability of FamilyTrace in efficiently managing all aspects related to client care was greatly admired by staff and integral to the Unit’s daily operation.

Dean used FamilyTrace and other computer programs to manage and run “clinical data investigations”. This involved the production of daily, weekly, monthly and bimonthly reports, used in various ways. One report reminded the clinicians of any outstanding
actions involving their clients for the month (e.g. a client’s risk scores that had not been entered). Another report collected a client’s mutation test results from the laboratories, which Dean sent to the clinical geneticists for confirmation before sending to the genetic counsellors to inform the client (see Chapter 6). Dean also used the system to organise the list of high risk clients and their doctors to be sent out medical “surveillance” letters, and the list of people to be risk notified.

All of the other members of staff also used FamilyTrace on a daily basis but to varying degrees. They used the database to enter, display and report on their client’s information, familial histories, demographics and contacts. For every electronic file, a hard copy card file existed and was visible in the meeting room (see Figure 9, page 111). Despite having card files, staff were very dependent on the FamilyTrace program. The major differences between the online file in FamilyTrace, and the physical file held in the meeting room was that the online file was able to run reports on the data and alert staff to which clients had to be contacted and letters needed to be written. The system enabled a more efficient and accurate approach to the management of thousands of clients.

Anne was employed as a data officer and had two main roles. Every month the clinicians and the research nurses provided Anne with the names of clients that had died. Anne then collected the death certificates from the State’s birth’s, deaths and marriages office and the details were added to FamilyTrace, noting whether the death was cancer related or not. The cause of death was important to know because it impacted on the calculation of familial cancer risks for other biogenetic relations and because it provided medical
authentication. In Chapter 4, I explain that in the process of becoming a client, each client was required to have filled in a “family history form” which was used to make a “pedigree”. Anne was responsible for entering the family history information that the client provided into FamilyTrace which used an in-built system to create a tree like structure known as a pedigree. This pedigree was then used by the clinicians to inform their clients of their risks (see Chapter 4 for detailed discussion).

**The Research Nurses**

The two research nurses, Jenny and Samantha, sat together in the first room reached when entering the Unit, along with one of the genetic counsellors. Jenny and Samantha recruited clients for research projects established and worked on by research collectives interstate and internationally. Their work contributed to research collections where researchers throughout the world shared data and biospecimens (blood, tissue, cells, DNA, RNA and tumours). At the time of fieldwork: Jenny and Samantha worked on two major worldwide research projects, one study involved familial colorectal cancer, and the other familial breast cancer. Their roles as research nurses involved recruiting participants for the studies, with interactions with potential participants mainly occurring over the phone, although some face-to-face interactions did occur. Generally the participants either already had a mutation identified, or were from families considered at high risk of certain types of familial cancer.

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17 These records were allowed to be collected because in the probate law of the State an (unwritten) Act (obtaining consent by proxy) deceased persons lose their ability to consent, or not, to its release.

18 The researchers included epidemiologists, medical oncologists, surgeons, radiation oncologists, molecular biologists, clinical geneticists, genetic counsellors, statisticians, psychologists and pathologists. The data were paid for by researchers who accessed them, and all researchers were required to conduct peer-reviewed and ethically approved funded research on familial cancers.

19 John was a collaborator on both projects.
In Chapter 6, I explain the process involved in becoming a client of the Unit, how clients were required to be referred to the Unit by medical practitioners. Before Jenny and Samantha were able to recruit participants to each specific project, they also required a referral so that they had ethical consent, justification for contacting an individual (via telephone or mail) and providing them with more details. There were in-house referrals and participant referrals. The in-house referrals came from the genetic counsellors and the clinical geneticists who referred certain clients of the Unit considered appropriate for the research projects. Once a participant was already involved in a study, a referral from their biogenetic relations was sought. This form of recruitment was somewhat similar to the practice of risk notification practiced by the clinicians, where their clients were asked to provide information about certain relatives so that they could be notified of their risk (see Chapter 7 for detailed discussion about risk notification).

Comprehensive questionnaires were administered in the research projects that spanned three periods of life (depending on the participant’s age) and were used to collect genetic, epidemiological, medical and psychosocial data from families.\textsuperscript{20} The research nurses also collected biospecimens (genetic material) that had either been sent to them in special packages in the mail, or that they collected from hospitals after their participants had surgery. The survey data they collected were entered into FamilyTrace, stored in a de-identified central database and the biospecimens were sent interstate. Interstate and overseas researchers contributed to both research collections which had been operating for a number of years and had accumulated data on thousands of “multigenerational

\textsuperscript{20} These questionnaires required that each participant go into minute detail about what their environment, dietary, exercise, and health was during certain stages of life. The questionnaire was set up to allow for a large amount of data to be collected. The questionnaires were generally set up to only require yes or no answers by the participants.
The research nurses had both previously worked in hospital wards and were previously used to responding to ill patients’ calls, or need, for medical attention. In their past roles, the treatment they provided to the ill patient provided immediately evident results from their work and care. In their current roles, it was seldom that they provided the research results back to the participant, or saw the influence the results had on their participants. This reflected the reality of the research collaborations to which they were contributing, where any results took a number of years to produce and publish before the affected participants could be informed. The research nurses negotiated this particular dilemma by expressing the value of their work, which they were confident would eventually benefit not only some of the individuals and families who were participants of theirs, but other researchers who used the research collections to which they contributed data. This demonstrated a belief and committed to scientific endeavour.

The research nurses dealt directly with participants, most of whom were also clients of the Unit, but the type and extent of their interaction was different than the clinician’s involvement with their clients. At times, this was a point of contention because, although Jenny and Samantha were health professionals, it was not appropriate for them provide advice, or to counsel the participants they recruited for their studies. As part of the questionnaires completed, their participants were required to go into minute detail about

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21 Multigenerational kindred are a group of related persons across several generations.
their own, and their families, living conditions, health history and history of cancer. The retelling of this history brought up stories, and emotions surrounding the impact that cancer had, and continued to have, on their lives. When the participants of their research studies asked Jenny and Samantha about their risk, mutations and options, although Jenny and Samantha felt they knew the answers, and had the experience to “inform” the participants, they were not allowed. In these cases, Jenny and Samantha directed the participant back to the questions and then referred them to the genetic counsellors.

The research nurses often referred to their participants as “one of my families”, demonstrating a responsibility and relationship with the participants. Since the participants were regarded as their “families”, and therefore their responsibility, not having the certification to answer questions and/or provide information, was at times a point of tension. It also had implications for the research nurses job roles, because once they had recruited a participant they were required to encourage the participant to identify between two and eight biogenetic relations that they could further recruit for the study. I was informed that sixty percent did not want to refer because they did not want particular family members knowing about the cancers and/or mutation identification. As it was not part of their role to ask why, and to try to work through such decisions with their participants, it impacted on the number recruited. In these cases, as before, Jenny and Samantha informed their participants that there were genetic counsellors they could speak to if they wanted to work through their decision.

There were some similarities between the research nurses and the clinicians’ work. The work of the research nurses contributed to interstate and overseas research projects and, much like the clinicians, they too were required to negotiate issues surrounding
ownership of information and the service’s guidelines around confidentiality, autonomy
and family information (see Chapter 6 for examples). Yet, the research nurses had no role
in shaping the direction of the research projects.

_The Genetic Counsellors_

Due to space limitations the five genetic counsellors sat in three separate rooms that were
either adjoined or adjacent to each other. The genetic counsellors’ and the clinical
geneticists’ job roles overlapped in many respects, with both involved in the provision of
information, risk and mutation status, and risk reduction and surveillance options to their
clients. The genetic counsellors and research nurses did not provide or receive medical
information directly to or from other medical staff – the clinical geneticists relayed the
information. This reflected a hierarchy based on occupation, role, knowledge and power.

Although both the genetic counsellors and the clinical geneticists were considered as the
clinicians of the Unit because they both saw and managed clients, the genetic counsellors
had the greatest amount of interaction with the clients. These interactions occurred
primarily in person, during clinic appointments, or when clients considered at high-risk of
familial cancer were invited to attend information sessions. Further correspondence
occurred in the provision of letters that were written summaries of the clinic appointment,
and over the phone when they or the clients had questions or information to be clarified.

Like the research nurses, the genetic counsellors also often referred to their clients as
“one of mine” or “my family”. This demonstrated an ownership, responsibility and close
relationship with clients. However, inconsistent to this, the relationship with their clients
was said to be purely a working relationship. There was a marked division and a rule that
all the genetic counsellors adhered to, of not getting emotionally involved with their clients. As explained later, the clinicians had ongoing working relationships with clients and families. Sometimes the individual genetic counsellors were invited to attend their client’s family celebrations or the funerals of clients, however there was a collective decision of not attending non-work related gatherings. Adhering to this rule was said to aid the genetic counsellors in remaining un-attached emotionally to their clients while instilling the working relationship between the genetic counsellors and their client.

The photographs on the next page are of a genetic counsellor’s pin board, which also reflects the contents of the other genetic counsellor’s pin boards. The material posted on the board is another example of the dominating nature of the clinical work practiced. The board was covered with genetic based diagrams, education and resources (see Figure 6, next page). One poster illustrated, with diagrams, the “molecular mechanisms of stem-cell identity and fate”. The only references representing any psychosocial aspects of familial cancer were a business card of a psychologist and a list of support organizations’ names and the support services offered. The content of the board was further evidence of a practice whereby the genetic counsellors passed on their clients to other appropriate services when they felt the need to refer their clients for further counselling and support, rather than providing the counselling themselves in clinic. This emphasised the medical practice of genetic counselling for familial cancer risk.
Another clinical practice undertaken by the clinicians is visible in Figure 7, which is a photograph of a document titled “extended family relationship”. The document displayed, using various shapes and lines, the appropriate or official way of displaying genetic family relationships in a pedigree. This specific way of documenting genetically related individuals and familial cancer was a practice ingrained in the work of familial genetics, playing a fundamental role in the mapping of “families” across time and enabling the calculation of familial cancer risk estimates (see Chapters 5 & 6 for further discussion).
The Clinical Geneticists

The two clinical geneticists had their own offices next to each other. Although their roles were complementary, there were also differences in their positions, work and expertise. Trish who trained as a paediatrician and clinical geneticist was second in charge of the Unit. Trish had joined the Unit a few years earlier working on the increasing number of familial cancer cases where endocrinology and metabolic disorders also exist.\textsuperscript{22} Her work provided a close link between the general genetics unit, involving collaboration on research and patients with the general genetics unit, as well as health professionals in other hospitals.

As head of the Unit, John had been there since the Unit’s inception and had degrees in science and medicine, specialised training in paediatrics, and a PhD in laboratory genetics. These qualifications made him both a specialist clinical geneticist and a genetic pathologist. Such qualifications resulted in both great economic and symbolic capital (Bourdieu 1975, p. 25). As well as being a practising clinician, John published articles and appeared on public television and radio. I once accompanied John when he presented ethical issues of general genetics to high school students in a rural location. At the time of fieldwork the issue surrounding the patenting of genes (which allowed for genes to be owned and all royalties to be paid to the discover of the genes involved with mutations) was being fought in the Federal Court in Australia (see Chapter 8). John was a strong advocate for the removal of the patenting of genes. These examples of his work roles demonstrate the range of topics and audiences he engaged with.

\textsuperscript{22} Endocrinology disorders were hormone disorders, and metabolic disorders involved problems caused by missing enzymes.
John was considered by his colleagues to be extremely good at establishing and maintaining work relationships between allied health professionals (e.g. surgeons, referrers) and the Unit. He did this by collaborating with them and linking them with information, research and clients. Having access to the latest information and important and powerful sources of knowledge proved beneficial to John, and those he collaborated with, because the transaction of knowledge resulted in more information being dispersed to colleagues, clients, research, media and the community. This collaborative practice reflected John’s strong immersion in the field, clinic, families and the public, roles which both worked to build up John’s knowledge and helped define the knowledge. It is an example of how medicine “switches” itself from objectifying patients and their parts, to reinvigorating individuals and communities with humanist thought (Latimer 2013a, p. 147).

John once informed me that the Unit’s clients wanted them (the clinicians) to be “cultural heroes” and that he too wanted to be thought of as such in the community. Latimer (2013a, p. 142) recounts how the geneticist, through presentation of one’s work as helping to save lives and prevent unnecessary suffering, reconfigures themselves as both having knowledge about genes and syndromes and, at the same time, reflecting a medicine that is “heroic and humanist”. John worked to obtain such status by viewing community involvement as an enterprise. He used the knowledge he had acquired to inform the public, work colleagues and the biomedical profession at large, of the benefits and value of the knowledge he had attained throughout the years. This practice of informing others reflected his belief held about the positive value of the Unit’s work. It demonstrated how scientific authority is a particular kind of capital that can be
“accumulated, transmitted, and even reconverted into other kinds of capital under certain conditions” (Bourdieu 1975, p. 25).

It was common for the clinicians to justify the value of trying to know about familial cancer risk and or mutations to their clients (see Chapters 6 & 7). However, whereas they expected to provide this justification to their clients, they did not expect to do so with medical colleagues and fellow allied health professionals. Despite this, a number of health professionals did not share the same belief in the importance of their work which largely involved prediction of risk and uncertainties. It was common to observe the two clinical geneticists sharing and expressing amazement over the opposing views held by others. In these circumstances they expressed how they also sought to educate and enlighten the other health professionals of their work’s validity. It was important for the clinical geneticists to substantiate their positions and the service as valuable to the community.

The clinical geneticists were the gatekeepers who oversaw most of the decisions made and actions by staff of the Unit. Their power lay in their capacity to decide who became a client, in defining cases of risk, deciding who was tested for a genetic mutation, which genetic mutations were tested for, and the provision of certain types of verbal and written information and knowledge. All of the clinicians sought to uncover familial cancer related mutations in their clients, with the rare and unusual mutations bringing greater value (see Chapter 8). By discovering mutations, the clinicians increased their symbolic capital, and progressed the science of genetics, while sharing the value of their findings with their clients who wanted certainty through the knowledge. As detailed in the process of becoming a client (see Chapter 6), the clinicians’ roles also overlapped in deciding who became a client, but the final decision rested with the clinical geneticists. As clinical
geneticists they had the medical education and with that knowledge came the power to make decisions regarding the provision of risk calculations, counselling frameworks, and what information and knowledge was imparted to the client.

Staff Satisfaction and Socialisation

Coffee, Bare Feet and Whistling

In a Unit dedicated to the prevention of familial cancers, I largely observed members of staff working in a relaxed atmosphere. The atmosphere reflected and reinforced the individual satisfaction that members of staff gained from their work, and the positive working relationships they had with each other. Members of staff expressed themselves in numerous ways that revealed a sense of pleasure and satisfaction in their work. Activities in the environment demonstrated and contributed to staff members’ sense of satisfaction. These included the strong smell of fresh coffee which permeated throughout the building daily, various staff members walking around the Unit in socks or with bare feet, and the common sound of staff whistling, humming, or laughing together. Whether the staff members were in their offices or not, the doors of the staff offices mostly remained open, only rarely closing when privacy was required for a private meeting or phone conversation. Not having doors creating a boundary between workers demonstrated availability and helped create a relaxed and informal environment, common in open-door policy environments (Hall 1969, pp. 136, 138).

All the members of staff tailored their workspaces, to varying degrees, so that they were personalised to them and reflected their personalities and interests. It was common to see photographs of their pets and children, pictures their children had drawn or pieces of clothing. There were also differences in what was displayed. Whereas one staff member
kept sea monkeys in a fish bowl, another displayed religious ephemera, while another had a computer screensaver that displayed three handsome Hollywood stars, which was often commented upon by visitors to the Unit. Research into work culture by anthropologists, sociologists, and psychologists have identified a number of factors related to the personalisation of work areas. These include identity, gender, emotion, culture, job satisfaction, work productivity, control, territoriality, status and company policy (Elsbach 2004, p. 101; Scheiberg 1990, p. 334; Tharp 2006, pp. 4, 5; Wells, Thelen & Ruark 2007, pp. 616, 617).

A relaxed working environment was also evident in the way that staff dressed. The members of staff conformed to a dress code of casual attire that was neat and professional, allowing for certain amounts of freedom of expression. There were expectations, but generally the dress code allowed the staff the freedom to change their attire to adapt with the outside temperatures. There was little difference in clothing worn between the genetic counsellors and the research nurses, both of whom saw clients, when compared to the administrative staff who did not see clients. The two clinical geneticists were the only exception, wearing covered shoes, long pants and collared shirts, usually with long sleeves. Their act of dressing more formally reflected the notion that with their positions came greater expectations, not just in the holding of certain knowledge, but also expectations surrounding dress. Such expectations consider doctors as members of a distinguished profession and by dressing accordingly (in professional dress) they uphold the image of doctors as responsible and competent. This way of dressing is also said to influence trust and confidence during the medical encounter (Dancer 2013, p. 346; Rehman et al. 2005, p. 1279).
The Use of Humour

During interviews with individual staff members, all emphasised their enjoyment in their working roles which they largely attributed to the relationships held with their colleagues. Evidence of positive relationships held between staff members was clear in multiple ways inside and outside of the Unit. In the Unit, and during work hours, humorous jokes were emailed to one another and it was a common occurrence to observe, from an outsider’s perspective, outrageous or sarcastic remarks by staff. For example, during one Monday clinical meeting the staff were informed that one of the clinicians had spent longer than usual in the appointment because the clinician found the client attractive, which resulted in laughter and various comments.

Humour could also be seen as a necessary and valuable part of their clinical work, playing an important part as entertainment and a coping mechanism while providing information. Medical anthropologist and psychiatrist Rob Barrett (1987, pp. 252–257) found and documented examples of how staff used humour in a psychiatric institution as part of their clinical work. From his work and other research conducted on humour in other organisational contexts come some common findings. These include that humour is a form of coping used to manage numerous anxieties and unresolvable problems that staff encounter in the health care system. It enables the expression of negative feelings about patients’ behaviours in a socially accepted “safe” way. It prevents burnout as it enables staff to separate themselves from the feelings of anger and grief that can be aroused by their work with patients. Other reasons for using humour include that it is an informal mechanism for negotiating status hierarchies in a multidisciplinary team, and helps build

23 Staff members who had worked in the Unit from the beginning mentioned that the positive work relationships between staff members had not always existed. They mentioned a very “turbulent period” in the past. Experiencing work place turmoil made many of the staff appreciate the current workplace relationships.
rapport, reduce tension or stress, distances self from threat, normalizes feelings and creates camaraderie in a group (Barrett 1987, pp. 252–257; Barrett 1996, pp. 85, 92, 93, 169; Huang & Kuo 2011, p. 141). Much the same explanations could be applied to the humour practised by staff in the Unit. In particular, demonstrations and explanations provided by staff showed that joking and humour helped staff release tension, resolve role conflict and reduce boredom, while bonding staff together.

The Role of Food and Drink

Food and drink played a significant role in the Unit and was not separated from work. Whether at Monday clinical meetings, or other staff work gatherings, food, and in particular sweet cakes, was shared. It was common for all members to sit down together, at a non-designated time, in the meeting room for a morning or afternoon “chat” over fresh percolated coffee. During this time, work related conversations and non-work related conversations took place. It was also commonplace for various foods, for example biscuits or nectarines, to be brought in from staff members’ homes to share. In the Unit, both the male and female employees, regardless of job role, made the fresh percolated coffee, however, all food was prepared or brought in by the female employees. Through enrolling staff members in particular relationships, food and drink are objects that can configure the gender identity of employees and the organization (Valentine 2002, p. 15).

Figure 8 (see next page), is a photograph where all staff worked in a production-like setup to prepare annual newsletters to be sent out to clients considered at high risk of familial cancer. John requested one genetic counsellor to bring her popular homemade brownies specifically for the process. The brownies, along with fresh percolated coffee, emerged as
an incentive to work harder, and a reward for employees’ efforts, while highlighting the power of the “boss” to request and the genetic counsellor to provide.

![Image of staff preparing annual newsletters](image)

**Figure 8. Photograph of staff preparing annual newsletters to be sent out to clients considered at high risk of familial cancer – alongside homemade brownies requested specifically for the process.**

Interestingly the genetic counsellors who had worked in the Unit the longest viewed their work-food relationship as due to a common interest in food while the newer members of staff however, viewed the extent of the practice as particularly unique to the Unit. Food as a “thing” has been shown to play a variety of roles within different organizations – but the role of food, and its effect, can impact on social relations in various ways (Valentine 2002, p. 3). In a study on food, place, and identity, Valentine’s examination found food and drink as “non-human entities”, things that helped build, maintain and stabilise links between staff (Valentine 2002, p. 2).

**More Than Just Breakfast**

Outside of the Unit, other than the Monday clinical meetings, the only other regular meeting involving all members of staff took place every fifth Monday of the month at a cafe near the hospital. Taking place prior to work, the group breakfast went for
approximately an hour and involved the staff members sitting together at one table enjoying breakfast together. During this time work was not discussed, rather, personal conversations surrounding children, travel, books and current news events took place.

A couple of days prior to the ritual breakfast approaching, I was always reminded by various staff members. This act intrigued me, because this did not occur in any other circumstance. Towards the end of my fieldwork I was informed at length of the breakfast meeting’s importance, which made apparent why it was important that I observed the regular gathering. It was explained that the group breakfast had grown out of the period of staff dysfunction, and so it helped keep staff engaged while reinforcing the importance of team bonding. The breakfast was a deliberate act to include everyone together within one team, in order to make all staff feel worthwhile. The breakfast provided recognition that there was a division between the groups, based on the separate roles and duties, and that the group breakfast was a regular way of trying to amend the division.

The purpose of staff reminding me to attend this out-of-work gathering, as opposed to other work gatherings, was both so I witnessed this important team bonding exercise, and so I was included into the group. In one of the case studies provided by Valentine (2002, p. 12), an organisation’s regular practice of having a “sit-down meal” was a way of diffusing staff tensions and temporarily erasing workplace hierarchies. In this context, food aided in directing, stabilising, and depoliticising the staff and their relationships. This example, along with the examples above, demonstrate how the range and roles of food served different purposes.
The regular occurrences mentioned above, including the breakfast gathering, individual expression and staff interaction, contributed towards the overall feeling of a Unit where staff felt comfortable to express themselves in certain ways with each other. Ethnographers’ studies of organizations and their inhabitants have commonly found an importance placed on autonomy for soliciting worker pride, enthusiasm, and effort (Hodson 2004, p. 8). These innovations have been described as changing the individual’s relationship with work, rather than transforming the organization of work production, leading to “pleasure in work” (Donzelot 1991, p. 251). In the Unit, the atmosphere both reflected the generally positive workplace where members of staff were happy in managing certain aspects of their work, and it reinforced the behaviour and the bond between staff members.

Gossip and Socialisation

Gossip appears to be a well-known phenomenon in both historical and contemporary societies (Crnkovic & Anokhina 2010, p. 12). An agreed-upon definition of gossip is difficult however, because of a lack of consensus concerning the phenomenon’s precise delimitations (Manaf, Ghani & Mohamed Jais 2013, p. 35). A generalised definition of gossip views it as any informal talk about other people who are not present (DeBecker 2005; Westacott 2000, p. 66).

Various disciplines have contributed to research on gossip including social science (Manaf, Ghani & Mohamed Jais 2013, pp. 34–44), sociology (Shibutani 1966, p. 9), psychology (Rosnow & Fine 1976, p. 8), organisational studies (March & Sevon 1988, pp. 429–442), and the health sciences (Laing 1993, pp. 37–43). In anthropology, a range of research has considered the social function, rules and place of gossip, viewing gossip
not as idle, negative behaviour, but as an important political tool in which power and values are maintained and transmitted, group unity is reinforced, and conflicts are resolved or exacerbated (Brison 1992, p. 15; Gluckman 1963, p. 307). Others view gossip as a type of informal communication, involving information management and exchange for profit or mutual benefit (Paine 1967, p. 278; Rosnow & Fine 1976, pp. 31, 35).

The fieldwork notes I gathered mainly came from events told by staff who perceived, and subsequently chose to retell, an occurrence in a certain manner. Staff chose, either subconsciously or unconsciously, the way in which they retold the story, the words which they used, what was said and left unsaid, and where and when emphasis and varying tones were used. The study of gossip is useful in understanding the telling of stories, however the term gossip can be problematic in the sense that there are different types of gossip with “true” gossip typically taking place between small intimate groups, where ethnographers as outsiders are often excluded. Keeping this in mind, the common practice of gossip that I observed in the Unit took place during the act of debriefing. A new theory of gossip, which supports my findings in the Unit, views gossip as a type of communication that is coupled to, or embedded in, other communication forms, both informal and formal (Mills 2010, p. 213). I argue that the act of gossip, as a process where value-laden information was communicated, was part of the practice of informal and formal debriefing in the Unit.

Debriefing and Gossip

The daily practices of debriefing by the staff of the Unit, in particular by the clinicians, across various settings demonstrated how such accounts were informative and integral to roles and practice within the fieldsite. Debriefing by recounting stories is an accepted part
of the culture of biomedicine and often called a “debrief”. The clinicians used debriefing the most, reflecting how debriefing is taught as “reflective practice” and encouraged as part of education and training as medical professionals. The role of gossip in the socialization of nurses has been found to function as information, influence and entertainment (Laing 1993, p. 37). Although all staff contributed to gossip, it was typically undertaken by the clinicians and in particular the genetic counsellors because they saw the majority of the clients and therefore had stories to express and share. This maintained the exclusivity of the clinicians and reflects Gluckman’s (1963, p. 308) findings that gossip unites and maintains staff relationships through the sharing of morals and values, and the controlling and regulation of outsiders.

Whether the clinicians retold a stressful client encounter in order to “get it off their chest” or shared a humorous event, it was an act of debriefing and gossip. Not all debriefs and gossip were the same; the act depended on the people involved, the object, the purpose and the effect. Two types of debriefs involving gossip could be differentiated: “formal”, or expected debriefs, and “informal” or ad-hoc debriefs. Whether “formal” or “informal”, gossip via debriefs served different functions from helping to cope, helping with practical understandings and dealing with uncertainty.

The “formal” debriefs by the clinicians, scheduled during the weekly held Monday clinical meetings, always contained gossip and so were a mix of case management and entertainment. The debriefing involved each clinician providing a description of the clients that they had seen the previous week, which consisted largely of medical information and details about the client’s case (see Chapter 4). The gossip element of the debrief occurred when humorous details or comments were added, by either those
providing the debrief, or by fellow staff in response to what they heard. Such comments included jokes about a humorous name displayed on a pedigree, something a client had said during clinic that was considered funny or the physical attractiveness of a client. As explained above, humour played an important role in the Unit – in debriefing, the use of humour was a mechanism for entertainment and coping. It demonstrated how gossip could be fun, providing an escape from “monotonous drudgery” and a release from routine and stress (Delbridge & Noon 1993, p. 7).

During these formal debriefs, it was also common for staff to gossip about unfortunate circumstances, such as the impact of cancers on a family. The genetic counsellors informed me that it was not appropriate for them to debrief at home, to inform their spouse or family about work details due to client confidentiality. This revelation revealed another reason as to why the clinicians debriefed so extensively in the workplace. Such a finding supports research by Waddington (2005, p. 35) who found that gossip is a feature of nurse’s emotional labour providing the opportunity for the expression of nurses’ true feelings about patients and colleagues, which cannot be expressed in public. In comparison to formal debriefs, “informal” debriefs containing gossip were also a mix of case management and of entertainment, however they were not confined to a specific space and time in the Unit. Instead, “informal” debriefs occurred at all times during the day and were, the majority of the time, triggered by phone calls from clients, tests results, or when the clinicians returned from clinic appointments. For example, when a genetic counsellor returned from visiting a client in their home, they often recounted to fellow colleagues anything from interesting medical details, family relationships, to commenting on the state of their home or the tea provided. Such commentary termed “surveillance work”, by Latimer, was considered a normal practice in the health care of child and
reproductive health when making assessments of the family (2013a, p. 83). Informal debriefs also occurred outside the physical space of the Unit, within the hospital including within the hallways, the lifts and in cafes while staff waited for their coffee or lunch order. Outside of the hospital space, informal debriefs took place in airports, cafes, cars, and at an interstate conference.

Rosnow (2001, p. 216) and Shibutani (1966, p. 20) have noted that when there is an absence of context and concrete information, people gossip to try and make sense. In this way, gossip may decrease the feeling of uncertainty which arises in circumstances of environmental ambiguity. Whether formal or informal, involving jokes or gossip, the act of debriefing by staff, and in particular by the genetic counsellors, played an important role in the daily routine of the Unit. Gossip delivered through debriefs helped in the exchange of information for individual gain or mutual benefit, provided entertainment and a coping mechanism, and was productive in building a positive work environment. If gossip is considered as playing a vital role in group formation, regulation and perpetuation (Delbridge & Noon 1993, p. 6), both the clinicians that shared the gossip, and the staff who listened were implicated in the practice.

The Meeting Room
In the centre of the Unit was the meeting room. The only room with a designated meeting space, large table and whiteboard, it served many purposes, which made it a significant space. The meeting room was the most consistent space that I was able to observe and in which I could obtain the most knowledge relating to familial cancer and the running of the Unit. It was made particularly significant because it was here that the weekly Monday clinical meetings were held (see below).
Although clients did not physically enter the Unit dedicated to their care, their presence was obvious and existed in other forms. When entering the meeting room it was impossible to ignore the abundance of client card files, the historical collection of a client’s medical familial information filed tightly together in bookcases that covered two of the walls (see Figure 9).

Figure 9. Photographs of the client card files in the meeting room.
On the two adjoining walls of the meeting room, there was a whiteboard and a large pinup board that displayed a mixture of work and non-work related items. These items included posters detailing the clinical guidelines for the calculation of familial cancer risks, alongside postcards that had been sent from members of staff while on holiday.

The Monday clinical meetings were the only regular work-related gathering where all twelve staff members attended for all, or part, of the proceedings (see Figure 10). Centring on the management of various aspects relating to clients’ cases, it was where all members of staff took turns in informing colleagues about their work before leaving the meeting. The Monday clinical meeting was the principle setting where individual job roles, teamwork and hierarchy were displayed and reproduced. As explained in the next chapter, the clinicians were the last members of staff to remain and work through clients’ cases together. This work involved constructing and reconstituting individual clients and their families into cases so that they were appropriate objects of their work.

Figure 10. Photograph of the meeting room prior to a Monday clinical meeting with coffee, milk and clinic appointment books ready for the meeting to begin.
When meetings were not being held, all members of staff regularly used the room to grab a client’s card file, to chat over a coffee, or to have lunch (together or separately, depending on the day/time). The room was also used to celebrate common cultural occasions, including Christmas (see Figure 11). Having a blend of work and non-work related social interactions occurring in the same space, and sometimes at the same time, made the room an interesting place in which to observe staff dynamics and work of staff.

Figure 11. Photograph of the meeting table ready for the Christmas celebration. The breakfast consisted of drinks (including juice, coffee and wine), homemade stewed fruits, creamed rice, yogurt, croissants with jam and cream, along with bonbon crackers and an exchange of gifts.

Conclusions
The working environment of the Unit, along with the key work roles of staff have been described and analysed in this chapter. The examples demonstrate how different roles and practices effected and perpetrated the practice of familial cancer work, in specific ways. These include a focus on clinical work, group hierarchy, team building, work satisfaction, and the use of humour and gossip through informal and formal debriefing. The work provides a background for understanding the next chapter which demonstrates in various ways in which staff worked in defining and constructing clients, families and familial cancer risk.
Chapter 4. The Construction of Clients, Families and Familial Cancer Risk

In my fieldwork site of a genetic counselling and genetic testing Unit, all members of staff were involved, to varying degrees, in the creation and reproduction of understandings around what a client, and a family, were and were not. The construction of clients and families in particular ways played a pivotal role and was essential to the work of the Unit. Relevant beliefs and practices arose from, were in accordance with, and contributed to ideologies that surrounded medical, genetic, and familial cancer knowledge in biomedicine. Furthermore, indications that the Unit and their clients were different from other hospital patients came in many forms. To be detailed in this chapter, these major points of difference included the term “client” that staff members chose to use for the individuals who utilised their services, the requirements needed to become a client, and the roles of staff which enabled the prediction of familial cancer risk.

Terminology as a Clue to Practice

In the introduction of this thesis, it was explained that the service users of the Unit were referred to as “clients”, rather than “patients” as might be expected in a medical setting. The decision to use the term “client” demonstrated both how staff viewed those who used their services and also the services the staff offered. According to the *Collins dictionary* (2014), the word “patient” denotes someone suffering from illness and/or a person who is receiving medical care. The term “client”, according to the *Collins dictionary* (2014), is derived from the Latin word cliēns, and means to listen or follow and describes a person who seeks the advice from a professional.
Both terms have been argued as problematic and debated by scholars. The use of the term “client” to refer to a patient dates back to 1970, when Pluckhan (1972, pp. 1, 2) considered the term “patient” to be inappropriate for a healthy person seeking “health-maintenance advice or going for an annual physical examination” (Wing 1997, p. 287). Since this time, scholarly debate has advocated a preference for one term over the other, while others argue that the choice of term should be situation specific. The term “patient”, it is argued, should be used in acute care situations and “client” used with preventive health care (King 1986, p. 51; McCallum et al. 1989, p. 151; Shum 1989, p. 150; Wing 1997, p. 288). Such a definition fits with the Unit’s preference, their use, of the term “client” over “patient”.

In the Unit, staff preference and use of the word client rather than patient (and any other term) demonstrated a critical difference in whom they saw and what they practised in contrast with the other Hospital Units. Ultimately their clients were not considered as patients because they did not meet the parameters that made one eligible to have the label of a patient. The majority of the clients they saw were not considered to be “sick” with familial cancer but considered to be at risk. The clients of the Unit who attended genetic counselling sought expert genetic explanation (and not medical care) about their current (at the time), or past experiences of cancer, and/or to obtain information and possible genetic testing about future familial cancer risk.

“Probands” and “Presymps”

All referred clients were considered at risk of familial cancer, however there was a differentiation made. The service further categorised their clients by dividing their
referrals into two categories, termed as either “proband” or “presymps”. The term proband clients simply referred to the first client of a family referred to the Unit.

The term presymps, used on a daily basis in-house by staff was short for presymptomatic. Genetic counselling and possible genetic testing was offered to first and second degree genetic relatives of a client who had a mutation identified in a cancer related gene, or those who were considered likely to have a genetic mutation based on their familial history. The people who became clients after they responded to being risk notified were considered as presymps. According to staff, presymps was used to describe individuals who had been risk notified because they considered them at high increased genetic risk of developing familial cancer. Various other descriptions have been used by scholars to describe individuals considered to be at risk of, but not suffering from, familial cancer. These include; “presymptomatically ill” (Yoxen 1982, p. 144), “patients without symptoms”, “perpetual patients without symptoms” and “asymptomatic patients” (Finkler 2000, p. 58; Finkler, Skrzynia & Evans 2003, p. 408).

Although the term presymps was used and favoured by staff in the Unit, it remained an in-house term because the word “predictive” instead of presymps was considered more appropriate, and was therefore required to be used in verbal and written communication with colleagues and health professionals external to the Unit. The clinical geneticist explained that although the terms were often used interchangeably in genetics, presymps implied that a client who currently had no symptoms would inevitably suffer from the disease that they were at risk of (such as in Huntington’s disease). In contrast, while a positive result in “predictive” clients indicated an increased risk compared to the general population, it did not mean that they would suffer from familial cancer.
The decision to use the term presymps in-house, regardless of the external use, and opinions and requirements by other units to use the word predictive, reflected and propagated how staff viewed their clients as being more than at risk – that the information the clients received from them would result in earlier diagnoses of familial cancer or the reduction or prevention of familial cancer.

I was able to attend one presymp clinic appointment (because it involved one of my participants who gave permission for me to be there) however, I was not allowed to attend or audio record any others. Whereas genetic counselling students were able to attend and assist in clinic appointments involving presymps, myself and visiting medical students were excluded. This reflected the view held by the genetic counsellors that clinic appointments with presymps involved the most “vulnerable” of clients, who required private appointments. They were viewed as being more vulnerable because they had physical medical evidence (via a confirmed mutation result in a biogenetic relation) supporting the belief that they had a greater risk of familial cancer. The finding of a mutation resulted in a diagnosis of familial cancer risk. Both the preference for, and use of the term client, along with the additional categorisation of probands and presymps, was used by staff of the Unit, further differentiating their clients from other types of clients or patients while demonstrating how familial cancer clients were viewed in specific ways.

24 The decision to allow me or other visitors to observe presymps appointments was discussed by the clinicians at various times. Although some staff acknowledged that presymps should not be exempt from any study, that their experiences should be documented and accounted for, the final decision rested with the genetic counsellors involved, and in each case they decided to conduct the clinic appointment on their own.
The Process of Becoming a Client

Referral of Possible Clients to the Unit

Before individuals became clients of the Unit, and accessed its services, they required a medical referral: a recommendation in writing from a medical practitioner (e.g. a General Practitioner (GP), surgeon or gynaecologist) to either of the clinical geneticists of the Unit. The coordinated referral system was established and required by the national medical health coverage system, known as Medicare.

National guidelines and visual guides regarding identifying and managing familial cancers were provided to medical practitioners. In general the guidelines for a referral in relation to identifying a familial cancer risk were simplified (see page 31). The clinical geneticists of the Unit also wrote and distributed familial cancer related electronic newsletters three times a year to the medical practitioners. The newsletters were sent out to inform medical practitioners about current knowledge and best practice, and to help facilitate the correct identification of people at increased familial risk of cancer. The committed practice of helping inform and educate medical practitioners demonstrated two things. First, it acknowledged the important role that medical practitioners had in referring and managing clients. Second, it demonstrated the rapidly changing state of knowledge and technology applying to aspects of familial cancer, which the clinical geneticists felt a duty to provide.

According to staff and statistics produced by the Unit, the greatest number of referrals came from doctors in metropolitan areas of the city, and the majority (68%) of all referrals were in relation to the risk of familial breast and ovarian cancer. Referrals about familial bowel cancer came in second (25%), and the remaining (7%) dealt with rare
disorders (Unit document 2009). Given the dominance of familial breast and ovarian cancer referrals, the majority of the Unit’s clients were female. As stated previously, the majority of their clients were Anglo-Saxon and very few were considered, by staff, as culturally and linguistically diverse (CALD). According to some staff, the lack of CALD clients reflected the limited number of CALD individuals referred to the service. The lack of referral was also attributed to the differing beliefs and superstitions held around illness causations and their difficulty to change.

Two recent Australian studies also describe how cultural beliefs and explanatory models, amongst Arabic-Australians (Saleh et al. 2011, p. 465) and Chinese-Australians (Yeo et al. 2005, p. 174), have implications for genetic counselling practice, affecting access to screening services, communication of diagnosis of cancer and management. Another influencing factor was said to be due to the risk calculations used to explain and predict familial cancers, as predominantly based on Anglo-Saxon and Jewish populations and therefore more relevant to explain and predict familial cancer in these populations.

When the service received a referral from a GP, it was either initiated by the GP or facilitated by an individual who went to the GP with knowledge or suspicion about their familial cancer risk. Referrals from medical practitioners, such as surgeons and gynaecologists, were also received. These usually occurred after their cancer patient’s family health history, cancer type, or biopsied results indicated a possible high risk of familial cancer.

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25 Rare disorders included: multiple endocrine neoplasia, familial melanoma, retinoblastoma, neurofibromatosis type 2, von Hippel-Lindau disease, and familial brain cancer.
In Chapter 5 I discuss the Braxton family, a family that was considered by the clinicians as unique and special because they had a rare occurrence of having two different familial cancer related mutations identified in their genetic relatives through genetic testing. When I interviewed Mary, one of the members of the Braxton family, as to what events brought her to the Unit, she explained that it was because of her daughter’s obstetrician. She described that while her daughter was pregnant with her first child the obstetrician collected a family history from them and commented on the prevalence of cancer in the family and the availability of genetic counselling. As Mary had already had ovarian cancer, and her sister breast cancer, Mary pursued the opportunity to have genetic counselling as she was interested in learning if their cancer experiences were due to a “familial error” and, if so, to determine if their children and/or grandchildren had an increased risk of developing familial cancer. Mary’s knowledge of a possible high familial cancer risk was communicated to some family members (including her sister whom had suffered from breast cancer), which prompted them to visit their GPs and ask for a referral to the Unit.

This example demonstrates how Mary and her family may not have known about the Unit and their high risk, nor subsequently discovered two different mutations in genetic relatives, if not for her daughter’s obstetrician. Earlier I explained that the National guidelines and visual guides regarding identifying and managing familial cancers were available to all medical practitioners and that the Unit also provided addition resources to them. Mary’s GP and surgeon (and her sister’s) were either not aware of the prevalent history of cancer in their family which placed them at high familial cancer risk, or they were aware but chose not to pass on the knowledge and refer them to the Unit. Mary’s referral by her daughter’s obstetrician, and not by other doctors and specialists,
demonstrated how for her, and for others like her, a referral to the Unit was triggered by, and dependent on, which medical practitioners were consulted.

“Weeding or Keeping” Referral Process

When client referrals were faxed, emailed or mailed to the Unit, the secretary (Ella) created an electronic file and a physical folder. If the referred client had relatives who were already clients of the Unit, they were also attached to the biogenetic relations electronically in a sub-file. The electronic file was created and stored in FamilyTrace, while the card file was given to the “green dot” genetic counsellor, the genetic counsellor whose turn it was to look after the new referrals for that week. The “green dot” genetic counsellor was responsible for undertaking the first stage of what could be considered a “weeding or keeping” process, which decided whether the person being referred met the criteria to be seen (see page 31) and if they were considered at increased risk of familial cancer.

Weeding Process

The genetic counsellor looked over the referral letter, considering the individual’s history and checking if they had any familial history on their database. When those being referred did not meet their basic criteria, the genetic counsellor passed the file to one of the two clinical geneticists to review the decision. Making the final judgement, the clinical geneticist involved held the knowledge and power, either disagreeing with the genetic counsellor’s decision, or agreeing and “signing off” on the referral. The Unit’s “signing off” involved documenting, in FamilyTrace, that no further action was required or permitted until further information was provided or obtained and warranted further action. The clinical geneticist also responded via a letter to the referring medical practitioner.
explaining the reasons for their decision. It was then up to the referrer to inform their patient of the decision. Where the referral involved a client considered to be at only standard or moderate familial cancer risk, the letter was also used as a tool to inform and educate the referrer about appropriate referrals. The above process highlighted how incorrect referrals of individuals were “weeded out”, so that future occurrences of inappropriate referrals could be prevented.

Keeping Process

When the genetic counsellor and the clinical geneticist involved considered those being referred as fitting their criteria, and as requiring an appointment, they became a client of the Unit. The secretary sent the client a referral pack. This consisted of a cover letter listing preferences for the location of a genetic counselling clinic appointment, a brochure about the Unit, a legally binding document asking for the release of the client’s medical information, and a form for the client to provide their, and their family’s, health history. Two other forms, a “consent form” and a “risk notification” form were included and will be discussed further below.26

Defining Kinship and Family

Kinship is a complex concept, generally referring to particular types of connections between people which vary between and within societies and across time, and may encompass ties of blood, legal relationships, emotional connections and, most recently, genetic links. It has been argued by Finkler (2000, p. 3) that the new genetics continues to

26 All clients were required to sign the consent form in order to access the services of the Unit. Information sheets and consent forms with signatures of their clients and health colleagues were part of the daily routine and acted as a physical record and form of accountability. Applying Foucault’s ideas, documents are said to make participants visible, archived, classified, measured, compared and controlled on a mass scale, and they shape behaviour within organizations; thus they are a mechanism through which risk is managed (Jacob 2007, p. 251; Riles 2006, p. 18).
greatly influence peoples’ lives, including their family and kinship interactions, leading to the “medicalization of kinship”. The identification of genetic mutations has enabled the meaning and cause of disease to be re-negotiated and the creation of a new genetic kinship community. Family and kin relationships are said to be drawn into the current notion that diseases are genetically transmitted from generation to generation – hence, diseases are no longer related to individual responsibility, and the true patient becomes the entire family (Finkler 2000, p. 3; Franklin & Mckinnon, 2001, pp. 1–25, 262).

Finkler, Skrzynia and Evans (2003, p. 410) argue that in our contemporary Western society, the routine of divorce and marriage generates diverse patterns of family structure and conceptualisations of its meaning, so that a family may consist of any grouping established on the basis of choice (single parent, blended family, adoptive and same sex partners). However in genomic practices, relatedness consists in the transmission of biological substances at the level of DNA which moulds people into an idealised form of family and kinship (through genetic ties), contrary to the changing practices where personal choice of family and kin are largely favoured.27

I was informed by some clinicians, and read Unit documents stating, that the Unit did not have a definition regarding what constituted a family. However, as discussed below, the everyday work of the Unit, including the forms that clients were required to fill out, highlighted specific ways in which the Unit viewed and defined family. What I observed was the opposite from what I had been told by staff and what I had read. The reasons for this contradiction can be informed by the work of Malinowski who uncovered three levels of data while undertaking fieldwork in the Trobriand Islands (1922, pp. 1–25).

27 See Chapters 5 and 6 for further theoretical discussions on kinship.
The first level, which involved “what people say about what they do”, could be obtained by talking with people. The second, “what people actually do”, could be gained by observing their actions, and the third, “what they think”, could be gleaned from a collection of folklore and received ideas. The reasons provided by Malinowski for unearthing such differences in data included: social standing of the researcher and the knowledge of and protection of information (Malinowski 1922, pp. 1–25).

Although a definition of family was sometimes disputed, the written and verbal use of family definitions (evident in multiple areas of their work) were not only used, but integral to the Unit’s practice. Malinowski’s concept, reveals that such a contradictory finding is commonplace in the field. Although I certainly tried to understand the culture to which the members of staff belonged, I do not claim to know, or think it is possible to know what staff thought. Listening and observing staff in action enabled me to uncover that staff did work with particular definitions of what constituted a family. What staff said, and did, highlighted how particular notions surrounding what constituted a family were created, endemic, imparted and integral in their everyday work in the Unit. Some of these notions will be discussed here.

**Defining Family through Forms**

The content of the client’s family history form, which was used to create a pedigree (discussed below), was one example of a document made to capture particular information, information that was most relevant to the work of the Unit. There were two sections to the form. In the largest section, clients were to list their “relatives”, including those who had not had cancer. Also required were the relatives’ names, dates of birth, dates of death, whether the relative had cancer, tumour(s) or polyp(s), age of diagnosis,
and the hospital and State in which treatment occurred. The type of “relation” clients should list was specified on the form. It prompted the client to provide details about their parents, siblings, cousins, parents’ siblings, and grandparents.

The relations listed above and embedded in the form were considered as “first and second degree relatives”. First-degree relatives are said to be an individual’s parents, siblings or children. Second-degree relatives are the individual’s grandparents, aunts, uncles, nieces, nephews and grandchildren (Barlow-Stewart 2012, p. 4). In general, the service considered the most relevant information as that relating to the first and second-degree genetic relatives of the client, and then, any other genetically related members of the family affected by cancer. The need for information relating to first and second-degree relatives was based on a particular belief about shared biogenetic material. In genetics, relatives are said to be people who share genetic material: the more DNA sequence that people share, the closer the relation and family (Peters, McAllister & Rubinstein 2001, p. 155).

On the second part of the family history form were three smaller separate boxes. The first box asked the client to list their “partner/s and children” and provide details of any cancers, tumours or polyps. The second box asked the client to provide details about “other relatives” that had cancer (including what their relation was to the client). The third box also asked for the client to list any relatives of their children’s other parent that had cancer (including what their relation was to the parent). The form demonstrated how content was structured in particular ways so that clients provided certain information that had been pre-grouped into particular “types of relations”.

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Even with these structured guidelines, some clients demonstrated confusion over what details to include. This sometimes resulted in the clients only providing information about family members who had cancer, thus not including the details of the relatives that had not had cancer. This practice made cancer appear as the norm rather than the exception and impacted on the ability of the clinicians to produce a more accurate risk assessment. This occurrence was not isolated to the Unit, with Patenaude (2005, p. 119) also commenting on this occurrence during a familial cancer counselling appointment.

Other instances illustrated a tension evident between what staff were supposed to practice and what they did practice in relation to familial relationships. I had been informed that familial relationships were not meant be defined and documented as either social or biological; however, delineating relationships in this way was integral to providing relevant risk calculation or risk notification. An example of when a social relationship was allowed to be documented was when a family member had been adopted or where first-degree and second-degree genetically related family members had married and/or procreated within the family. These relationships were required to be defined; in the former, it was so that they were not included in the risk assessment and, in the latter, it was because the information impacted on the risk assessment of the individual and their genetic relatives.

Furthermore, the forms illustrated how only particular types of medical information surrounding familial cancer were sought. The examples provided here highlight some of the ways in which the biogenetic relationships of clients were sought and favoured in the Unit. It also became evident that through the provision of information (filling of forms),
that the client and family were required to show a commitment and capacity to participate in the practice of genetic diagnosis, an argument further explored later.

**Acknowledging the Practice of Risk Notification**

Staff members were part of extending the boundaries of ideologies surrounding medical, genetic, and familial cancer knowledge in biomedicine. The formal practice of “risk notification” was a prime example of how these boundaries and ideologies were extended. As written earlier, a form seeking permission from the client to conduct risk notification was always included in the forms sent off to potential clients. The particular form of risk notification practised by the Unit involved gaining signed permission and information from the client for the service to contact and offer genetic counselling (and possible genetic testing) to certain biogenetic relations who were also considered at high risk of familial cancer (see Chapter 7). The process of risk notifying other genetically related relatives did not take place until a client considered at high risk had gone through genetic counselling (see Chapter 6 for detailed discussion on this process). For clients considered at high risk of familial cancer they were then reassured in the clinic appointment of the advantages of providing consent and encouraged to pass the information on themselves or provide details for risk notification to go ahead.

Clients did not need to sign the risk notification form in order to become a client but they were required to sign a consent form that required the client acknowledge the role of the service in providing risk notification. Part of the consent form read:

I understand that a diagnosis of familial cancer may have implications for other members of my family and that I may be asked to assist the Service in informing them.
This demonstrated how clients were required, before attending genetic counselling, to sign a consent form that acknowledged the potential role of the service in informing other members of their “family”, should a mutation be found. Everett (2007, pp. 375, 379) raises classical anthropological questions about the nature of gifts and commodities, describing a tension between individual and collective rights to things, a tension that extends to the body and products that come from the body. Through the required acknowledgment of the practice of risk notification which was embedded in the consent form, tensions surrounding the collective rights, the sharing of familial information between the individual client, the family and the service were demonstrated.\(^{28}\)

**The Processing of Clients**

When a client returned their forms, the data officer entered the details the client provided into FamilyTrace and the originals were placed in a card file. As analysed in the next chapter, the clients who sent back their completed forms and attended genetic counselling appointments were individuals who were pressured to go by family members, or who saw value in learning about the information provided and/or believed in the advantages of being informed about their familial cancer risk (like Mary mentioned earlier).

When forms were not returned, the individual was reminded three months later, and then again at six months before the referral was cancelled and the referring doctor notified. Due to confidentiality guidelines, I was not able to contact those who were referred to the Unit but chose not to return forms and/or attend genetic counselling. Staff expressed that they did not understand why individuals did not want the beneficial information they could provide. Staff considered individuals who chose not to attend or proceed with

\(^{28}\) See Chapter 7 on risk notification for further detail and analysis.
genetic counselling as both unfortunate, for having missed out on the opportunity, and unaware of the benefit of the information that the clinicians would have provided to them. As discussed in greater detail in Chapter 7, similar opinions were expressed towards “non-responders”, biogenetic relations who were risk notified but chose not to respond.

The Creation of Pedigrees and Risk Status

The fascination with family genealogies in the form of a pedigree is said to be common throughout the “West” and has a long history which can be traced back to biblical pedigrees in eighteenth-century Europe (Bouquet 2000, p. 173). Although an interest in family history occurs throughout all cultures of the world, the “Western” interest in family genealogies has taken on particular form, in the way of pedigrees. Palsson (2002, p. 338) argues that family records are phenomena with a social life, informed by “hierarchy, authority, citizenship and control”. This argument views such records as products of the contexts that produced them, and not untainted descriptions of relations and histories that many believe (Bouquet 2000, p. 187).

Latimer’s (2013a, p. 87) research in medical genetics describes how: “The family tree engages family in the construction of the family’s formation, and associates issues of health, illness, and deformity to the form of the family”. Similarly, when clients of the Unit provided their family history and consent they became part of a practice that helped to redefine, or cement existing notions of family and risk of familial cancer. The information that the client provided was used to create a “family pedigree” in an electronic pedigree drawing program. To be detailed below, a family pedigree was a physical timeline displaying certain details about relatives in a tree-like structure.
The “enrolment” of clients and families in the creation, and use, of pedigrees in the Unit was essential to the operation of the Unit.

**Family History Books**

Some of the clients also provided family history books, which provided another source of information to the clinicians. A collection of approximately a dozen copies of family history books sat in the Meeting Room. Clients had given the books to the Unit to keep or copy. The family history books provided extensive access to selected aspects of a client’s familial genetic and social history (see Figure 12).

![Figure 12. Some of the family history books in the Unit.](image)

Generally the family history books included photographs, pedigrees, timelines and interviews. Of particular interest was how some of the books stipulated that they were for “family use only”, yet somebody had provided the book to the Unit, demonstrating how the ownership of, and about, shared genetic information varies (see Chapter 6 for further discussion). It was said by the clinicians that, in general, the clients hoped the information the books contained could be used to risk notify appropriate relatives, if not at that time,
then in the future when medical advancements in mutation identification improved. The familial records reflected their clients’ interest in their family history – as they searched for information not only about their risk of familial cancer but about where they came from, and to whom they were related.

Latimer in her work on medical genetics of dysmorphology, described how once family members supplied personal, and social parts, through photographic and other representations of family members (accounts, histories), the work of dysmorphologists made them into things – “the currency of the clinic” (2013a, p. 170). Similarly, the family history books supplied became clinical currency through the copying, scrutiny and assessment by the staff. The collection was appreciated by the clinicians who used certain information from them to trace familial cancer and extend clients’ pedigrees.

The most comprehensive family history book in the Unit was that of the Gilmore’s beginning in 1583 (see Figure 13, next page). The collection had a prologue detailing the “many hundreds of hours” worked on the book and the ongoing nature of the work that required it to be updated yearly. Other than providing information such as who and how relatives were related, 1,200 out of 4,000 images had been selected, images that provided various snapshots, mostly of Australian history and culture. Images included: maps of homes; modern and historical photographs of individuals and families; of cars, land, horses, wildlife, gravesites, school classes, certificates, and letters. The images had been collected from various sources: family members, newspapers, the Internet, birth, death and marriage notices, national archives of Australia and the public records office.
On one page of the book there was a copy of a letter from an interstate familial cancer service with the heading “note to all descendants of the Gilmares”. The letter detailed how a familial cancer mutation had been found and mentioned the availability of genetic counselling for descendants. The insertion of this letter demonstrates how family and kinship are being medicalized as a result of the current emphasis on medical genetics and its clinical application, an argument made by Finkler (2000, pp. 181, 188, 210). The growing power of the “new genetics” is said to come “not through brazen and totalizing claims but manifests through subtle strategies suggesting that life is governed through genetic composition” (Betta 2006, p. 55). In this case, the insertion of a letter into the Gilmore’s social and historical family book, reshaped an illness from being an individual matter to a familial matter. Framed as a familial matter it includes both family histories and potential family futures.

The family history books kept in the Unit also highlighted the power and knowledge surrounding what the clinicians knew about their clients and what information they went
on to share. As discussed in further detail in Chapters 6 and 7, for the practices in relation to genetic counselling and risk notification, the clinicians had the knowledge and power to determine what knowledge they provided their clients with and what they did not. In relation to the Gilmore’s family history book this involved the clinicians being “careful” not to disclose to a particular client the knowledge that they were genetically related to this family (see Chapter 7, “The Maintenance of Genetic Information as Confidential and Private”).

The Creation of Pedigrees

Anne, as the data officer, was the only staff member other than the clinicians who “created” pedigrees from the information the clients provided in the forms. The difference between their constructions was that Anne used technological equipment in the form of the FamilyTrace database to create pedigrees, while the clinicians also frequently used a pen and paper, in addition to the database. The development and ability of FamilyTrace to manage clients’ familial data, cases and create pedigrees illustrates Sharp’s (2000, p. 309) argument that associated inventions with the new genetics has led to the commercialisation of minute body fragments, such as genes.

Before discussing the role and the importance of the pedigree to the work of the clinicians, I briefly describe first how Anne created the pedigrees using FamilyTrace. Having received each client file, Anne added the name of the client into FamilyTrace. When the client’s name was recognised in FamilyTrace, the system returned matches. Anne then used information relating to relatives’ names, dates of birth and diagnosis to match them to the existing family. The information from the new client was then added to any existing pedigree. Anne particularly enjoyed matching the clients and viewed each
one “like a puzzle to solve” as it was often the case that different members from the same family had differing information regarding relatives’ names, dates of birth, and diagnoses. 

As discussed later, the type and extent of information provided by the client impacted on the risk assessment provided, with the wrong types of information, too little information, or too much information being problematic.

When Anne entered a client’s name into FamilyTrace and it was not recognised as matching already existing clients then Anne created a new pedigree. The FamilyTrace system made the actual process of creating a pedigree quick and simple. Using the system Anne inserted a box. Each box stood for each family member and had the name, date of birth, date of death and a number which symbolised their genetic position in relation to the proband. As previously noted, the term proband describes the first family member who became a client and is widely used in medical genetics. It was important to identify the proband, so that the relationship to other individuals could be seen and patterns established. With each addition of a box, a screen prompted for the input of the relationship between the client and family member. The pedigree consisted of peoples’ details, straight lines, boxes shaded in and numbers (see Figure 14).

Figure 14. Photograph of a pedigree created in FamilyTrace and printed out across 2xA4 paper. Photograph is intentionally small and slightly distorted to maintain anonymity.
Different number values were applied to each incidence of cancer mutations found in family members. The number given depended on the number, and type, of cancers and/or genetic mutations, and whether they occurred in first, second or third generation family members. The type of mutations involved influenced the risk provided because of the possible impact. Staff also considered the ethnicity and gender of the client (and their family members), because in families that had more males there was little chance for a breast cancer related mutation to express itself, by way of cancer. Using the tallied numbers, the clinicians created a risk status. The lower the final number, the lower the risk, and the higher the number, the higher the client’s risk of familial cancer.

When creating pedigrees Anne once expressed that the clients were “not just names on a pedigree”, and seeing the many experiences of cancer through families, particularly in younger females, made her “hyper-vigilant on health”, and empathic for what some families go through. With every click of the mouse Anne created a virtual pedigree that made certain relationships, and the impact of familial cancer (or the lack of thereof) visually apparent. Some pedigrees were considered very small in scale with only a few boxes and lines, while others spanned many generations, with the relationships hard to understand for someone unfamiliar with pedigree structures. It was also apparent when looking at the familial relations on the pedigrees that the contents were unique when compared to other ways of documenting family relations. Kinship studies have a long history in the discipline of anthropology, with early cross cultural studies using biogenetic relationships as a starting point. It was Schneider’s work that challenged the idea that kinship relations consisted of reproduction and blood. Instead Schneider argued that blood kinship was an “American”, “biogenetic” construct, a Western construct that was not necessarily shared by people around the world (Schneider 1980, p. 23).
In the pedigree, the names and ages of individuals were added under standard symbols used to denote gender, cancer, other illnesses, and deaths (see Figure 14, page 134). Lines joined the symbols to illustrate relationships and births, while lines drawn between the symbols (as strikes) showed divorce, remarriage and adoption. By focussing on genetic kin, family members were related in specific ways, sometimes referred to as “kinning” or “re-kinning” (Howell 2006, pp. 8, 15). Many academics have commented on the difference between the genetic family mapping which reflects and promotes “traditional” notions of family and relations, and actual social familial relationships (including Finkler, Skrzynia & Evans 2003, p. 410; McLaughlin & Clavering 2011, p. 400; Svendsen 2006, p. 146). Traditional notions of the nuclear family, comprising of a male provider, female homekeeper and their children living consanguinity (bilateral and descended from the same ancestor) derives from Europe in the 6th century A.D. (Finkler 2001, p. 237).

Termed the “post-modern family”, some now argue that there is no longer a dominant family structure, which was brought on by transformations of routine separation, divorce and remarriage in the 1960s. These have generated diverse and fluid families that are not blood related. Established by choice they include single parent households, blended, adoptive and gay families (Finkler 2001, p. 238; Finkler, Skrzynia & Evans 2003, p. 410). Finkler, Skrzynia and Evans (2003, p. 410) comment that it has been necessary to bring back the traditional pre-1960s definition of a family in “biogenetic terms” for the purpose of genetic counselling even though they may be “frequently at odds with a person’s experience”.

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29 Bilateral decent is where equal weighting was given to both blood parents decent lines, with identity of the child born being derived from both parental decent lines (Finkler 2001, p. 237).
Genetic discourse is considered a new strategy in reproducing and privileging the heterosexual nuclear family form as the norm (Crabb & Augoustinos 2008, p. 303). The development, and use, of this particular form of pedigree, where “blood” or biogenetic relations are key, reflects notions held in the bio-medical culture. The construction and the interpretation of a pedigree draws from knowledge of genetics and its mode of transmission for particular genetic illnesses (Novas & Rose 2000, p. 503). Whereas the traditional biomedical model was based on the doctor and sole patient, the new genetics brought with it a reversal – requiring various family members be involved for a diagnosis to be made (Finkler, Skrzynia & Evans 2003, p. 409). The new genetics with its tools (such as genetic testing) have given genetic relatedness a renewed status, allowing little room for culture and social construction (Palsson 2007, p. 62). The result of such notions and tools is said to be “the medicalization of family and kinship” which changes people’s experience of reality and has profound ethical and practical dilemmas which can be both debilitating and empowering (Finkler 2003, pp. 403, 411). Emerging knowledge about genes has the ability to radically transform family relationships (Chilibeck, Lock & Sehdev 2011, p. 1768). Such ethical and practical dilemmas are demonstrated and discussed throughout this thesis.

In the Unit, once the pedigree had been matched to other related pedigrees, and added to, various copies of it existed in the system. The corresponding pedigrees were printed out and placed in the client’s file. Only the information the client provided about their family was shown to the client during clinic appointments. As explained in Chapter 5, in clinic appointments only limited versions of the pedigrees were shown to clients, added to and changed as needed, and used to inform clients of their familial cancer risk.
The next step in the processing of clients involved providing the client with a genetic counselling clinic appointment. A client’s priority for an appointment depended on whether they were probands or presymps. As stated earlier, the demand for the Unit’s services outweighed their capacity, which resulted in waiting periods for new clients of up to six months for proband clients. Appointments for presymps were to be held within six weeks, reflecting how they were considered at further increased risk (because a mutation or high risk had already been identified in their families) and in need of information sooner (see Chapter 7).

Case and Client Management and Construction

Risk Calculation (Prior to Monday Clinical Meeting)

Two weeks prior to a client’s appointment the genetic counsellor assigned to the client consulted the client’s electronic and physical file using the referral letter, and then using the information provided by client and any existing knowledge held about the family, they produced a risk calculation. Risk assessment models (empirical scoring systems) were used to calculate and determine a client’s risk. Based on the Mendelian principles of inheritance, the three main risk models which were utilised to calculate a client’s risk, were called, “Manchester”, “BOADICEA” and “BRACAPRO”. All the models differed in terms of the cancers they included, the populations they were based on (e.g. British, Jewish), the family history required, and their exclusiveness (e.g. what to include). The Manchester model was the only model which was designed to be calculated by the clinicians, whereas the others required software (Evans & Laloo 2010, pp. 237, 244).

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30 The genetic counsellor phoned the client if clarification of any information was deemed necessary.
The pedigree and risk assessment models were very important and considered by the clinicians as being more useful than mutation tests in revealing a client’s risk of familial cancer. Such a belief was also evident in the clinicians’ practice of providing a risk status to clients based solely on the contents of their family pedigree. The belief in the traditional method, of a family pedigree being more useful than the ability of the genetic screening for a mutation, was also shared with clients during clinic appointments (see Chapter 5 for one example). Even with genetic testing, only a small per cent of familial cancer mutations (one in five) were detectable. Most clients ended up with a risk assessment based exclusively on their family tree (see Chapter 6 for explanation of genetic testing mutation results). This indicated how the knowledge obtained from translating the family pedigree was privileged over a genetic mutation test. This was not unique to the Unit but reflected the limitations of the genetic mutation testing. Similar findings have been commented on by Svendsen (2006, p. 144) and also reflected in the genetic community with a news article quoting a cancer geneticist who said: “Family history remains the best genetic tool we have…It’s the best-kept secret in health care” (Eng & Scheuner 2010). As discussed further below, the reliance on the family trees, made up of family information can be problematic, due to unknown knowledge, or misunderstandings about health and illness within families and across generations. Although it was apparent that the clinicians privileged the pedigree over genetic mutation tests, in contradiction, the practice of performing mutation tests was still used in conjunction with many risk assessments when the clinicians believed it would result in the finding of a mutation. Such a practice highlights the belief in, and need for, biomedical evidence, diagnosis and validity.
As described in the following section, the clinicians who interpreted the pedigrees prior to and during the Monday Clinical Meeting did so because the pedigrees held knowledge that the clinicians used to produce knowledge about familial cancer risk by designating clients into risk categories. The client’s pedigree in the Unit was the gateway to familial cancer knowledge for the clinicians, the client and relatives involved because (as detailed later in Chapter 5) the clinicians used the pedigree to produce a familial cancer risk and then inform each client about their risk during genetic counselling appointments. The interpretation of pedigree information sometimes paved the way for mutation testing and/or risk notification of particular genetic relatives (Chapter 6).

**Risk Calculation (During the Monday Clinical Meeting)**

Up until this point in the process of client management, the administrative staff and the clinicians all worked individually but as part of a team to process and identify clients considered at increased risk of familial cancer. Further decisions made prior to a client attending clinic occurred collaboratively during the Monday clinical meetings. In Chapter 3 I introduced staff hierarchy and roles and explained that it was in the Monday clinical meeting where I was able to see how the staff and their roles worked together and separately to manage clients of the Unit. The meeting was the only time in which all staff of the Unit came together and contributed at various points to manage and discuss clients’ files, pedigrees, risks and the complexities surrounding familial cancer. The type of cancers discussed at the meeting were most often breast and bowel cancers, reflecting the majority of the referrals, but other familial conditions were included to a lesser extent.

For the Monday meetings, although no seats were assigned to staff, where they sat around the table, both initially and by the end of the meeting, largely depended on when they
arrived and when they were expected to leave the meeting. The genetic counsellors were usually the first staff members to arrive, and were always the last to leave. Generally, they sat at one of the longer sides of the table positioned in between the two entry doors because it allowed for remaining staff to enter and spread around the table with ease. Staff members filled their cups with coffee and milk, passed around food that may have been baked or brought in for the meeting, and were well into conversations when the clinical geneticists, John and Trish, entered and sat at the remaining free seats at the head of the table. Their last minute attendance reflected tight constraints on their time, often coming from meetings or phone calls.

John led each meeting and when John was away, Trish was in charge. Following an agenda, the structure of the meeting was always the same, only changing when relevant staff members were absent or late. John began the meeting by greeting everyone, welcoming back those who had been away and introducing any visiting doctors or students. The first agenda item was “social news” where current or planned staff absences were discussed, and it was during this time that staff provided details about conferences attended or recent holidays. The next item on the agenda was reserved for any pressing issues that were of immediate concern and was the only agenda item where all staff contributed. Various items were raised during this time; examples included new software, reminders of a staff farewell dinner approaching, or a question posed about the approval process for sending blood samples overseas for testing.

While every staff member was still present, the next agenda item was usually opened with a vigorous drum roll on the table by John and the question posed “any new mutations?” or “any mutation families?” The purpose of this practice was to build anticipation amongst
all of the staff members and it signalled the approach of the possible exciting news that a “new” mutation, and/or a previously undiscovered mutation, had been found. Where none of the clinicians’ clients had been “found” with a mutation then the next item on the agenda was brought forth. This happened the majority of the time, as four out of every five of the genetic tests came back as negative or equivocal – meaning that the results were unclear (see Chapter 6 for discussion).

The clinicians shared when a mutation had been found in one of the genetic samples provided by their clients. The staff member in charge of the case provided the name of the mutation and shared any details that they considered as relevant about the client or family in which it was found. In Chapter 8 I discuss how the identification of a mutation in one of their clients resulted in displays of excitement. The more common mutations (e.g. BRCA1) were greeted with a cheer or applause in recognition of the find. When a rarer mutation or when an unexpected mutation result was found, the clinician involved took more time to explain the case surrounding it, and fellow staff responded with increased interest. The finding of a mutation was seen as significant and resulted in displays of enthusiasm, because it was rare to find a mutation. Genetic testing for a mutation was only performed on individuals who met certain criteria and in whom staff believed a mutation would be found (see Chapter 6). The finding and sharing of the mutation was understood to be empowering for the individual client and the clinicians involved. Mutation identification was thought to display which clinicians were more accurate in their predictions and was considered as resulting in greater information and knowledge to manage the familial cancer risk of clients and families. With rare and unusual results being shared with other clinicians (via research or genetic material), the clinicians were also at the forefront of the discovery process (see Chapter 8 for more information).
After any mutations had been reported, Ella then brought up administrative issues that staff needed to be aware of. Anne reported on her progress on the completion of pedigrees, and every month she reminded the clinicians that she was going to collect the death certificates of recently deceased clients and/or clients’ relations. When Anne and Ella did not have anything else to discuss, they were thanked before returning to their desks in the next room. The focus then shifted to Dean who asked questions, or updated his colleagues, about issues related to the running of FamilyTrace, or other technological equipment, before leaving the meeting. The research nurses then provided updates on the studies they were undertaking. This included informing the clinicians of any difficulties in recruiting certain clients for their studies, or about new findings made in studies that they, or their interstate colleagues, were working on. They then left the meeting unless the clinicians informed them to stay because discussion was turning to a client’s case which the research nurses may have been interested in for recruitment.

The above practice, where each staff member reported and then left, continued until only those with the greatest amount of contact with clients remained – the clinicians. The formal debriefs then occurred. Each genetic counsellor brought to the meeting a pile of client folders. These files were divided into clients that had been seen in clinic the week prior and clients that were to be seen that week. The client files were further separated into probands and presymps. The formal debriefs involved each genetic counsellor reviewing and discussing the details of the clients that they had seen in a clinic appointment the previous week. A client’s name, age and reason for referral along with the client’s history related to familial cancer, their risk and plan of action, was discussed. The Monday clinical meeting provided a structured time for medical decisions to be made. The accounts often also contained a mixture of seriousness, humour, gossip, facts
and innuendos made by either the clinician involved, or by others in response to what they heard.

Following this discussion, the clinicians discussed the clients to be seen in clinic the following week. Other than providing the name, age and details of referral, the printed pedigree was looked at by the genetic counsellor and the clinical geneticist together so that an agreement could be made on the risk status and recommendations to be provided to the client. The seat movements at that time were reminiscent of musical chairs as each genetic counsellor took turns moving next to the clinical geneticist. The physical action of sitting together was an integral part of the meeting and was so they could both look over the file and pedigree together to discuss and decide the risk categories in which clients would be placed. Looking over a pedigree the clinicians could be considered as going “back to the future”. The clinicians went back in time, reflecting on the past, through the memory and pedigree of their clients and then using current knowledge of genetics and medicine to trace the history of cancer in their family. This process was required in order to predict the possible impact on future generations and recommend appropriate surveillance or surgical measures to reduce or prevent the chance or severity of familial cancer and/or prolong life. It was impressive to watch and listen to the clinicians interpret the pedigree – at a quick speed, they distinguished between familial inherited cancer syndromes and non-familial cancers, along with associated conditions which provided clues as to what type of mutation they were possibly dealing with, how it had moved through the family, and the risk of familial cancer.

The meeting process allowed for a snowball effect where most staff members expressed, disagreed, clarified, justified and sought help, leading to diligence, greater understanding
and confirmation that the client/case had been, or would be, managed appropriately. The impact of a client having a lack of kinship information, the wrong type of information or too much information became apparent at the Monday clinical meetings. In most circumstances, the more information the client provided the better as it enabled a more thorough grounding on which to make a decision about their risk of familial cancer.

A Lack of Familial Information

When people lacked knowledge of their family health history, it affected their chances of becoming clients of the Unit and the risk assessment made by the clinicians. There was often confusion and a disconnection between what clients knew in relation to their own history, and what they knew about their biogenetic relations. The incorrect information was not just in relation any cancer diagnosis or health problems family members had suffered or died from, but also to differing accounts of birth and death dates, marriages and siblings. In relation to the health history of family members, the national guidelines that the Unit followed recommended that where there was no medical evidence of cancers having occurred, then such information was not to be used to calculate the final risk figure (NHMRC 1999, pp. xi, xv).

When clients had no information about their family, it was within guidelines to offer genetic testing, based on this limited knowledge of family structure. In those circumstances the clients were considered as having a greater chance of finding a mutation. One genetic counsellor commented on the irony of a client having more chance of having a mutation found with less familial information, than clients who had an abundance of cancers in their family.
In one circumstance, a genetic counsellor planned to inform their client that they were at moderate risk of familial cancer based on information reported by the client that her sister had a familial cancer mutation identified. With the genetic counsellor not able to find evidence of the existence of the mutation on their database, during the clinic appointment the client was informed of her moderate risk assessment being dependent on a mutation having been identified. And so, the client was asked to provide evidence of the mutation. This demonstrated how the clinicians privileged certain knowledge about family history by a client, which they determined to be adequate, medically confirmed and correct. The clinician explained that during the genetic counselling appointment the client cried and admitted that she had made up the existence of the mutation because she wanted genetic testing. Due to a poor relationship with her family, although the client was aware of many cancers in her family members, she was unaware about who was affected with what forms of cancers, and if mutations had been identified. This demonstrated how much some clients wanted genetic testing. In this case, the client tried to hide their lack of information and say that a mutation had been identified, which she thought would have resulted in her being offered genetic testing – which she thought would provide a definitive yes or no answer in relation to her own risk.

The Wrong Type of Information

In general, the more information the client provided the better as it helped inform a more thorough risk calculation. There were two main exceptions to this. The first, mentioned earlier, occurred when the information provided by the client was either the wrong type of information, because it was not useful, or the information the client had provided comprised of inaccurate or differing accounts. Earlier, I discussed how the emphasis placed on biogenetic information and meanings had implications for the social definitions
of family that contributed to the complexity of staff work. The second exception occurred where there was not enough knowledge provided by a client due to unknown biological paternity and/or maternity associated with adoption, artificial insemination, or because of a lack of family knowledge due to communication breakdown or death. In some cases staff held knowledge and information about biogenetic family members that they knew was different from, or unknown to, the client, and it was used to inform their clinical decisions unbeknown to the client. In instances when clients asked staff directly about being adopted or whether certain family members had been to genetic counselling, staff members had the power to withhold knowledge, responding that they were not allowed to disclose that information. Due to adherence to confidentiality and privacy laws (ComLaw 2014), staff were very careful in not relaying such information to the client. The clinicians largely managed this by having different copies of family trees, with only the one corresponding to the information provided by the client being shown during clinic appointments. This demonstrates a critical difference in the transfer of knowledge between clients of the Unit and the clinicians in the clinic. Clients were required to provide the Unit with information about the certainty of relatives, whereas the clinicians were only required to provide familial cancer risk knowledge back to the client and not information or certainty of biogenetic relations. However, as argued in Chapter 7, through the practice of risk notification the clinicians managed and undertook particular practices that attempted to provide certainty of biogenetic relations by contacting and sharing familial cancer risk information to those deemed as being particular biogenetic relations to the client.
**Too Much Information**

Although a lack of information and the wrong type of information was problematic, too much of the *wrong* kind of information caused problems for staff due to the cost of storage. During one of the Monday clinical meetings one of the genetic counsellors expressed being overwhelmed by “papers” of information a client had provided. The client had many health problems, and genetic testing had discovered a mutation. The genetic counsellor suspected the mutation was from the client’s father’s side, but the client did not have any information on her father’s side, with the papers of information about her mother’s side. The clinical geneticist gave the genetic counsellor permission to make distinctions and include only the important information (relating to the “side” they were interested in) on FamilyTrace due to the costs of storage space on the program. This example demonstrated the degrees of information – how clinicians using clinical judgement decided and selected what they considered to be useful information and worth including, and what they did not consider as useful, and not worth including.

**Classifying and Disclosing a Client’s Risk Status**

The biopolitics of genetic risk is riddled with risky estimations that gloss over the uncertainties embedded in rapidly changing molecular genetic knowledge. (Lock 2012, p. 141)

In Australia, the National Health and Medical Research Council (NHMRC 1999) has outlined clinical practice guidelines for familial aspects of cancer. The guidelines were prepared by “panels of experts” in the field, and were based on evidence; where there was no evidence, the advice provided was based on expert opinion (NHMRC 1999, pp. preface, xi). As a guide it was “not meant to be prescriptive” but rather was designed “to provide information to assist decision making” and “to be followed only subject to the clinician’s judgement in each individual case” (NHMRC 1999, p. viii). The clinicians of
the Unit largely followed the guidelines, which included the classification of risk status. Whether there was, or was not, appropriate NHMRC classification for risk status, the client’s risk of familial cancer was determined on the basis of clinical judgement by one of the clinical geneticists. Clinical judgement in each case was needed because of the “rapid evolution of knowledge in the field” (NHMRC 1999, p. xiii).

The familial cancer risk status a client was considered to have depended on the familial cancer it concerned. According to the NHMRC guidelines, “virtually all” individuals who inherit a gene mutation that predisposes them to Familial Adenomatous Polyposis (FAP) will develop colorectal cancer (NHMRC 1999, p. 13). In comparison, the lifetime risk for cancers in individuals with a mutation in BRCA1 or BRCA2 are 40%-80% for breast cancer, 11%-40% for ovarian cancer, 1%-10% for male breast cancer, up to 39% for prostate cancer, 1%-7% for pancreatic cancer and melanoma (Petrucelli, Daly & Feldman 2013). The large difference in the numbers above, demonstrate the uncertainties surrounding the estimation of the impact of a mutation on one’s risk. With the majority of the Unit’s client referrals concerning breast and ovarian cancer risk, the information below focuses on how breast and ovarian familial cancer risk was defined.

**Clients Considered at Risk of Breast and Ovarian Cancer**

During the Monday clinical meeting, one of four risk categories was decided on and assigned to a client independent of any genetic mutation test. The categories were: “at average or slightly above average risk”; “moderately increased risk”; “potentially high risk”; and “high risk” (Unit’s Guidelines). In the beginning, a client’s risk was based on three main factors: one, the information the client had provided; two, any additional information the service already held on family members; and three, the “risk model” or
criteria the clinicians used to inform their decision, which as described earlier was condition specific. The first and second factors were closely linked. In most cases the risk category a client was placed in was only as good as the information that the client was able to provide about themselves and their family’s health history, as well as any reports of cancer the service was able to verify through medical records.

The purpose of the clinicians identifying a risk category was not just to inform the client but it served a purpose in informing what level of surveillance and risk reducing options the client could access. The surveillance and risk reducing options for clients belonging to moderate and high-risk categories were similar. Without going into the details specific to each category, there was a range of measures including regular self-examination for breast changes, clinical breast examination, mammographic screening every year, participation in clinical trials to prevent cancer, prophylactic mastectomy and oophorectomy, ultrasound screening for ovarian cancer, cessation of oral contraceptive use, hormone replacement therapy and colorectal cancer screening.

The category the client was assigned into was important and could be viewed as a type of membership, with different knowledge, recommendations and access to services. In a pamphlet provided to women identified with a BRCA mutation and/or those considered at high risk of developing breast and ovarian cancer, it noted that high risk women were the “winners” of a new clinic considered a “one stop shop”. The clinic enabled the client during the one visit to see a breast surgeon, gynaecologist and genetic counsellor, and have access to surveillance tests (such as mammograms, or breast ultrasound).
Actual and False Risk Categories

During a client’s initial clinic appointment, they were provided with information about familial cancer, and ethical and practical considerations, and they were “counselling” in relation to their risk (see Chapter 5). During the Monday clinical meeting when clients’ risk assessments were decided, it became apparent that the clinicians sometimes decided to inform a client of a different risk status than that to which the clinicians believed they belonged. This was demonstrated by the clinicians either making the client’s risk higher or lower, depending on whether the clinicians believed that the client would have benefited from knowing the truth. Of the times this was observed, it only involved clients who were regarded to be in either the moderate and high risk categories because the surveillance and preventative measures were largely the same.

For example, in one case it was agreed that a female client, who was considered by a genetic counsellor to be highly anxious, be informed that she was moderate risk rather than high risk. The decision was said to be made because, as stated above, the options available to the client were largely the same for both. The clinicians therefore viewed it to be in the best interest of the client for her not to be informed of her actual higher risk status. In another instance, it was decided during the meeting that a female client would be informed that she belonged in the high risk category, rather than the moderate risk category in which she was considered, because the clinicians believed the client would be complacent when informed that she was moderate risk and would be more likely to act on being informed she was high risk. Foucault argued that with knowledge comes more power, and that each society has its own “regime” of truth and the means by which it is sanctioned (Foucault 1980, p. 131). The decision by the clinicians to provide the client with an alternate truth demonstrated how the clinicians negotiated a client’s risk based on
their own assumptions of how the client may have interpreted, handled and ultimately appropriately managed knowing the risk category in which they were judged as belonging. This also illustrated how the clinicians had the power to not only classify but to disclose a client’s familial cancer risk.

The Power to Classify and Disclose Risk

An explanation as to why the clinicians manipulated the truth, in the way described above, was influenced by the explanatory model they followed – which considered an individual’s response to their risk as occurring on the basis of a perceived risk, and not an actual risk. The psychological framework which underpins this model argues that perceptions vary depending on whether information is framed in a positive or a negative way and is important to consider because it effects the health decisions that are made (Marteau 1999, p. 425; O’Doherty 2005, p. 24). The clinicians of the Unit knew that their clients held their own perception of their familial cancer risk. As knowledgeable and powerful health professionals they were in the position to assist some of their clients by providing a “real” or “false” risk category to help shape the information in a certain way, with the aim of influencing clients’ risk perception and subsequent health decisions.

Power is pervasive and, in organisations, power is said to be embedded in everyday life and a dynamic social process affecting the emotions and behaviour of the people in the organisation (Hardy & Clegg 2006, p. 632). Individuals became clients of the Unit and by being a client of the Unit they were subject to power relations. The clinicians wielded substantial power in deciding the weight they gave influencing factors and the category in which the client and their family members belonged. They also demonstrated significant
power during the clinic in informing clients about the risk category to which they were allocated.

Conclusions
This chapter has described the intake, diagnostic and classification process that all the staff members of the Unit contributed to and undertook in differing ways, which resulted in the particular reproduction of understandings around what a client, a family, and risks were, and were not. It has demonstrated how the clients provided family history information which was judged by the professionals, the clinicians, as being adequate, not enough, the wrong type, or too much. The clinicians used the information to categorise clients with a risk and then finally they decided how to inform the clients of their actual or false familial cancer risk.

The next chapter describes the practices occurring in the clinic, where genetic counselling was a medical practice largely focussed around the dissemination of particular genetic knowledge surrounding familial cancer risk. Genetic counselling was not separate from the diagnostic process mentioned above; rather, it was in clinic where classification was further constructed. Chapter 5 also further demonstrates particular aspects of uncertainty, knowledge and power surrounding familial cancer information. In particular, the chapter will discuss how clients were imparted with knowledge by clinicians, and how the clinicians engaged and managed the uncertainties and tensions surrounding autonomy and confidentiality surrounding an individual’s right to genetic information and familial cancer risk.
Chapter 5. Genetic Counselling, Knowledge and Power in the Clinic

Previous chapters have described some of the different ways in which it was apparent that there was a focus on familial cancer as largely a medical practice and not a psychological practice. The emphasis placed on clinical work was not just reflected in the working environment of the Unit, but it was also evident in the genetic counselling clinic appointments, an argument extended in this chapter. The main service provided to clients of the Unit was genetic counselling, known as “clinic”, undertaken in a clinical appointment space.

A brief history of genetic counselling begins the chapter followed by a description of the clinical spaces and general sequence of the clinic appointment. This chapter then provides insight into how genetic counselling was a clinical practice rather than a psychosocial practice. The clinical (biomedical) emphasis involved imparting certain types of knowledges (through information provision), rather than the provision of mental and emotional support (therapeutic and psychological counselling) to clients. The clinic was not separate from the diagnostic process, extending on the diagnosis and risk estimates undertaken prior to the clinic; it was in the clinic that classification was further constructed. The clinicians concentrated on identifying clients at increased risk of familial cancer, imparting to the client particular knowledge about familial cancer and risk, and some ethical and physical considerations. This chapter is about the expression of particular knowledge and power in the clinic – how clients were informed to various extents about their familial cancer risk, the extent to which the clinicians imparted
knowledge about what was “known” and “not known”, and some of the clients’ reactions to uncertain information.

The Practices of Genetic Counselling

A History of Genetic Counselling

Along with the emergence of genetic technologies in the 1970s, and the ability to predict and/or test for genetic mutations, a new emphasis was placed on treating disease “susceptibilities” rather than diseases (Rose 2007, p. 113). In acknowledgement of the complexities inherent in such practices, and a belief in the importance of its provision, the practice of genetic counselling was developed and used in adjunct to the technology. The two separate disciplines, genetics and genetic counselling, were developed to work together to achieve understandings of genetic knowledge and how genetic factors influence the health and psychological well-being of individuals, families and communities (Collins 2005, p. viii). The term “genetic counselling” was created by Sheldon Reed, an American genetic scientist in 1955 who described genetic counselling as “a kind of genetic social work without eugenic connotations” (Reed 1975, p. 335).

Genetic counselling was said to have allowed for three key contributions in the area of medicine and genetics, leading to improvements in the health and psychological well-being of clients (Collins 2005, p. viii). The first contribution was to enable clinical geneticists and genetic counsellors to work as a team to understand how both genetic and non-genetic (environmental) factors interact. Second, the team works together to determine how individuals, families and communities learn, integrate and use knowledge about genetics. Third, as a team, the clinicians were both contributors and leaders in dialogue and decision-making related to genetics and concepts of “identity”, “race” and
“ethnicity”. A more critical view, as discussed further in the chapter, considers the explicit aim of genetic counselling to affect the client’s health behaviour (Rose 2007, p. 113).

**Global Practices**

The recognition of genetic counselling (as an essential service), particularly in relation to familial cancer risk, is said to be growing throughout the world. However, complexities surrounding the accreditation and practice have resulted in differing education, accreditation and practices (Yashar & Peterson 2013, p. 687). Research conducted on genetic counselling across China, Hong Kong and the United Kingdom (UK) identified similarities and differences in the practice (Sui 2009, p. 392). Overall, Sui argued that the governance of genetic counselling and its practical application have their own characteristics within each particular society and its social contexts. For example, factors that contributed to the application of Chinese genetic counselling were economic conditions, the status of the genetic counsellor, the lack of coverage of the healthcare system, and the national population policy. In comparison, Sui argued that individual well-being and autonomous reproductive choice were more respected in the UK and Hong Kong (2009, p. 404).

In other case studies drawn from India, Cuba, Germany and Israel, Rapp (2014, p. xvi) also argues that medical practice, genetic testing, screening and therapeutic technologies vary across regions and countries. This often results in different dilemmas being raised about the presence and absence of BRCA testing (and other genetic tests) and knowledge production. The different practices and dilemmas are shaped by specific local contexts including historical consciousness, national regulation, medical education and laboratory
practices. Rapp (2014, p. xvi) considers the rapid increase in, and changes regarding, BRCA research, testing and treatments, as causing “genetic entanglements” because the significance and limitations are complex and not clear.

**Australian and Local Practices**

Even within Australia, various definitions and descriptions of genetic counselling exist, demonstrating the ambiguity of the practice. The Australian Society of Genetic Counsellors (ASGC) describe genetic counselling as:

> A communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions. (Resta et al., 2006 cited in ASGC 2011)

The Human Genetics Society of Australasia (HGSA), of which the ASGC is a part, also defines genetic counselling as a process but goes into further detail describing it as:

> The process by which patients and relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and of the ways in which this may be prevented, avoided or ameliorated, integral to this process are the clients’ and families’ emotional and psychological responses as well as social issues. These need to be acknowledged, addressed and supported for effective genetic counseling. (Harper cited in HGSA 2011)

**Definitions from the Unit**

In comparison to the above definitions, the Unit’s website wrote that the purpose of genetic counselling was simply: “to assist you in making the best decisions for yourself” (2013), placing emphasis on the individual. In the Unit, the clinicians emphasised information provision and non-directive discussions with their clients, however, as discussed further in, direction was provided in the form of information provision. The genetic counsellors were interviewed separately about what they provided to their clients during clinic. Three extracts from the interviews below provide different
perspectives about the role of genetic counselling, while demonstrating and explaining why information provision was emphasised.

In relation to a client that Sheryl (and I) had just seen in clinic, Sheryl said:

It’s about working out fairly quickly where that client’s at, and you probably noticed that I checked how she was and yeah picked up that she was pretty relaxed and would be ok about having a joke and making light of things, while still recognising that there’s a serious side there as well, I thought she had a lot of issues.

In the first example, Sheryl described how genetic counselling involved “reading people reasonably well”, checking how the client was at the beginning of the appointment and then providing the client with relevant information and gauging, using her experience, how the information was handled by the client. Although Sheryl recognised that the client “had a lot of issues”, she further described her role, saying:

it’s not to psychoanalyse or to do in depth counselling, it’s to make sure that I’m not traumatizing her with information...Cause our role is partly educative so it’s the information provision and there’s a counselling component to that, to make sure you’re not upsetting the client and you’re not stepping on toes and going in bombastically. But yeah if, if someone’s got obvious issues then it’s about then making sure you refer them on to someone else who can deal with that, another counsellor or whatever, not to take that on ourselves...A woman I saw last week her mother was dying and so obviously she was upset so you wouldn’t refer her on because it’s a normal grief process that she’s going through, so it’s about being able to recognise that.

The extract above demonstrated that the main role of genetic counselling, for Sheryl, was to provide educative information. Where a client appeared too upset, outside of what was considered normal, then the client was referred to either a psychologist or social worker whose work it was to work through the issues.31
In another example, when Sue was asked about the aim of genetic counselling, she began answering the question by explaining how clients sometimes came to the appointment knowing they had a family history of cancer, but not knowing what cancer was. Sue then said: “they (the clients) just think that they did something wrong, or it (cancer) just comes, cancer just comes and zaps you”. Due to this Sue found it integral to provide her clients with an understanding about why the prevalence of cancer in their family may have happened using genetic explanations.

In a third and final example, when May was asked if the term “genetic counsellor” accurately reflected her role, she explained how the need to provide information to the client dominated over providing “actual counselling” because the need to inform the client was linked with obtaining informed consent. For May the “actual counselling stuff” involved spending time exploring social and family issues.

we have a lot of information we need to give as well and sometimes it feels that it dominates over our other roles…And if at the end of the day you're doing a test and you need the informed consent then you need to give the information, you can't skip on it as then you're not getting informed consent. So I guess that’s my work battle, or internal battle, or something, is how much opportunity we actually get to do the actual counselling stuff.

The three examples provided above demonstrate different elements emphasized about the role, the aim and definition of genetic counselling. The first example explained how their roles were educative, the second example described how information provision was required because of clients’ misunderstandings of the area, and the third example, linked with informed consent, explained how clients needed to be considered informed appropriately to make the correct decision about their risk. All of the examples demonstrated and explained why during clinic appointments emphasis was placed on the
provision of information over counselling clients in relation to managing and processing the personal, familial and social issues.

Clinic Spaces and Clinical Emphasis
The clinicians of the Unit conducted clinic appointments on a daily basis, Monday to Friday. The majority of clinics occurred in either the separate outpatient area of Fairbank Hospital, or within other local and rural hospitals that had been chosen by the client from a list. Each genetic counsellor had a specific hospital site, or two, that they conducted clinics in, while the clinical geneticists shared attendance at the different locations depending on their schedules. The clinics were held in the outpatient areas of the hospitals where various other health units and departments saw patients. The outpatient areas used mostly specialised in seeing people either with cancer and/or pregnancy related health care needs, and so the rooms were set up for physical examination by nurses and doctors.

The Power of Clinic Spaces

Rather than being one definite sort of thing – for example, physical, spiritual, cultural, social – a given place takes on the qualities of its occupants, reflecting these qualities in its own constitution and description and expressing them in its occurrence as an event: places not only are, they happen. (Casey 1997, p. 27)

The Unit did not have room within their office space to see clients. The genetic counsellors commented that they (as a service) were not considered as important, when compared to other medical departments, to be provided with a larger space (by hospital administrators) in which they could both work and see clients. They perceived that their

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32 Occasionally appointments were conducted in the client’s home when clients were unwell or unable to attend clinic in a hospital.
comparatively lower status reflected the lower value placed on the largely preventative medicine which they practised when compared with treatment medicine. The staff members viewed this as unfair and unjustified because they believed that what they were providing was as significant as other forms of medical care to the health care of the community.

It was said by the clinicians that the reason the clinic appointments were held in clinical spaces within various hospital locations around the State was to enable the service to be accessible to all areas and parts of the community. Hospitals have “power” in biomedicine; they are usually the places where sick people go to see medical professionals, who have the power and knowledge to heal. Genetic counselling clients did not have to be seen in a hospital environment for physical or medical reasons, because the majority of the time, the only physical procedure undertaken was when a client’s blood was taken for genetic testing, this was not undertaken by the clinicians from the Unit, rather this was undertaken by nurses from the hospital or at a pathology collection centre.

Rather than for medical reasons, the location of genetic counselling in a hospital reflected and sustained relations of authority. The genetic counsellors did not have the same power as the clinical geneticists, as they were not expected to heal or be located in a hospital. By placing the genetic counselling in a hospital setting, the genetic counsellors were given a power that they would not have had if they worked outside of a hospital (in an office for example). They were placed “under” the clinical geneticists, which served to further empower the clinical geneticist and yet to also empower the genetic counsellors, who were viewed by clients to have medical authority and knowledge by virtue of the fact that they were located in a hospital. This demonstrated how genetic counselling clinics
were held within clinical spaces and within hospital environments as an effort to acquire medical authority. Using Goffman’s arguments, one could view the clinical location as an example of an “institution” involving a medical model that provided “medical servicing” and “tinkering services”, involving a technical expert server and an individual client in need of repair (1961, p. 321).

*The Influence of Clinic Locality*

With all of the local clinics being held in outpatient areas that either provided obstetrics or oncology services, the possible impact that it could have had on clients was sometimes dismissed, and at other times acknowledged by the clinicians. For example, in one circumstance a genetic counsellor exclaimed that it was “not nice” for their male clients to sit waiting with pregnant women. This comment acknowledged that there was a mismatch between males and females, and between pregnancy and cancer.

In another instance, after attending a regular clinic held in an oncology ward, I asked two of the genetic counsellors about the possible impact on clients attending a clinic in the oncology outpatient ward. I had asked this question after being unprepared and confronted by seeing people sitting in a large open area on reclining chairs each with a tube inserted into an arm vein which were attached to black intravenous bags. It was obvious that the individuals were sick and receiving treatment. As these occurrences would have been visible to those attending clinics I asked two genetic counsellors if the space impacted on their clients.

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33 I learnt later that the bags and tubes were black in order to mask to colour of the chemotherapy drugs.
After my question was readily dismissed, I explained that I found it confronting and thought that it could also be confronting for clients attending clinic with the purpose of finding out if they were at risk of familial cancer, or if they or family members already had cancer. I was informed by a genetic counsellor that cancer was “nothing to be afraid of”, that cancer was “a part of life” and that their clients attended the clinic because they were “used to cancer”. The statement that cancer was “nothing to be afraid of”, reminded me of an argument put forth by Susan Sontag while attempting to de-mystify the notion that cancer results in death. Sontag called for people to regard cancer “without meaning” as if it was a very serious disease, but “just a disease”, rather than a curse, a punishment, and not necessarily a death sentence (Sontag 1988, p. 14). However, whereas Sontag was referring to actual diagnosis of cancer, the clinicians of this Unit were dealing with familial cancer prediction and essentially wanted the clients to take responsibility and action (which sometimes involved surgery) to possibly avoid cancer that was also considered as “nothing to be afraid of”.

*Description of Clinic Spaces*

The clinic is the key institution which the new genetics must pass through in order to impact on society (Latimer 2013a, p. 7). All of the clinics, regardless of where they were held, had similar working procedures (see next section). As long as they were in a hospital setting then the clinic provided what was intended: medical authority, power and knowledge. Kirmayer (1988, p. 61) discusses the ways that medical practice distances the body from the person. The architecture of hospitals provide physical barriers between the sick body and the social person, which removes the patient from the possibility of relating to the institution on a personal or emotional level.
It is evident from the photographs above, that the spaces in which genetic counselling was undertaken were clearly medical spaces, rather than therapeutic-psychological spaces (see Figure 15). The rooms were not tailored in any way to make clients comfortable, and the only things used in the rooms were the chairs and the desk to put their files on. The sink made it appear as though the clinicians needed to wash their hands or wear gloves, while the bed appeared as though clients needed to lie down for examination.

It was clear that the clinic spaces were for the examination of sick people yet the majority of their clients at risk of familial cancer were not sick with cancer – and may never suffer from cancer. This highlighted a tension. Being at risk of familial cancer meant that the

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34 There was one instance where I observed a clinical geneticist examine the hands and feet of a client, but this was in relation to a different familial condition where this was a required part of diagnosis.
health status of clients was somewhere in-between being well and being sick. Whether those considered at risk are viewed as cancer “survivors”, “pre-symptomatic patients”, “perpetual patients” or “previvors”, they are forging new identities living under a geneticized gaze (Chilibeck, Lock & Sehdev 2011, p. 1770; Rapp 2014, p. xviii). This gaze considers them as needing medical diagnosis, guidance, and health surveillance, all which required they be seen within a “clinical” environment.

Genetic Counselling Clinic Appointments
Latimer argues that it is the clinic that is the location for the discovery of medical knowledge (2013a, p. 27) and a “new domain that is engrossing medicine, science, social science and the social body” (2013a, p. 18). It was in the clinic where the uneven alignment between the new genetics, medicine, the gene, the individual and the family became further apparent.

Clinic appointments ranged anywhere from forty to ninety minutes in length – which was considered an appropriate length of time to impart enough information to obtain informed consent. There was a common pattern of procedure with slight differences in how they were conducted, due to whether the client was a proband or a presymp, and the clinicians involved. The genetic counsellor arrived approximately ten minutes prior to the appointment, preparing for the clinic by going through the information within the client’s file, which helped them remember what needed to be covered.

When I was present in clinics, the genetic counsellor first asked the client for permission for my attendance, usually in the hallway prior to reaching the door. This occurred regardless of whether I had obtained prior consent from the client and reflected the duty
of care the genetic counsellors placed on the right to confidentiality and decision making. In the majority of the clinics I attended, the client was female and, where the client was a male, they were accompanied by female family members. Later in the chapter, I introduce Michael, a male client who attended with his wife, daughter and his sister who he said had “made him” attend. This reflected the familial nature of the health concerns, where females were considered the matriarch of families; both information holders and information distributors. As such women are considered as “kin-keepers” of the family and more likely to be asked about family medical history (Green et al. 1997, p. 57).

After the genetic counsellor introduced themselves they informed the client of what they expected to cover at the appointment and the amount of time they had. It was explained to the client that they worked in a “double team” or “tag team”, with a clinical geneticist who attended later in the appointment. Sometimes the genetic counsellors told the client to save the difficult or “curly questions” for the doctor. This provided the client with a heads up about the extent of their medical knowledge.

The genetic counsellor asked the client why they were at the first appointment, which was done in order to clarify the events and reason that led to their attendance. This was followed by the client being asked what they hoped to gain from attending. Clients largely expressed that they attended to find out if they carried the gene that had caused cancer in them, or in their family members. They also wanted to know if they

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35 In all of the circumstances, those both planned and unplanned, no client or their family member refused my presence.
were going to get cancer and for some if they were going to get cancer again, including when and where it would most likely occur.

During one clinic appointment that I observed, Lilly (the client) had been undertaking chemotherapy treatment at the time of the clinic after being diagnosed with breast cancer and recommended by her oncologist to attend. On her own at the appointment, Lilly expressed that her decision to attend was based on her wish to have genetic testing. Lilly viewed this as a necessary step to take in order for her to plan the future care of her thirteen year old daughter. With her husband deceased, Lilly wanted to make sure that if she died from her cancer, that her daughter could prepare for her future health and avoid cancer by knowing if she had a mutation. This is an example of how the decision to attend genetic counselling was based on the perception that the information received from attending, and from a genetic test result, would help family members avoid cancer and/or death from cancer in the future. Lilly was like many of the clients of the Unit who attended clinic with the view that the knowledge gained from mutation testing would help them and their family members avoid cancer in the future.

**The Pedigree as a Tool**

As mentioned previously, along with the information provided by clients, medical documents, specialised software like FamilyTrace, and genetic test results, the clinicians used the clinical pedigrees created in the Unit, to map, connect and predict relationships of familial cancer risk between clients and genetically relations. They used the information collected to sort or “triage” the clients into different risk categories (Gibbon 2007, p. 52).
The next part of the clinic appointment involved the genetic counsellor taking out the pedigree and showing the client. In the clinic, the pedigree was a very important physical tool that the clinicians used to confirm and clarify information, using a pen to add in changes and cross out information as the client instructed. Although the majority of the clients looked over pedigrees that had already been printed out on paper, one of the clinical geneticists preferred drawing the pedigree in front of the client. By building the pedigree in the appointment, it was said to slow the process down and allow for the client to take in the information better. All of the clinicians agreed that their clients preferred the pedigree to be “built up”, and/or added to in front of them, saying: “it gives them a story, it makes it fact”. This statement supports the argument that the pedigree enables the visualization of genetic disease by depicting genes as travelling irreversibly in time and through bodies and allowing for future illness to be imagined (Svendsen 2006, p. 147).

When informing the client about which biogenetic relations the risk was suspected to have come through, one clinical geneticist preferred not to use the names on the pedigree, saying it personalised it too much which could result in particular family members being made to feel guilty. On the other hand, the other clinicians preferred to use the names while discussing their client’s risk, viewing it as an important part of the client taking in the information. This demonstrated how there were particular differences in how the clinicians confirmed information in the pedigree and informed their clients of the familial risk. It also illustrated how the clinicians had the power in the relationship to use their own techniques to gather appropriate information.

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36 Outside of the clinic the clinicians talked about the appreciation of the pedigree by client because the pedigrees were viewed as important tailored diagrams. Sometimes the clients asked for copies of the pedigree to be sent to them so they could keep the visually impressive familial information.

37 Sometimes this was undertaken by a clinical geneticist who sometimes independently saw clients for the whole clinic appointment without a genetic counsellor present (e.g. for rural clinics or last-minute appointments).
Educating the Client

After using the pedigree for discussion and clarification, rather than keeping the pedigree on the desk, the clinician placed the pedigree back in their file in order to obtain the client’s full attention. The genetic counsellor then informed the client that they wanted to talk about cancer, explaining the difference between “normal” cancer and familial cancer, and how particular patterns of cancer and types of cancers in families indicated if cancer experiences was familial and due to the inheritance of a genetic mutation.

For the majority of the clinicians it was important to know what their clients already knew about cancer and familial cancer so they could then decide how much, and what, information to provide to the client. Most of the clinicians argued in favour of knowing whether their client believed that the prevalence of cancer in their family was due to certain events, such as chemical sprays used in their farms, Hormone Replacement Therapy (HRT), the experience of great stress and/or family abuse. The connections between certain events as the cause of cancer were commonly expressed by clients in the clinic and reflects how cancer is a predominant disease metaphor (Sontag 1978, pp. 9, 72, 77). When considered as a disease of suppressed emotion or inhibited passion, cancer is said to emerge from the unconscious self as malignant growth (Sontag 1978, p. 27).

It was argued that knowing the client’s knowledge about cancer and of genetics, along with any religious or spiritual beliefs, meant they could be acknowledged and that the language they used and the information they provided could be appropriately tailored. Clients wanted answers as to why they had experienced cancer; asking the clinician whether it was due to the HRT they took, an accident, or stress. The clinicians largely informed their clients that their experience of familial cancer was due to reasons beyond
how they (the client) behaved. As explained later, if clients had genetic testing and mutations were subsequently found, it provided a biomedical/genetic explanation for the experiences of cancer. On the other hand, another clinician adamantly argued that they (as clinicians) did not need to know the client’s opinion on their causes of cancer, because the clients were there to learn, saying: “I don’t need to know what they know or believe, for the purpose of efficiency and because they have come to hear me”. This again demonstrated the differences in the opinions held, and the genetic counselling styles practised by the clinicians, and the power of the clinicians to decide what was important and covered in clinic.

The content and extent of information provided to the client in the clinic largely followed the recommended NHMRC (1999) guidelines, varying depending on the genetic counsellor, the context and the medical history of the client and family. The type of information the client received also depended on the client’s risk status and the perceived capabilities of the client. Generally, the genetic counsellor informed their client if they were considered at increased risk of familial cancer (when compared to the normal population) and, if the client had children, their children’s risk was also discussed (if the children were biogenetically related). If the client had already suffered from cancer, then they were provided with an estimation of the chance of a familial cancer re-occurring or occurring elsewhere in their body. In the appointments that I attended, clients did not ask how their risk was calculated. The genetic counsellors once explained how it was very rare for their clients to ask how their risk was calculated. In the times it had occurred, because of the difficulty involved in explaining the system to a layperson the genetic

38 In contradiction however, clients considered at high risk of familial cancer were encouraged to exercise and eat healthy with healthy recipes provided in newsletters/handouts sent.
counsellor was not able to answer, and so the clinical geneticist who came into the appointment half way through answered the questions.

After the client was informed of their risk, they were then provided with information about their medical surveillance and cancer risk reducing or preventative options. As explained in Chapter 4, these included early detection, reduction or prevention strategies and procedures relevant to their risk and case. The greater the client’s predicted risk resulted in the recommendation of earlier, different and more intensive screening or procedures. The provision of such recommendations about the type and timing of screening and procedures also demonstrates how genetic risk encompasses personal responsibility over one’s health decisions (Novas & Rose 2000, p. 501). It is argued that the surveillance and risk reduction procedures undertaken by those identified at increased risk have no efficacy in reducing cancer incidence or death (Patenaude 2005, pp. 25, 252). This argument, of the unproven benefits of such surveillance, was not explained to clients of the Unit because the clinicians believed such measures did result in benefits to the client and the community. There are also other arguments supporting these benefits, which demonstrate the diversity of opinions and research on the subject.

**Content and Extent of Information Provision During Clinic**

Clients were considered as individuals in need of particular information, information of potentialities and hope. The need to inform clients reflects the contemporary “Euro-American” idea of personhood, that science provides people with better knowledge and understanding of their bodies which will help them make decisions and choices, and be autonomous (Strathern 2009, pp. 152, 155, 156). It also reflects the neoliberal idea where clients are viewed as autonomous and rational consumers (Lupton 1999, p. 10).
All of the staff expected that their clients came to the Unit to be informed, however it was said that client’s desire to be informed varied (discussed below).

The very process of information provision is not value neutral because it also provides particular perspectives. Practitioners and policy makers may constitute clinical knowledge practices of prediction and diagnosis in genetics as simply “revealing the facts of the matter”, however, in so doing they reproduce medical theories that constitute the medical gaze as simply detecting and describing “what is” (Latimer 2013a, p. 183).

As mentioned earlier, the content and extent of information provision provided to the client followed recommend NHMRC (1999) guidelines but it was also dependent upon on the clinicians. This decision was based on the extent of knowledge the client appeared to have around familial cancer, their “burdens” (issues they were dealing with) and capacity to understand and handle concepts. Overall it was stressed by the clinicians to their clients in clinic (and in written material) that the clinician’s role was to provide non-directive and non-judgemental assistance so the client could make the best decision for themselves. This neoliberal idea demonstrated how clients were reminded that it was ultimately their responsibility to make decisions surrounding their health and the health of their children. There were however tensions evident with this concept and practice of attributing responsibility, with an example discussed later in regards to a client’s decision to have a genetic test.

**Informing Extents**

Although emphasis was placed on the benefits of knowing one’s familial cancer risk and acting accordingly, ultimately the most important messages the clinicians were required
to pass their clients (in order for informed consent to be considered as achieved), were to do with uncertainties – of not being able to know. These uncertainties surrounded the prediction of familial cancer risk, genetic test interpretation, ownership, privacy and implications of familial cancer risk information. Apart from the consent form that all clients had to sign prior to clinic, in clinic the uncertainties, largely framed as “considerations”, were explained in various ways and to various extents. When clients expressed confusion after having been informed of certain concepts or processes, the clinicians gained professional satisfaction from educating the client about what they, as part of the scientific and medical community, did not know. A genetic counsellor once said that she “loves that she doesn’t have to give answers”. This supports my argument that genetic counselling, in the context of familial cancer, was about information provision but not necessarily about providing answers.39

Previously it was explained that the clinicians displayed preconceived ideas about their clients’ level of genetic knowledge. Clients who were seemingly more informed and/or intelligent, beyond the clinician’s expectations, sometimes referred to “switched on”, were said to have made their role easier. The clinicians were more confident that these clients either were already aware of familial cancer, or appeared to have the mental and emotional capacity to understand the ambiguities. When the clinicians did not have to inform their clients about “the basics” and the benefits of knowing their risk, this meant they had more time to cover other aspects. This belief resulted in the clinicians discussing with those clients, not all they knew, but some of the things they did not know in regards to the uncertainties surrounding the concepts of estimating familial cancer risk and

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39 As a way of negotiating and coping with the impacts that such institutionalised unknowns had, the clinicians strived for more knowledge and scientific progress in the area (as discussed in Chapter 8).
mutation interpretation. Interestingly, the goal of providing the client with the uncertainties was to leave the client more informed and empowered by the information to act appropriately on their risk – which was a contradictory notion and practice.

For some clients, the right to know may surpass their ability to know because no matter how skilled the clinicians, a proportion of the population may not conceptually understand the messages being provided (McBride et al. 2010, pp. 91, 95). Although the clients I interviewed post-clinic appreciated information about the current limitations, the information caused confusion. One client commented to me: “the more we know the less we know”. This popular quote acknowledged that the growth of knowledge entailed the growth of ignorance. Such a response to information was labelled by nineteenth-century philosopher James Ferrier as “agnotology”, the cultural production of ignorance where more knowledge of a subject leaves one more uncertain than before (1854).

On the other hand, clients who apparently displayed little understanding about genetics and/or familial cancer, or those seemingly less educated and not understanding the information in the way intended were sometimes referred to as “slow” or “not all there”. As a result, the clinicians used their clinical judgement and power, selecting what information they provided and what information they left out, with more emphasis provided on what was known, and less emphasis was placed on what was not. In the Unit it was common to hear the clinicians debriefing after clinic and expressing surprise over the lack of knowledge that their clients held around genetics. The surprise was in relation to a question asked by a client during clinic, or when the client asked for more information regarding certain statements made by them. Some examples shared included: clients being confused after being informed that a genetic test on them was also a genetic
test on their family members, or that they were made up of genetic material, half of which came from their father and half from their mother. The clinicians argued that such knowledge should have been learnt in the early years of high school.\footnote{The disbelief of known genetic knowledge also extended to visiting medical students. One visiting medical student asked the clinical geneticists about the number of chromosomes in humans. The clinical geneticist expressed disbelief believing it was common knowledge to know that 21 chromosomes existed.} This resulted in the clinicians having to “go back to basics” when informing clients about genes, cancer and familial cancer risk.

Below is an extract of an interview with May, one of the genetic counsellors. It illustrates how a belief in not adding more information, viewed as “burdens” to their clients was important to consider.

They (the clients) are coming to you for a specialist consultation in a way rather than seeking someone to help manage an issue...you certainly have to be aware of thinking about and exploring not just what they (clients) are giving you at the appointment but what the whole situation has been like. How they have coped at the time, if you are adding more burdens to them and all those sorts of things.

This highlights how there was an apparent contrast and contradiction in information provision. Clients considered as being less informed were provided with basic information but greater certainty regarding familial cancer risk, testing and interpretation, when compared to the clients considered “switched on”, who in contrast, received more ambiguous information. This illustrated the “agnotology” surrounding familial cancer.

Being Informed of the Decision to Test for a Genetic Mutation
The majority of the Unit’s clients were said to have attended genetic counselling with the desire to have genetic testing due to the perceived benefits of “knowing”. Research by Marteau and Croyle (1998, p. 694) found that people seek genetic testing because they...
wish to decrease uncertainty and so they want to know with certainty if they are likely to
develop a particular illness. Clients of the Unit had a sense that clinicians could provide
certainty; however, certainty was not something the clinicians or the laboratory could
provide. This demonstrated a knowledge gap that existed between the clients and
clinicians. The clinicians had the power to decide whether clients were eligible for genetic
testing, illustrating how they were gatekeepers to genetic testing. However, the clients
offered genetic testing still had to decide whether to have the genetic test.

During clinic appointments, the clinicians explained to their clients whether or not genetic
testing was appropriate for them. Their explanation to the client centred around whether it
was thought the test would be useful or not to the individual’s circumstances. When the
clinicians believed that they would find a familial cancer mutation in a client, they offered
DNA testing (through blood samples) to possibly identify the underlying genetic
mutation. Where clients were told that they would not be offered genetic testing, it was
very common for the clients to ask why they had viewed individuals with similar stories
on the television or online, who had genetic testing. In these cases, the client was
informed that often media portrayals were incorrect and the clinicians emphasised and
explained how genetic testing was not useful for them. For the sake of mutation
identification, clinical judgement was adapted to fit the situation, demonstrating the
power that the clinicians had in deciding what was known and what was passed on to
whom. This demonstrated the clear power practice where power was exercised through
clinical judgement (because of a possible genetically viable result) and not an emotional
caring decision (because the client wanted one).
Clients were informed that when genetic testing was undertaken the majority of times the results were negative or inconclusive and when they were positive, it did not mean that the client would go on to suffer from cancer (see next chapter about test results). With the odds against the finding of a mutation the clinicians were selective with offering genetic testing, favouring clinical and medical discretion over the psychological need of some clients. Some clients expressed confusion and disappointment when informed of this knowledge, because they believed that a positive genetic mutation test would provide confirmation and/or certainty about an increased risk of familial cancer.

**Translating the Uncertainties and Implications of the Genetic Test**

Where clients were offered genetic testing, some of the implications of having a genetic test were provided for the clients to consider. These included: the ethical consideration that a test on them was a test on the family; possible insurance implications; the technical limitations of the genetic test; and its interpretation at the present and future time. The clinicians were powerful in predicting, testing and justifying choices as discussed below.

The clinicians largely varied in how they explained the ability to detect whether cancer experiences was due to an inherited familial cancer mutation or “by chance”. One genetic counsellor used diagrams to aid in the translation, while one of the clinical geneticists used the analogy of a genetic recipe book and the Da Vinci code (a popular novel). This explanation described how there were about 20,000 recipes that made up a person (spelt out with four letters) and if one ingredient was wrong when their cells made copies, but the rest of the ingredients were ok, than the recipe may still turn out. It was explained to the client that the difficulty for the clinicians was in not knowing the effect or extent of
having the wrong ingredient. It was further explained, that “spelling mistakes” and errors in the genetic code were looked for when genetic testing was undertaken. However, it was said to be like looking for spelling mistakes in a “deluxe edition of the Da Vinci code”, but they were not allowed to look at the beginning of the book, as they are not good at looking at the beginning. They also would not know if the version they were checking was the English or American version and so it was difficult to know when a spelling error or a variation did not matter. Emphasis was then placed on how there was a chance that the laboratory would find mistakes but that they, as clinicians, had difficulty in interpreting if, and what, effect these findings could have (if they could result in cancer, or not). This specific explanation demonstrated how a mix of scientific and lay knowledge, through the use of popular analogies of recipes, novels and spelling mistakes, were provided to the client to try to help them understand the difficulty in identifying their familial cancer risk through genetic testing and what it could mean if a mutation was found, or not.

**Clinician’s Discretion and Power**

In accordance with their guidelines, clients considered to be at moderate or high risk were generally offered genetic testing. There were some exceptions to this, involving both the testing of clients who did not meet their guidelines and not testing clients who did meet the requirements. In one instance a client considered at standard risk, who did not meet guidelines for genetic testing, was offered testing because they had Jewish ethnicity and had screening for the common Jewish BRCA mutations.

Another exception, which saw moderate and high risk clients not offered genetic testing when normally they would have been, occurred when the clinicians did not think a
mutation would be identified. In each of these cases, risk scores clinical discretion and judgement superseded guidelines, however, as mentioned earlier, this was a legitimate practice, acknowledged and recommended by the NHMRC (1999, p. viii). These occurrences demonstrated the power of the clinicians and the ongoing negotiation needed when working in the field of familial cancer. It also demonstrated an argument made by Latimer (2013a, p. 201), which considers the genetics clinic as the centre of both calculation and discretion, because even where there are tests, clinical judgements are performed deciding whether or not the test result is accurate or significant. By maintaining “uncertainty” it legitimates the need for more “genetic technoscience” and more clinical judgement in the future to help fix diagnostic possibilities (Latimer 2013a, p. 201).

Earlier, I described that there was a tension present for clients receiving non-directive and non-judgemental counselling, or information provision. When a client expressed certain strong opinions either for or against genetic testing, in order to provide the client with all the tools necessary to make an informed decision, the clinicians played a role once described as: “the devil’s advocate”. For clients considered at high risk of familial cancer and eligible for a genetic test, emphasis was placed on the benefits of identifying and knowing if a known mutation existed. In another case, when a client was not offered genetic testing but expressed the need for genetic testing to confirm her future risk of breast cancer, the clinician provided risk figures that emphasised her chances of not developing breast cancer. This demonstrated how the clinicians were gatekeepers to information and genetic testing, deciding who was offered genetic testing, before the client could decide whether to have a genetic test and how to proceed with the information received (see Risk, Responsibility and Surveillance further in the chapter).
“Get the Doctor”
The majority of clinics involved both a genetic counsellor and a clinical geneticist for the second half of the appointment. When the genetic counsellor felt they had covered everything with the client (towards the end of the appointment) they informed the client that they would “get the doctor”. They then left the clinic room and briefed a clinical geneticist waiting in a nearby room. During the quick brief (no more than five minutes), the clinical geneticist was reminded about the client’s referral, risk, what had already been covered in the appointment, and any other relevant information. Both clinicians then returned to the client together.

In the clinic appointments that I attended, although the clients had been informed prior that the clinical geneticist would attend, some of the clients initially seemed confused as to their role. This is evident in an extract below, taken from a clinic appointment with Mary (the client) and Kym (her sister):

Clinical geneticist: Hi, my name is (first and last name) and I am one of the clinical geneticists with the team, and I am one of the doctors (shakes hands before sitting facing the sisters)
Mary: ok
Kym: So you are checking for spelling mistakes?
Clinical geneticist: No the laboratory person does that
Kym: You just read their reports?
Mary: Tell them what to look for?
Clinical geneticist: I read their reports and I am involved in the interpretation of their reports, but I work with people rather than work with specimens in the lab.

The exchange above demonstrated how two sisters asked questions in an attempt to understand the role of the clinical geneticist/doctor, requiring the clinical geneticist to further explain their role. Kym, the sister, used the “spelling mistakes” analogy that she had been informed of only moments earlier by the genetic counsellor, which demonstrated how she took on the language and information provided in that way.
In the next part of the clinic appointment, the clinical geneticist substantiated and reiterated the information that the genetic counsellor had provided, including explanations about cancer and familial cancer, their risk assessment, whether genetic testing was “useful” for them and the options available for them to manage their risk. The overlap of information ensured that correct medical advice had been provided, and with the information explained in a different way, with different examples and analogies provided, it was considered as resulting in better understanding by the client. It was also an opportunity for the client to ask medical questions, or difficult questions that the genetic counsellors did not feel comfortable answering, or did not have the ability to answer. Clients considered at high risk of familial cancer were then encouraged, by both clinicians, to provide contact details for certain biogenetic relations. If the client agreed, then the formal risk notification procedure was undertaken – a procedure set up to facilitate the sharing of the information with “family members” considered at risk (see Chapter 7 for details and discussion).

On the one hand, the teamwork process in clinic reflected the practice of genetic counselling which consisted of the genetic counsellors and the clinical geneticists working in close partnership as a team. On the other hand, it highlighted a hierarchy amongst staff. Although they largely worked as a team, the clinical geneticists were the only ones also able to provide genetic counselling to proband clients on their own. Proband clients were considered as requiring a doctor to determine if they had been informed enough to provide consent, and reflected the greater authority, knowledge and responsibility that the clinical geneticists, as doctors, were bestowed with.
Clinic Appointments for “Presymps”: Almost Having the Tiger by the Tail

For presymp clients, those that attended clinic after being risk notified, there was some difference to the information they received during their clinic appointment. Although I was only able to attend a couple of presymp appointments, the appointment described below was typical of the process and contains different accounts of information. The presymp clinic occurring in a rural hospital involved some members of the Bruan family.41 Attendees included Michael (who was in his sixties and the primary client at the time of the appointment), Lisa (Michael’s sister), Sofia (Michael’s wife) and Kate (the daughter of Michael and Sofia). After the clinician asked why Michael had attended the appointment, Michael answered that he had been “told to come” to the appointment by his family. His sister Lisa (in her fifties) was also a client of the Unit and had been identified with a BRCA1 mutation (a mutation in a gene linked to an increased risk of breast and ovarian cancer). Lisa had been tested as a result of being diagnosed with breast cancer twelve months earlier. In order to minimise her risk of further cancer, Lisa had since had a double mastectomy (the removal of both her breasts) and although a hysterectomy had been planned for the following month she had re-scheduled it to the following year because she was not ready.

When the clinician asked each family member why they were there, all of them responded similarly emphasising the importance of finding out the information, not for themselves but for their children and/or grandchildren. This was a common reason given by many clients of the Unit for seeking information. In this appointment, Lisa pointed out to her relatives that it was important for them to know so they could look after themselves

41 Being a rural location only one clinician saw clients, unlike in the city locations where both a genetic counsellor and clinical geneticist were involved. In the rural setting, this was changed in order to maximise the number of clients seen.
in order to be “around” for their children and grandchildren. It displayed how Lisa believed in the direct correlation between knowing this information and living. Michael also expressed that they attended the appointment to find out if they, or their children, carried the same mutation as Lisa; why their family had the mutation; which parent the mutation had come from; what their cancer risk was; which people and biological material could be affected; if there was a cure; and if they had the mutation and there was no cure, what they could do about it.

The clinician explained that the Unit’s role was to “help families use genetic information in the way that the family wanted to”. It was then declared:

In talking about things today I don’t have anything you must do X, Y, Z, but there may be you could do X, Y, Z, which are options that you can then explore with family members and doctors, whatever you may want.

This quote demonstrated how the clinician was direct from the beginning, telling the family that he was not going to tell them what to do – but rather provide them with information about the choices they had and that any decisions were then up to them. This non-directive form of genetic counselling, involving informing and guiding, was used because it is a guiding principle of risk communication in genetic counselling (O’Doherty 2005, p. 7). Its use illustrates how certainty sought by clients was rendered uncertain.

As the appointment went on, a marked difference became apparent between what was wanted and expected by the family and what could be provided. During the 90 minute appointment the clinician provided a mix of facts and uncertainties about cancer and familial cancer (in particular relating to the BRCA1 mutation identified in Lisa) to the
family. The family’s reaction to being informed about the many uncertainties was that of frustration and confusion.

The first frustration the client expressed related to the family’s need, or not, to have a genetic test to see if they were also carriers of the mutation. With the family’s “pedigree” laid out in front, the clinician informed the family that the experiences of cancer were likely to be familial, which was evident even before having a genetic test simply by looking at their family history: a family history which included a pattern of early onset cancers and multiple cancers. The clinician stated: “I don’t need any fancy genetic test to do that, I can do that with the mark one eyeball and the pedigree that we have”. This statement demonstrated how the clinician did not need a genetic test to identify their risk, that it was apparent from using their clinical knowledge and judgement regarding the family history. However, in contradiction to this statement, Michael and his children were then offered the option of genetic testing (for the BRCA1 mutation known to be in the family). The clinician informed them that instead of talking about it in “an abstract sense”, genetic testing would take it to the next step – of them finding out if they had the mutation.

The clinician went on to emphasise how there were essentially two sides to finding out this information: “a good news, bad news story”. It was explained how knowing that one has a mutation could be viewed as a threat because the person who has it “is certainly at increased risk of cancer”. The clinician then emphasised how it was “also an opportunity and knowing this information can be very helpful in reducing the impact of cancer on your family”. The clinician went on to describe how Michael and his children each had a fifty-fifty chance of having inherited the abnormal BRCA1 mutation gene, the possible
ovarian and breast cancer implications associated with it, and the choice of following a
course of actions that could be undertaken to detect cancer and/or avoid earlier (through
undertaking regular surveillance or risk reduction surgery).

After having previously wanted genetic testing for family, Lisa said that even without
having the genetic test, they knew they were at risk and they knew what they could do
about it, emphasising to Michael that they (including his children) could all undertake the
suggested measures and consider undertaking risk reducing surgery to remove their at-
risk body parts (even without having the genetic test). Michael, confused, questioned Lisa
as to why she would say that after she was the one who told him to come and have the
mutation test and stressed the importance of knowing their risk. The clinician asked Lisa
why she did not recommend the mutation test after having it, and previously
recommending it, and Lisa explained how she regretted having the test done, saying:

I’ve got it (the mutation) now I know. I’ve advised the others not to have it done at the
moment cause it is a scary thing knowing that you’ve got it. And it’s like will I be here
tomorrow, things like that, and that’s what is very scary for me and I wouldn’t want them
to go through it. Like my kids, I’ve advised them not to have it done cause it is a scary
thing cause you go to sleep at night-time and you think of something and think am I going
to be here next week, will they find something somewhere else, yeah cause one thing will
lead to another, that’s how I look at it. Cause I’ve had breast cancer…. the blood test I
wouldn’t recommend it cause I think, I’ve got it we must have it in the family
somewhere, unless my mother played around and I doubt that very much. So I wouldn’t
recommend it to my brothers and sisters but I wouldn’t even my children….they are still
young and life insurance that’s unknown cause if they don’t know they can get life
insurance for all these things but not once you’ve declared it.

In the above extract, Lisa conveys how the new knowledge she had been provided with
(that she had a BRCA1 mutation) brought with it greater unknowns. Instead of assisting
her to make her feel more secure about her future, she became more aware of the
uncertainty and the unknowns about what a mutation meant for her future. After Lisa had
said this, Michael, confused by his sister’s opinion, responded by stating: “Well if I don’t
get it done how are my kids going to know?” Lisa explained that they just needed to inform their doctors of the family history of BRCA1 so they were aware of it. The clinician then responded that although he did not disagree with what Lisa was saying, he asked if he could be excused while he “just argued the opposite”, before stating:

This is where you almost have the tiger by the tail, the tiger’s not absolutely controlled. I cannot cure this particular BRCA1 mutation but I now know where the tiger is, he’s at the end of that tail and it means that we’ve got, we can provide much more targeted and useful information to members of your family.

Using Kate as an example, the clinician went on to say that it was one thing for Kate’s doctor to know that there was a BRCA1 mutation in the family but it was very different if the doctor knew that Kate had inherited the mutation. It was explained that without the mutation confirmation the doctor could not work with Kate to make the difficult decisions in deciding what they could do about her risks, before saying that the doctor could not justify surgery without knowing if she had the mutation. Without knowing Kate’s status, without knowing if surgery was really necessary, the clinician stressed that it would really be an “unjustifiable risk” to do any surgery. The clinician further advised the family of the danger in compromising their health for the sake of not getting life insurance.

This scenario was an example of how the clinician picked up on Lisa’s regret about knowing of her mutation and the possible influence her experience might have on her relatives. The clinician felt the need to play what was termed as “devil’s advocate” where they argued the opposite. This displayed how the clinicians did not let their clients make their own decisions without first providing them with the appropriate or correct decision to consider. Concerned that Lisa and the others were not fully aware of all of the information, the clinician placed emphasis on the importance of the family knowing if they had the mutation by relating it to the possible future care they could and could not
receive, which depended on their decision. The clinician used both abstract and colourful imagery (tiger by the tail) and concrete examples (involving Kate needing to justify surgery) to convey the argument. This example demonstrated how some clients, who were given the opportunity of mutation testing for a familial cancer mutation, were made to feel accountable and responsible for having a genetic test due to the emphasis placed on the possibility of greater care and prevention (justifiable) and the lack of care and prevention available to those who rejected the option to know (unjustifiable). The clinician assumed that surgery would be taken up and that the mutation confirmation would be enough justification required in order to undertake the procedure. The clinician did not inform the family that clients considered at high risk of familial cancer were able to have appropriate surgery to minimise their risk without mutation confirmation, saying in this appointment that such surgery would be an “unjustifiable risk”. The need to find out one’s mutation status and have surgery reflected the promise of predictive health, which brought about an obsession with the “taming of chance” (Gibbon 2007, p. 6).

After the clinician had “argued the opposite”, the family were then informed that there “was no rush to do any testing” and for Michael to have the genetic test when he was ready. Ultimately, it was stressed that they needed to be comfortable with their decisions, how the information was “both a threat and an opportunity” and how an informed decision was important. However, the notion that one can be informed when talking about risks could be considered as contradictory to the underlying meaning of the word risk.

Michael, confused by the choices and swaying advice of the clinician, used a forceful and tense tone directed at the clinician saying: “you say one thing then say another”. Michael highlighted and questioned the non-directive counselling style, and, specifically,
the inability to receive the answers to the questions Michael came to the appointment wanting. He had expectations that could not be met by the clinician, who shared the varying arguments, which reflected the realistic nature of the unknowns, and the uncertainties that existed around familial cancer.

The second frustration/confusion revolved around the inability of the clinician to provide the family with answers regarding their risks of developing cancer, the other types of cancers they were at risk of, when cancer could occur and who it might happen to in the family. Regardless of how the clinician answered the questions – which included using a mix of detailed, short and sharp, scientific, lay, abstract or honest responses, e.g. “we don’t know why” or “there are no black and white answers”, – the confusion around the uncertainties remained for the Bruan family. Michael, using the information he had gained from the appointment, asked the clinician:

If your body is made up of one good one rusty, fifty-fifty, how much do I have to cut away to get rid of that bloody fifty bad half your body?

The clinician explained that it depended on where the cancer risk was (what particular organs it had more chance of affecting) and what age the clients were. Wanting more specific responses, Michael became increasingly agitated as the appointment went on, interrupting with a loud voice whenever he heard something he did not agree with or understand. The display of these emotions demonstrated how clients displayed agency, through interrupting and questioning what the clinician was saying and why what they were being told did not make sense to them.

These reactions provide examples of possible misunderstandings that occur in genetic counselling. The examples highlighted the differences of opinions held, the value of
knowing, and the frustrations in not being able to know. Apart from the clinician informing the family that a positive mutation result provided justifiable evidence to have surgery, they did not provide any information with certainty, and this reflected the unknowns and uncertainties in the area of familial cancer. This impacted on Michael’s ability to comprehend, make decisions for himself and his family members, and it resulted in the family questioning the value of the knowledge that the clinician provided.

Being at risk
Prior to entering and once in the clinic, clients provided the service with access to their own and their familial information. In return, the clinicians provide their clients with information either that they were at standard risk or that they had increased future risk of familial cancer, a risk that could be reduced. Such an exchange, involving the providing and receiving of information, has been described by Latimer as an exchange of “gifts” and demonstrates how families become “allies of the clinic” (Latimer 2013a, pp. 179–184), and included in the process of information provision.42

Mainstream approaches in medicine treat risk as an objective, quantifiable entity, however the concept of risk is socially and historically constituted because genetic counselling relies on the sociocultural concept of risk, a mathematical expression of probabilities to forecast the future based on past occurrences (Finkler 2000, p. 7; Lupton 1999, pp. 1–11; Petersen & Lupton 1996, p. xii). Research is being conducted on the social, cultural, psychological, and ethical aspects of health risks, illness prevention and the impact of knowing one is at risk because precisely what it is that is known, or can be known, is open to question. A number of studies have also highlighted the varying and

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42 The notion and description of reproductive gift exchange in biomedicine was discussed by Konrad (2005a), in her work on ova donors and recipients in Britain.
ambiguous meanings involved in the communication of risk, how risk information may be presented as relative or absolute, in percentages, as probabilities, or in descriptive terms (Marteau & Croyle 1998, p. 695) and how the communication of can risk impact on individuals’ level of “perceived risk” (Marteau 1999, pp. 417–418; O’Doherty 2005, p. 20). Research from cognitive psychology has described how people often do not understand what the genetic test results mean in terms of their risk (Cameron, Sherman, Marteau & Brown 2009, p. 307; Hanoch, Miron-Shatz, Rolison & Ozanne 2014, pp. 1142–1143). In the previous example, Michael demonstrated a misunderstanding of the information he had received, evident in him asking how much of his body had to be “cut away” to remove the “bloody fifty bad half of the body”.

For clients who have been informed that they are at an increased risk of familial cancer, or for individuals who consider themselves as being at increased risk, they may view themselves as sick. This is because there is an entrenched assumption that the term “risk” constitutes a threat (Malaby 2002, p. 286). All people, not just those with a familial cancer risk, can be termed “perpetual patients”, “hypochondriacs” or “pre-symptomatic persons”, stigmatized and considered sick with conditions they do not have – and may never have which can lead to a fear of the future (Finkler 2000, pp. 7, 183, 197; Finkler 2001, p. 241; Hubbard & Wald 1999, p. 38; Konrad 2005b, pp. 1–6). In Kirmayer’s work on the hidden values of biomedicine, he distinguished between real disease and imaginary disease (1988, p. 69). In biomedicine, it was argued that a person is only considered to be “really” sick when a biological process takes hold. However, he also contends that the “potential” for disease sustains the interests of physicians and so the potential patient receives evaluation and care as if they needed attention (1988, p. 75). Considering this argument, for clients who were informed of an increased risk of familial cancer and for
individuals who were risk notified, they could be considered as having a “real disease”. This is because by becoming an object of medical study and attention, they were provided with an intrinsic value associated with having a real life-threatening diagnosis.43

Risk, Responsibility and Surveillance

Those considered at risk are described in disease terms but they are not sick with cancer, and may not be sick for years, or perhaps ever (Jonsen 1996, p. 8). Potentiality, as the partner to risk, is complex because it requires imagination and talk about what does not, and may never, exist (Taussig, Hoeyer & Helmreich 2013, p. 4). It is argued that familial diseases provoke the “genetically risky individual” to be genetically responsible and cautious (Novas & Rose 2013, p. 487). Once diagnosed with being at risk of developing diseases or at risk of “susceptibilities”, the responsible asymptomatic individual is enrolled for the rest of their life in the world of medicine, taking tests, drugs, or self-examination, and self-defining themselves as a “prepatient” suffering from “protosickness” (Rose 2007, p. 94).

Being at risk can shift human fate from a number of diverse etiological theories of disease to the notion of a mathematical probability or statistic, however Boia (2004, p. 106) illustrates that although statistics appear “objective” they depend on the ideologies they wish to demonstrate. What is more, being at risk of familial cancer places the social responsibility of prevention on the client and their family, leading them to believe that by taking precautionary measures (like prophylactic mastectomy or family planning measures), they can control the outcome and avoid familial cancer (Beck, Giddens &

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43 This is not limited to clients with a familial cancer risk, with risk factors also extending to the monitoring of individuals prone to other illnesses such as high cholesterol or diabetes.
Lash 1994, p. 3; Finkler 2000, p. 8; Lupton 1999, p. 4; Petersen & Lupton 1996, pp. ix, xiii; Petersen 1997, pp. 192–202). Gifford (1986, p. 216) wrote how the term “risk” “has not always embodied ideas about chance”. In the Unit however, individual and familial responsibility to avoid familial cancer was emphasised by clinicians in clinic and in written material as an “opportunity”.

At the beginning of the chapter, I introduced that the overall aim of genetic counselling was to affect the client’s health behaviours (Rose 2007, p. 113). On a local level, the need to affect the client’s health behaviours was evident in the reasons behind the establishment and continual funding of the service. The service aimed for their clients to take action and reduce their risk by undertaking “surveillance strategies” – which they also believed led to medical savings. Apart from informing their clients in clinic about their options, clients and their referring doctor also received letters post clinic. The letter summarized what had been discussed with the client about their familial cancer risk and it advised and recommended the appropriate measures recommended to the client and their genetic relatives in regards to their risk. The letter’s purpose was to provide accurate detailed written information that the client and the doctor could refer to, a way of informing their client of any important information that their client might have forgotten and, most importantly, it was a way of organising adherence to surveillance strategies. In addition to the letter, clients also received a standardized detailed (A4 paged) brochure explaining the risk and considerations which included cited research, graphs and figures which substantiated their advice.

All clients considered at high risk of familial cancer also became part of a “scheduled surveillance” program managed through FamilyTrace. The service reminded clients, and
their doctors, when a surveillance check-up or procedure was due and what type (e.g. mammography). The outcome of the check-up or procedure was provided back to the Unit and recorded, and where a diagnosis of cancer was made, cancer risk was re-evaluated for other biogenetic relations. The surveillance service was practised in order to remind and encourage clients to follow the surveillance strategies with the help of their doctors. This observation is an example of Foucault’s arguments when he discussed the place of medicine in the monitoring and administration of surveillance of populations and their bodies (Foucault 1980, p. 155). Although disciplinary, the form of power in the Unit was not openly coercive, rather it took a moral approach. With clients organising the booking this meant that power was exercised most effectively as the high risk client (the subject of the power) “interiorizes” the gaze, so that they exercised the surveillance over, and against, themselves (Foucault 1980, p. 155; Lupton 1997, p. 99).

The decision to co-ordinate medical surveillance reflected the view in the Unit that the clients at risk of familial cancer were prone to disease and therefore in need of constant monitoring. This surveillance program had individual and societal benefits – which was integral to the practice of familial cancer risk identification because it was considered as helping prevent their clients from getting cancer and/or the severity of cancer and becoming a health burden on society – as members of the public would otherwise have to pay the taxes for treatment and care associated with the diagnosis of cancer. I observed this concept of financial benefits being disclosed by the clinicians to some clients, demonstrating how it was sometimes used to encourage the client to undertake certain measures to reduce their familial cancer risk.
Uncertainty Surrounding the Diagnosis of Familial Cancer Risk in Clinic

The majority of the clients of the Unit came with expectations (both acknowledged and unacknowledged) that the clinicians could provide them with black and white answers as to whether they had a familial cancer mutation and what it meant for them or their family. These expectations reflected the wider held socio-cultural assumption that the area of genetics is far more promising and advanced than the reality. It also reflected one of the contradictory views held by the clinicians as demonstrated in them arguing that if clients came they would find out about their risk and if they did not, then they were missing out on an important “golden opportunity” (see page 238).

The clients and the general public hold great hope that genetic technology will identify a gene and minimise health risks. In regards to familial genetic testing, the notion that genetic technology can provide answers is promoted through various forms of popular media stories across print, the Internet and television. Via the Internet, an array of private services are available to decode part of a person’s genome and provide information about ancestry and health risks (Palsson 2012, pp. 190, 193). On Australian public television during 2009, three stories about familial cancer risk had broadcast titles of: “Breaking the Curse” (Thomson 2009); “The Killer in You” (Rice 2009) and “In Your Genes” (SBS 2009). The increase in referrals resulting from the media exposure of Angelina Jolie’s experiences, known as the “Angelina effect”, has captured people’s attention and made many people aware of the potential risk of familial breast cancer and the possibility of managing it through prophylactic surgery, leading to increases of referrals and attendance across genetic clinics (Borzekowski et al. 2013, p. 520; Evans et al. 2014, pp. 1–6). These public discussions of familial cancer demonstrate ways in which the media describes, reflects and performs particular beliefs about genomic advances.
The role of clinicians, it has been argued, needs to involve managing the client’s risk, helping them cope with uncertainty about health and disease and to be competent at reducing uncertainty and comfortable in addressing it (Wellbery 2010, p. 1666). To different extents, clients were informed of, and came to learn, that the clinicians were unable, as a consequence of the knowledge and technology they had at their disposal, to provide them with definitive answers. Upon learning about some of the unknowns and uncertainties inherent in the area of familial cancer, some clients expressed surprise and/or disappointment (to different extents) as the reality diminished their expectations and the hope they had invested in knowing.

Generally the topics discussed in the clinic appointments were: the purpose of genetic counselling; familial versus “normal” cancer; the client’s and their children’s risk; the usefulness of having a genetic test; and how the clients could manage, prevent and/or reduce their risk of cancer. These eligibilities and explanations differed depending both on which clinician was conducting the appointment, on the client’s history, their knowledge and displayed reactions to the information. Regardless of the client’s risk category and genetic testing, clients of the Unit were ultimately unable to know what would result from being informed of an increased risk of familial cancer. The clients were not uniformly all made aware of the uncertainties, possibilities and implications, which meant that all clients left the clinic, still not knowing, to different extents, about their familial cancer risk.

For all clients of the Unit, uncertainty remained regardless of whether they had genetic testing or not, and regardless of the outcome of that testing. Clients of the Unit fitted into one of four possible categories of being unable to know. The first category included those
who were not offered genetic testing yet were still informed of their risk category during their first (and sometimes only) clinic appointment. The remaining three categories of being unable to know involved clients who were offered genetic testing and provided with a genetic test result.44

Unable to Know – Denied the Option of Genetic Testing and Assigned to a Risk Category

As mentioned previously, clinicians offered genetic testing to clients who met their defined criteria and/or in whom the clinicians believed a mutation would be found. When clients were denied the option of genetic testing, it left the clients unable to know if they carried a known familial cancer mutation. When reasons were provided for not offering a mutation test, some clients verbally expressed their disappointment at not being able to have a test and some questioned the decision. As stated earlier, clients often brought up examples of what they had seen on the television of similar individual or family stories where they had been tested. Clients used these examples to question why they could not access the same services, why they were any different.

In an attempt to demonstrate their need for having genetic testing, and based on the certainty they thought it would bring, some clients asked if they could pay for testing themselves. This idea was mostly rejected by the clinicians saying they would not order the genetic test even if the client paid because the test would not be useful in their circumstance. After the clients had been informed of their options, which the clinicians stressed were based on scientific facts and knowledge they held at the time, some clients

44 As detailed in the next chapter these were: clients who were informed that their genetic test results were “inconclusive”; clients who were informed that their genetic test results were “negative”; and clients being informed of a “positive” genetic test result.
seemed to accept the decision while others were not convinced, preferring to favour their own opinion and reject what they were informed. These reactions displayed how, in some cases, the clinicians’ professional opinion and decision was not believed, valued or accepted because it was not in keeping with the beliefs and knowledge that the clients possessed in regards to their cancer experiences.

Regardless of whether a genetic test was conducted or not, the clinicians informed their clients of their level of risk during their first clinic appointment, and this was adjusted if new information was received (including genetic tests results). With specific recommendations and options associated with each risk category, the risk categories had physical, psychological and economic implications for that particular individual and their family members. The clinician’s decision about what to disclose and what risk to inform the client of, was based on what the clinicians knew about the family’s history, what the family knew about their history, and which clinician was undertaking the clinic appointment.

The practice of assigning clients to risk categories without any genetic testing was commonplace and reflected the confidence that the clinicians had in their clinical judgement to work successfully without the need of genetic testing. Being able to predict a client’s risk based purely on their family history was a skill that the clinicians had been trained to develop and use. This kind of predictive risk assessment is common in preventative health (Rose 1992, pp. 65–86). As previously mentioned, it reflects the notion held in genetics that the information gained from family history remains most accurate in predicting future health concerns. The clients who were denied the possibility
of genetic testing were ultimately left not being able to know if they did or did not have a familial cancer mutation.

The clients were informed about particular uncertainties by the clinicians who decided upon its relevance to each case. The clinicians did not inform the clients about the uncertainties in order to avoid unnecessary confusion for their clients, once saying that they needed to conserve the majority of the appointment for the information about the value of knowing. As discussed earlier, this emphasis on the possible positives of knowing one’s increased risk not only reflected clinicians’ overall belief in the power of the information they provided to impact positively on their clients, but it also substantiated their reason for being and practising as a service. Regardless of the outcome of a genetic test for familial cancer, there are always degrees of unknowns. Although all of the information was uncertain (the very nature of risk), according to the clinicians, they distinguished between sharing the “known unknowns” – those that were good for the client to know – and not the “unknown unknowns” that were viewed as not necessary to share. This distinction demonstrated how there was a dichotomy of knowing/not knowing for both the clinicians and the clients – which the clinicians reproduced even though they knew they could not predict if, who and when familial cancer could occur.

Conclusions: Extents of Knowing and Not Knowing
The anthropological literature on knowledge has grown rapidly over the last twenty-five years, and commonly regards knowledge as an accumulation of what people claim to know (Crick 1982, pp. 307–308; Dilley 2010, p. 176). Knowledge’s opposite, “not-knowing” or “ignorance”, has been rarely considered (Dilley 2010, p. 176).
According to Mair, Kelly and High (2012, p. 3), “a new and distinctive anthropology of ignorance” has recently emerged. Regardless of the group of people under study, ignorance can be the ethnographic object, considered not simply as the absence of knowledge, but as a historical phenomenon that incorporates certain knowledge, logistics, ethics, emotions and social relationships (Mair, Kelly & High 2012, p. 3). The notion of “not-knowing” was used by Murray Last (1981, pp. 387, 391) to challenge anthropological assumptions held about the systematic nature of medical knowledge. More recently Adriana Petryna (2002, pp. 13, 39, 44) has used the term “non-knowledge” to describe the contested nature of bio-medical knowledge post-Chernobyl and the value of that uncertainty for people who made claims on the state.

Horgan (1996, pp. 10, 23, 116) considers ignorance as being manipulated by people in power relations, constructed and imposed either intentionally or due to the limitation of science. Dilley (2010) also differentiated between unknowns that are both unintentional (due to the limit on one’s knowledge) and wilful (to deny, or refuse to take notice of, or to disregard).

If knowledge provides a sense of certainty about things, and has a reassuring effect regarding our place in the world, ignorance, by contrast, can suggest uncertainty and a discomfort about the world. (Dilley 2010, p. 188)

This quote helps explain why some clients of the Unit wanted to know risk: they predicted and/or obtained some certainty and comfort in knowing, and avoided anxiety which is a product of not knowing.
Genetic counselling focussed on the clinical work of providing certain information to the client about their risk of familial cancer, so it had educative elements that also provided the client with further access to health related options and services. In clinic, the counselling component was considered as secondary and involved the referral to psychological services if the client had unresolved emotions affecting their ability to make decisions regarding the uptake of surveillance and preventative measures.

The clinical emphasis of the appointment was an extension of the diagnostic process and was evident in the medical information sought from the client, the clinical medical spaces in which clinic appointments were held, the time spent on information provision and the decision to genetically test. The information provision involved identifying risk and relaying possible physical implications of being at increased risk of familial cancer, rather than providing psychological and emotional support. The emphasis placed on the provision of medical and genetic information was so the client could be considered informed, as much as possible, but there were various extents of knowing involving the clinicians and the clients.

The emphasis placed in the clinic on the medical practice or “clinical work” over the psychosocial work, supports the broader idea that the Unit was involved in, and focussed on: the production and dissemination of scientific discovery and knowledge, and the identification of cancer-causing genes. In the clinic, a tension between the existence of doubt, and the desire for certainty, revealed and drove medical discretion and judgement over whether something was pathological, and included or excluded from diagnostic categories (Latimer 2013a, p. 9). Similarly, in the described circumstances above, a particular form of power to include and exclude information surrounding the client’s
familial cancer knowledge, and their risk, revealed and maintained tensions between doubt and certainty, and medical discretion and judgement. The next chapter leads on from the clinic appointment examining what occurred post clinic. The chapter focusses on the identification of mutations through genetic testing, the provision of genetic test results to clients, and examines how the clinicians emphasised the potential value of mutation identification.
Chapter 6. Post Clinic and Beyond: Mutation Identification

Concepts of health and disease, normality and deviance and what humans can, or ought to be, effect understandings of DNA and genes because they incorporate “ideological baggage”. (Hubbard & Wald 1999, p. 7)

This chapter highlights some of the different ways in which clinicians and clients learnt of, provided and responded to uncertain information surrounding genetic testing for familial cancer mutations. Divided into five main sections, the first part of this chapter describes what happened after a client’s clinic appointment, how genetic testing was undertaken and how genetic test results were provided. With the term “mutation” regularly used and taken for granted by staff in their day-to-day practice, the chapter also provides and reflects on the usages and knowledges surrounding the term, not just in the Unit and by clients but also in the public domain. Regardless of the uncertainties of genetic test results, the potential value of mutation identification and genetic evidence was emphasised and practised by the clinicians. This focus demonstrated another way in which familial cancer counselling and genetic testing, undertaken by the clinicians, emphasised the potentialities – the possibilities.

Genetic Testing and Result Management
When a client was offered genetic testing during their clinic appointment, they had the option of having blood drawn immediately after the clinic, by nurses in the hospital, or at a later date at a pathology collection facility. The procedure involved the collection of two separate blood samples, via a syringe either at the same time by a different nurse or
on separate occasions. Two samples were collected so that both samples could be tested, which decreased the likelihood of errors in the identification of a client.45

No staff member of the Unit physically undertook the genetic tests on a client’s samples, as laboratories undertook this process. The clinicians decided which clients had genetic testing, what they were tested for, and where they were tested.46 A laboratory undertook the actual search for a mutation but testing a person’s genetic code was complicated because a “normal” person could have thousands of variations with unknown certainty about what they mean (Lau & Suthers 2011, p. 49). In regards to an increased breast and ovarian familial cancer risk, when the laboratory analysed the 20,000 nucleotides of the BRCA1 and BRCA2 genes, they found many genetic variations known as variants which they then compared to a database. The laboratory identified either: a benign (common) variant; a pathogenic variant which placed a woman at high genetic risk of developing breast or ovarian cancer; or variants which were of unknown clinical significance (Lau & Suthers 2011, p. 50). How these results were explained to the clients of the Unit is discussed further below.

Back in the Unit, the laboratory results were received by the data manager, who entered them into FamilyTrace, before sending them to the clinical geneticist for review. This review included the interpretation and authorisation of the results which was required because of the significant potential implications for both the client and family tested. The fact that genetic tests results required interpretation reflects Latimer’s (2013a,

45 In clients who had collapsed veins as a result of chemotherapy treatments or had a fear of needles, a cheek swab was performed.
46 The majority of the testing for mutations occurred in laboratories based in the same State, however interstate and overseas laboratories were also used. The decision as to which laboratory a genetic sample was sent depended on the type of mutation being tested for because each laboratory had different capabilities and timeframes to turn around the results and the cost involved to the service.
argument that the laboratory gives perspectives into the living body and defers to the clinic – and so it is in clinic where the relevance of the results is made explicit.

The test results either confirmed or disconfirmed the original risk status the clinicians had predicted and the results were generally provided to the client during an appointment.\(^\text{47}\) Primarily it was a genetic counsellor who provided the results because the clients had already been informed about the result possibilities in the first clinic and so were considered informed enough to understand the results, and therefore did not require a doctor.

**The Provision of Genetic Test Results**

In the results appointment, the clients were provided with the information about their genetic test result, which either reiterated or changed the risk and surveillance advice provided already. Where appropriate, relatives were risk notified (see Chapter 7 about risk notification). As mentioned earlier, certain clients were also referred (because of their cancer experiences and/or genetic mutation result) to the research nurses to be involved in research studies.

The clinicians expected *not* to find a mutation in their clients which reflected the low rate of positive mutation results (one in five). There were four instances in which clients of the Unit, regardless of genetic testing or not, were unable to know if they would experience familial cancer. In the previous chapter, I already described the first category relating to

\(^\text{47}\) Although there was a Unit policy that preferred all result appointments to take place in person, results were given over the phone at the discretion of the genetic counsellor. Exceptions were said to be made if it was in the client’s “best interest”; due to a client’s lack of transport, distance issues, the genetic counsellor believing that the client could “handle” the information over the phone and time constraints.
clients who were not offered genetic testing and who were left unaware if there was a genetic explanation to help explain their familial experiences of cancer. As explained in further detail below, the remaining three categories of being unable to know, involved clients who were offered genetic testing and were provided the genetic test results. The second category related to clients who were informed that their genetic test results were “inconclusive”. The third involved clients who were informed that their genetic test results were “negative”, and the fourth category of being unable to know, involved clients being informed of a “positive” result.

*Inconclusive Test Result*

Known by the clinicians as “a variant of unknown clinical significance”, some clients were informed of an “inconclusive” result, which left them confused and unaware if they carried a mutation. Even though such results were termed as “a variant of unknown clinical significance”, in contradiction, these results were considered by clinicians as a “negative” result because they had no clinical significance. During result appointments, clients were informed that this result meant it was not possible to interpret whether the client did or did not carry the mutation. The clinicians explained and emphasized to the clients that at the present time they could not provide them with a result. However, the clinicians provided future hope and promise to their clients, by saying that with the increase in genetic testing sophistication and knowledge, a more definitive answer would be possible in the future.

*Negative Test Result…but Not Negative*

Whenever a client had a genetic test, it was most likely that they would receive a negative result (meaning that that a mutation was not found), because four in every five tests
undertaken resulted in no mutation being found. This meant that the majority of women with familial breast or ovarian cancer did not have an identifiable mutation in the BRCA1 or BRCA2 genes. The clinicians said that where a client had genetic testing and was not found to have a mutation then that information was said to generally have reduced their client’s fear about cancer. This demonstrated how the client viewed that not having a mutation lessened their risk of familial cancer. However, a negative result did not necessarily mean that the client did not carry a possible familial cancer causing gene mutation and it did not exclude the diagnosis of familial cancer. In the results appointment, it was explained to clients that a negative result meant that the laboratory technicians could not find the mutation they were looking for because either it did not exist, it was too hard to find, or it involved a mutation that was “yet-to-be-identified”. These various possibilities were evidence of the ambiguous meanings associated with a negative test result – which were not negative. Being informed about the ambiguity resulted in greater client confusion and uncertainty for some clients, while for others the concept was not understood.

A Positive Mutation Test Result

The identification of a “positive” mutation result, known by clinicians as a “pathogenic variant”, had significant implications for both the client and their family because, even though a mutation was confirmed, it still resulted in only a prediction of familial cancer risk. Regardless of this, the clinicians emphasized the value of identifying a familial cancer mutation (see below for further discussion). The management of a client carrying a BRCA1 or BRCA2 mutation depended on the mutation, the gender and age of the client. As discussed earlier, options included increased breast cancer surveillance, risk-reducing surgery, medicines and reproductive decisions.
The first two categories described here (an inconclusive or negative genetic test result) demonstrated how not knowing shifted responsibility and accountability away from the clinician and fuelled the clinicians in the pursuit of knowledge, while the last category showed how knowing a mutation existed highlighted the clinicians’ powerful position and substantiated their expertise. How clients responded to the various prognoses will be discussed after describing the meanings and connotations surrounding the term “mutation”.

**Meanings and Connotations of Mutations**

*Terminology Matters*

When clients were informed that they had a familial cancer mutation, it was sometimes evident that a disparity existed between the client’s and the clinician’s understandings of what a mutation was and what it represented. With some clients asking the clinician to use a different term than mutation, it showed how the term mutation sometimes represented something that the client was not comfortable with hearing. Suggested alternatives by clients included, “genetic error” and “funky gene”. Other research has suggested that when health professionals discuss cancer related mutations with clients, preferred alternatives to the word mutation may include; “variation” or “change”, (Condit et al. 2004, p. 249), “alteration” or “functionally-challenged gene” (Cotton 2002, p. 3) “faulty gene” (Wakefield, Juan & Kasparian 2009, p. 395) and “altered gene” (Hodgson, Hughes & Lambert 2005, p. 417). In terms of lay understandings of the term mutation, it has been well documented that mutations have negative connotations as they are increasingly associated with abnormalities which in turn create misunderstanding (Ando et al. 2008, pp. 75–76; Condit et al. 2002, p. 69; Condit et al. 2004, p. 245; Marshall 2002, pp. 76–78; Wakefield, Juan & Kasparian 2009, p. 395). In two separate Australian studies, it was found that the terms “mutant gene” and
“mutation” were not only considered offensive by their participants but that they increased stigmatization and caused unnecessary anxiety (Hodgson, Hughes & Lambert 2005, p. 416; Wakefield, Juan & Kasparian 2009, p. 395).

When clients did not express concern over the use of the word mutation, their apparent acceptance could have been because it did not bother them (as different people have different interpretations of terms), or perhaps because the client did not feel comfortable in expressing a concern. For the clients who did object to the term, the ability to re-name, thereby re-framing meaning, enabled the clients to express agency over the result that a “mutation” had been identified. These acts also illustrated how clients respected and accepted some knowledge provided by the clinician, while resisting other knowledge. They are examples of Foucault’s work on the rejection of knowledge and power (Foucault 1980, pp. 15–16, 141), demonstrating how the clinician’s knowledge was contested (with their term “mutation” rejected) and the client’s preferences provided and used in the clinic space resulting in a shift of power.

The clinicians and clients of the Unit were not the only ones who considered mutations in a particular ways. Condit (et al. 2002, p. 1) found substantial discrepancies between the scientific usages and popular, or lay usages surrounding the term mutation. What a mutation is and what it means has historically been viewed as ambiguous and remains so, with many meanings applied to the term and no agreement about its definition existing in both scientific and popular circles (Cotton 2002, pp. 2–3).
**Historical Overview**

In order to understand why the term mutation can be viewed as both inappropriate and/or offensive by the public, it is important to first consider the history of the term. Mutation is derived from the Latin meaning to “mature”, “change” or “exchange” (Condit et al. 2002, p. 69). The term was used by the scientific community when discussing heredity in the first decade of the 20th century and, as stated, from the beginning of its use inconstant meanings and usages among researchers were apparent and these continue today (Condit et al. 2002, p. 70). The uncertainty and conflicting definitions in its meanings and usages are said to be reflections of the lack of clear evidence about the role of the genetic mutation and the differences in the “said functional applications” (the cause and purpose) of mutations by evolutionary scientists and medical practitioners (Condit et al. 2002, p. 73). A somewhat neutral and simple scientific definition is said to be that a mutation can be seen as a “change from a genetic previous state” (Cotton 2002, p. 3). Condit et al. (2002, p. 74) stated that formal definitions proposed by experts or those appearing in dictionaries rarely reflect the actual meaning of the word’s use because language and its meanings are too complex, too various and constantly changing to be captured.

Negative opinions by the public were said to have developed during the middle of the twentieth century because of the public fear of genetic mutations which saw reports directly linking genetic mutation damage with those exposed to nuclear radiation, atomic weapons and bombs, particularly after Hiroshima and Nagasaki (Ando et al. 2008, p. 75; Cotton 2002, p. 2; Condit et al. 2002, p. 72). The nuclear fallout was then heightened by the media which sought to reflect and play on the fears and anxieties by creating a plethora of science fiction novels, comics, films and television series (Condit et al. 2002,

Condit et al. (2002, p. 72) found that the public’s lay understandings and attitudes about the negative nature surrounding the word mutation had stemmed from the strong emotional connections they made between cultural meanings from both science fiction movies and the historical experience with radiation. Condit et al. (2002, p. 73) suggest that the negative connotations of the term mutation is also associated with a rejection of hierarchical understandings of variations in genes.

*Current Representations*

It was common to hear clients of the Unit mention that they had used the Internet to find out about familial cancer mutations, with some purposely not undertaking further searches because what they had initially uncovered was either confusing, and/or alarming. An image search of “mutation” in a search engine results in an abundance of photographs depicting children and animals with abnormalities (such as those born with extra digits or with malformed body parts), illustrations of DNA spirals and also animations featuring abhorrent creations. With these images, illustrations and representations readily accessible to the public through the Internet, comics, television and films, it is understandable why one might assume that human mutations are not only “abnormal” but result in frightening outcomes for the individual, their family and society. The ongoing negative exposure through popular media around the word mutation is said to account for the maintenance of the negative view held by the public, yet regardless of this the word, mutation remains part of popular discourse within genetics (Condit et al. 2002, p. 73).
Ando et al. (2008, p. 79) has stated that, unlike the public, experts regard mutations as being necessary and not undesirable. The taken for granted, frequent, daily use of the term by the clinicians both in the Unit and in the clinic reflected the increased positive role that the identification of familial cancer mutations have had since the completion of the HGP.

During one result appointment, the clinician used a white board in the room to draw the client’s family pedigree. Using the names of the family members, the clinician explained how and where they believed a BRCA1 mutation found in their family had come from and who else could have them. In this and other clinics, there was the abstract idea that familial cancer mutations existed in both individual and some biogenetic relations that were depicted and explained on white boards and on pedigrees as shaded-in boxes. These depictions are examples of how mutations and genes were on the one hand separated from the individual human being yet re-configured and connected in biogenetic ways. As argued below, the clinicians informed clients that the discovery of a familial cancer mutation resulted in greater clarity and the opportunity to prevent or minimise the severity of cancer. Such representations were very different from the images and representation of mutations in visual media and on search engines.

**The Potential Value of Mutations: Usefulness of Information**

**Familial Cancer Mutations: Confirmation, Access and the Possibility of Prevention**

Clients were said by clinicians to have largely expressed a desire to know if they had a familial cancer mutation. Cameron et al. (2009, p. 307) found that whereas some clients hoped that a mutation would be found to explain cancer occurrences and enable decision making, for others knowing they did not have a mutation provided relief and reassurance.
This illustrates how people vary in their understandings and responses to these various prognoses.

The identification of a familial cancer mutation confirmed suspicion that something was not right, helping to explain a family’s experience of cancer. When some women were informed that they had a BRCA mutation, it provided confirmation of what they already knew (given the history of breast or ovarian cancer in the family), while for other women, in addition to the mutation confirmation, they searched for secondary causes to explain the cancer experiences, a finding similar to Navarro de Souza et al. (2014, p. 673).

Other ways in which a client experienced being informed about a positive mutation result are described in a case study later.

For the clients, they were informed that the finding of a mutation was not able to result in certainty about the development of future cancer, but rather, in further knowledge and sometimes access to certain services. A common expression used by the clients was “forewarned is forearmed”. This reflects how clients felt that knowing they had a familial cancer mutation was useful and helpful as they could use that information to prevent the impact of cancer.

As discussed previously, the clinicians also considered and emphasised the value and usefulness in informing the client of their mutation status. Positive results “justified” the increased health surveillance measures and prevention options considered appropriate for their possible risk. The specific measures depended on the cancer, and were different to population based measures. For breast and bowel risk (the most common seen in the Unit) they included: clinical breast examinations, mammographic screening, prophylactic
Knowledge about one’s risk and mutation status was seen as useful as the chance of getting cancer could be reduced or treated more successfully if detected early. As explained earlier, the identifications of a client’s risk and/or the discovering of mutations was also argued to have economic benefits as the savings to the health system offset the cost of running the service. However, informing clients of their increased risk of familial cancer was seen as useful only if the knowledge altered their behaviour (by undertaking the increased health surveillance and preventative options) to help prevent familial cancers or to reduce their impact. I observed the clinicians speak candidly to some of their clients during clinic appointments about the economic benefit, for the government and society, of discovering people at high risk of familial cancer and mutations. By disclosing that it was the government who sought economic benefits the clinicians used it as ammunition to support to benefits of this knowledge while separating themselves from part of the economic process.

In an interview with Sue, one of the genetic counsellors, she explained that the “fortunate” finding of a mutation resulted in better access to services. The finding of a genetic mutation, it was said, made the client feel more empowered to go and discuss “preventative care” because they felt better informed. The clinicians provided the client with information, letters, groups and sometimes the names of specialist doctors.
The clinicians considered themselves as providing a service that was about “talking and access”.

In addition, for the clinicians, the rarity of finding a mutation was empowering in a different way than that sought and experienced by their clients. In Chapter 8 I discuss how any mutation positive results evoked emotive feelings of excitement in the clinicians, with rarer mutations resulting in further celebration. The identification of the mutation was considered exciting because it allowed for new knowledge and represented the discovery of new information, or “data”.

**Familial Cancer Mutation: A Paradox of Future Hope and Future Concern**

In Chapter 4, I introduced Mary from the Braxton family who had attended the clinic with her daughter and her sister after being informed about a possible familial cancer risk from Mary’s daughter’s obstetrician. Mary had suffered from ovarian cancer and her sister had suffered from breast cancer. I interviewed Mary prior to, and post clinic, attended their initial clinic appointment and a test result appointment. I was also fortunate to attend the home of Mary’s daughter during a home visit with a genetic counsellor, where Mary, her sister, Mary’s daughter and her elderly mother were present.

When both sisters attended the result appointment (having both had genetic testing), the genetic counsellor informed both of them that they were positive for a BRCA1 mutation. The mutation found was different from a mutation that had already been found in a biogenetic relation. The reaction of the two sisters to the positive mutation test result mirrored the belief held by the clinicians of the Unit – they largely found value in knowing that they had a BRCA1 mutation identified. The knowledge gained was viewed
as a positive thing as they felt it helped explain their experiences of cancer in the family and helped them to prepare as best they could for the future of their children and grandchildren. Although they acknowledged that the information about the impact of the mutation was not concrete, they saw the value in the information saying: “forewarned is forearmed”.

As evident throughout this thesis, knowing and not knowing is complex. During the result appointment where the value of the mutation was largely emphasised by both the genetic counsellor and the sisters, the overall belief in the value was lessened by a concern about the future implication of knowing about the mutation. Mary expressed concern that they could be held “responsible” for their mutation status asking the genetic counsellor if the government could end up “killing” those with mutations because they could be characterized as “burdens” on society. After taking time to think, the genetic counsellor responded that they (as a service) existed and were funded because identifying people at high risk of familial cancer resulted in less associated health care costs, as by preventing or reducing the severity of cancer the economic burden on society was reduced. This response illustrated how the idea of being a burden and therefore possibly harmed was improbable and dismissed by the genetic counsellor. In the appointment, Mary did not respond to the genetic counsellor’s argument, however in email correspondence with Mary post appointment, in regards to that subject, she wrote to me:

I am concerned that in the future (50 years or so) the burden on the health care system may bring about euthanasia on those individuals considered a high cost liability, eg. elderly, cancer sufferers etcetera, etcetera. If a definitive test for defective/mutated genes can be found, I am concerned that thought could be given to prevention (euthanasia, abortion) of the genes being passed to the next generation. Even with early detection and intervention, there is a high cost associated with cancer detection and treatment. I don’t think the counsellor could believe that humans could get to that degree of inhuman behaviour. I believe it is possible as money is a big decision maker.
Mary’s email illustrated a concern that with money being “a big decision maker”, then in the future the most efficient way of reducing costs of individuals considered “high cost liability”, would be to conduct euthanasia or abortion on individuals and/or on the genes themselves. Apart from demonstrating personal understanding about genetics, it also elicited a concern over the future genetic discrimination and ownership of genetic material.

During the result appointment, Mary and her sister also asked about the possible future insurance and employment effects not for themselves but for the sake of their children and grandchildren. In Australia, health and life insurance agencies were (at the time of writing), legally able to discriminate against people considered at future risk. They legally did this by insuring at a higher rate, excluding conditions or declining to insure altogether, resulting in an “insurance Pandora’s box”, where many ramifications are possible (SBS 2013). The genetic counsellor informed Mary of the current insurance implications and that the insurance premiums are based on a family history of cancer regardless of whether a gene or not has been identified. The sisters were informed that the Unit could not predict any future insurance or employment effects – demonstrating future uncertainty around possible discrimination.

The questions and concerns of Mary and her sister had not been raised in the clinic prior to genetic testing. This demonstrated how the reality of being informed about the mutation resulted not only in contradictory emotions – but new concerns and questions. These comments and questions showed concern over future discrimination on the basis of familial cancer risk and concern over the future protection of genetic information. Such concerns exist because of uncertainty regarding current and future ownership of
genetic material that is constantly changing. As discussed further below, those in public, legal and academic spheres are making similar concerns apparent. One such concern argues that genetic research, ownership and discrimination is not far removed from the eugenics movement associated with Nazi Germany (Hubbard & Wald 1999, p. 23; Petersen 1998, pp. 63, 64).

Current and Future Uncertainties

Genetic Testing for Potentiality

…calculations about future risk for specific diseases based on genetic testing of individuals are inextricably embedded in technologically produced data about the natural world, much of which is in a state of constant flux. (Chilibeck, Lock & Sehdev 2011, p. 1769)

Aspects of an individual’s past and future have the potential to be revealed through genetic testing (Taussig, Hoeyer & Helmreich 2013, p. 3). However, the possible results of genetic testing show that even with the most detailed sequencing machines, genetic testing cannot predict the biological, physical and social occurrences that effect human development and changes (Hubbard & Wald 1999, p. 36). Hall, Mathews and Morley (2010, p. 1) also highlight the limitations of genetic testing to predict disease. The presence of specific mutations (such as those involved in familial cancer) only informs about potentiality, not certainty, because the mutations are predictive and not prescriptive (Lock 2012, p. 129). As detailed in this chapter, people vary in their understandings and response to a diagnosis of potentiality.

Genetic Testing and Genetic Relatedness

By locating disease in individuals, to locating the pathological in the family and across different bodies in genetic clinics, biopower is radically extending medicine’s gaze
Through genetic testing, there was a focus and emphasis placed on genetic relatedness in the Unit. Rabinow (1996a, p. 102) has expressed concern with the implications of the new genetics as those who share a gene or chromosomes for a disorder are considered as related, and so it constructs different forms of “imagined kindred”.

Akesson (2001) described a broad spectrum in people’s attitudes to kinship and blood ties after they had undertaken genetic counselling for various diseases. These included both positive and negative attitudes from families, from how the process increased cohesion and communication between distant relatives, through to how it damaged relations between siblings, or between parents and children (Akesson 2001, pp. 129, 132). When Finkler (2001, pp. 246–247) interviewed women diagnosed as at risk of breast cancer, she concluded that families who were dispersed and not in contact could be reunited by medicalization through history taking, genetic testing and DNA storage. Latimer (2013a, p. 157) also found that some relations became more socially close because they had something new in common. This may be a positive outcome for some families, but equally, the information received may hold negative implications. With a positive mutation result it was possible for other biogenetic relations to find out that they carried a mutation, showing complications surrounding how an individual’s right to know can interfere with another individual’s right not to know, a finding also described by O’Doherty (2009, p. 465). These examples demonstrate how the right to know one’s risk, and the right to know your relatives, is mediated by genetic testing in various ways.
The Management of Genetic Material and Knowledge

Human genes are being treated by science in the same way that indigenous ‘artifacts’ were gathered by museums; collected, stored, immortalized, reproduced, engineered – all for the sake of humanity and public education, or so we were asked to believe. (Mead 1996, p. 46)

A number of ethical considerations surround the practice of familial cancer and one of them relates to who owns genetic information. There was an apparent tension surrounding who owned the genetic information that was gained from clients and/or from genetic tests. I observed how genetic material was viewed as existing both inside the individual (in a gene), inside some genetically related members (those whom shared the same genes), and also how genetic material and genetic information was separated from the person and family through the commodification, patenting and sharing of cells and genetic information.48

Ongoing decisions around the patenting of genes, DNA storage banks, the prevention of future discrimination relating to genetic information, and the keeping of genetic information as private, are considered by courts, governments and commissions (Hubbard & Wald 1999, pp. 145–157). Anthropologists have raised questions about these issues, in particular about who owns the information after samples are taken either publicly or privately (Palsson 2012, p. 190). In the rapidly changing field of genetic research, scientific discoveries are often out-dated before they are published. Gene stories derived from research promise better health, longer life and personalised medicines (Hubbard & Wald 1999, p. xvii). Such stories extend to the public domain, reach into people’s homes and, as demonstrated earlier, appear on the Internet, in news stories, documentaries and popular magazines.

48 See Chapter 8 for more information about the commodification and objectification of genetic material and information.
Complexities surface regarding the limits of confidentiality when it comes to familial disorders. A client’s familial cancer risk assessment was dependent on the genetic and health information from biogenetic relations. The client’s genetic and health information also had healthcare implications for biogenetic relations. Where a client was identified as having a mutation in a high-risk cancer gene, their biogenetic relations may carry the same mutation and therefore be considered as having an increased risk of developing cancer. Information was collected from clients about their family relations and their histories of health problems, which was then used in risk calculations and to inform decisions about genetic testing.

Sharp (2000, p. 310) highlights that there is current concern among research participants that their bodies, through being commodified, have been violated and their boundaries breached. Finkler raises questions over ownership rights when patients request or consent to genetic testing, asking “To whom does genetic information belong: the individual or the family?” (2000, p. 4). The current answer, gathered from current practices, is that genetic information belongs to the individual, the biogenetic family and others that may profit commercially from the knowledge, such as pharmaceutical corporations, insurance companies and nations.

Under the arm of the Human Genome Diversity Project (HGDP), genetics research has and is generating debates over the ownership of genetic material and knowledge (United Nations Educational, Scientific and Cultural Organization ((UNESCO)) 1997). In particular, the focus is on patent claims. Depending on the country, ownership rights may be granted for the discovery, creation, and, in turn, marketing of associated genetic processes (Andrews & Nelkin 1998, pp. 53–57; Rabinow 1996b, pp. 19, 31; Suzuki &
Knudtson 1989, pp. 178, 335–336). Rabinow’s work on French DNA illustrates how various parties may lay claim to an individual’s genetic material or coded fragments, associated inventions, and new categories of scientific knowledge (Rabinow 1999, pp. 4, 125). With the current involvement of multinational pharmaceuticals in genetics research, ownership of DNA supersedes national boundaries and enters a transnational arena, but the state ownership rights may supersede these.

Arguments surrounding patents are typically about two issues: what should be patentable, and how patents affect research practice. Whereas some argue that patents promote research, others believe patents hamper research and the free exchange of information (Hubbard & Wald 1999, p. 176). Deciding what parts of genetic material, information and its associated inventions, should or can be owned, and who can own them is complex with arguments currently playing out in court systems. In 2013, a panel of nine judges in the United States Supreme Court took three months to rule that genes extracted from the human body were not eligible to be patented (Wells ABC 2014). Perkel (2013) states that for researchers in the pharmaceutical and biotechnology industries, the decision resulted in lower cost diagnostics, more competition in the genetic marketplace, enabled diagnostics developers to design new tests without paying royalty fees and not under threat of litigation, and enabled drug companies to tailor drugs to different patient populations.

However, not all countries and courts agreed with the United States decision. In July 2014, an Australian Federal Court of five judges dismissed an appeal against the patenting of BRCA1 for the second time. The defendant, Ms D’Aracy, a breast cancer survivor but not a carrier of the BRCA1 gene, launched the case against United States
based company Myriad Genetics and Melbourne-based company Genetic Technologies (Corderoy SMH 2014). Previously, the court had ruled that the patent applied because the genetic material needed to be extracted from the body to be tested. This is an example of how human choice and action is not recognised when “discovering” the “potential” of what resides inside a gene even though it requires technological action to be realised (Taussig, Hoeyer & Helmreich 2013, p. 7).

In this case, lawyers for the defendant argued that because genes existed in nature, they were discovered rather than invented (Corderoy SMH 2014; Wells ABC 2014). Public interest and concerns surround the long-term consequences for genetic research, genetic testing and genetic medicines as the patent is viewed as preventing other companies doing research around improving health outcomes. For example, the Australian Cancer Council (ACC) released a statement arguing that gene patent laws needed to be changed to protect healthcare consumers from gene monopolies saying:

> The patents system should reward innovation and help deliver affordable healthcare, not stymie research and increase costs by allowing commercial entities to control the use of human genetic materials. (ACC in Wells ABC 2014)

Questions over whether or not the practices should condone such pursuit of improvement is continuously debated (Latimer 2013a, p. 5). In this era, individuals are made increasingly responsible for their own health and that of their children. Those with high costs, the elderly and people with mental and physical impairment, are considered and characterized as economic burdens on society. In the case study earlier, Mary expressed concern over the future of knowing her mutation status, because the government may end up killing people with mutations because they will be considered a financial burden to society. Such concerns are not unusual as knowledge and myths surrounding genetic
mutations are resulting in, and can lead to, genetic discrimination by prospective employees, health and life insurance, and dangerous medical manipulations (Hubbard & Wald, 1999, pp. 1, 6).

In the Unit, genetic discrimination was defined as the “differential treatment of healthy asymptomatic individuals based solely on their genetic makeup, predicted from genetic testing or inferred from their family history” (Unit Newsletter 2009). There are industry and public concerns held over future practices of genetic engineering and the possibility of “designer children”. Who will decide what is acceptable and not acceptable to eradicate – predispositions to medical problems, the removal or selection of traits to enhance the child – these concerns surround unequal social access and where it will lead as those with better or good “genes” will have social advantage with education, employment, health care, marriage and procreation (Hubbard & Wald 1999, p. 23; Patenaude 2005, pp. 268–269; Petersen 1998, pp. 61, 62, 68). During one Monday clinical meeting, the clinicians looked over a pedigree that illustrated multiple genetic mutations, which resulted in various familial illnesses (including cancer) across many generations and “sides” of the relations. One clinician joked saying that such occurrences (serious multiple genetic mutations and illness) could easily be avoided if only people had to provide their genetic test results to a potential partner before dating. Although the remark was made in a joking manner, it displayed a perceived use for genetic mutation testing to be undertaken and swapped with potential partners, with the hope of avoiding and eradicating particular familial genetic mutations. This illuminates how partner selection and/or procreation decisions may be impacted by genetic test results and how it could possibly result in genetic discrimination as those with less serious familial genetic
mutations are considered favourable over those shown to have more serious familial genetic mutations.

Conclusions: Post Result Appointment and Summary
After the result appointment, a letter of the results and the implication of the results were sent to the clients, their referring doctor and any other medical clinicians nominated by the client. A copy of the laboratory result was sent to the medical clinicians but not to the client unless they requested it. This was because the clinicians believed that the laboratory report could be confusing to the client due to the “technical language” used by the specialists to communicate the results.

There was an ongoing relationship between the clients and the service – reflecting how familial cancer risk care and surveillance extended beyond the clinic. Apart from the client receiving surveillance updates by the Unit and their doctors, the client was asked to inform the service of any additional individual and/or familial experiences of cancer. Likewise, the client was informed of new information obtained by the service about biogenetic relations, or information related to medical advances, technological or legal changes, which changed the information provided. The clinicians also used some of their client’s information, the medical and/or genetic material collected, and incorporated them into scholarly investigations – which they either shared with other clinicians and/or wrote up in journal articles in order to advertise their findings and to advance genetic knowledge. The process of risk notifying other biogenetic relations also took place after this time, a practice which is described and analysed in the next chapter.
It is a difficult area because it’s frustrating when you see there’s a potential lifesaving gene in the family that people need to know about and I don’t understand families who don’t talk to each other. I don’t understand that really. I recognise... I mean I recognise that it’s, it’s very common but I don’t understand that. I think if you’ve got a family member you should be talking to them, telling them look you may be at risk of a bowel cancer gene or a breast cancer gene and you can do something about it. Why would you knowingly let someone not, not know. So that’s a bit frustrating, but I guess at the end of the day it’s not my responsibility, I can offer to do risk notification, I can encourage them to do it but I’m not, it’s not a nanny state and people have to take responsibility and it’s not my responsibility ultimately, I can’t force people to give me contact details or to risk notify if they won’t and yes it’s frustrating but it’s just c’est la vie. (Interview with a genetic counsellor)

In Chapter 4, I described how each client received a consent form prior to undertaking any genetic counselling. In relation to the practice of risk notification, the consent form required the client acknowledge the role of the service:

I understand that a diagnosis of familial cancer may have implications for other members of my family and that I may be asked to assist the Service in informing them.

The function of the service included working with those considered at high risk of familial cancer, and those termed “family members”, which referred to genetically related individuals. Apart from clinicians encouraging the clients of the Unit to share the results of their risk diagnosis and/or their mutation test results with their family, the clinicians managed and undertook particular practices which involved the sharing of information between biogenetic relations. They did this by undertaking a service called risk notification. Considered a pro-active strategy, risk notification was a formal process that involved the clinicians obtaining from their client the names, and dates of births and addresses of certain biogenetic relations. Only certain biogenetic relations were risk notified when the clinicians considered them at high genetic risk of developing familial
cancer on the basis of the family history of cancer and/or because of a cancer-related mutation having been identified in a biogenetic relation.

Although the risk notification of relatives was not a legal obligation of the service, or mandatory, it was undertaken whenever the clinicians could justify it because notification by the service resulted in a greater proportion of relatives seeking further information than when notification was left in the hands of their clients. A statistical study was conducted and published by the Unit demonstrating the benefits of the practice; it was more effective at recruiting new clients when compared to just encouraging their clients to inform their biogenetic relations about the risk.\(^{49}\) Acknowledging that communication was not “uniformly effective” in the families they worked with, risk notification was viewed as having an essential role in informing relations of information that although potentially confronting could be useful to them.

Earlier, it was argued that clients were enrolled in the clinic with the genetic counselling process dependent upon their participation, requiring their information and/or genetic material as forms of evidence. This enrolment also included the clients’ acknowledgment that they would be asked for the contact details of biogenetic relations, in order for them to be risk notified. The clinicians believed that the knowledge provided through risk notification was valuable and so potentially high-risk biogenetic relations had the right to be informed. However, as emphasised throughout the thesis, knowledge in the context of familial cancer risk is uncertain and so it can be considered helpful, but it can also cause fear and have unknown and unforseen negative implications. The bioethical issues and considerations surrounding the notions of pre-symptomatic risk notification are many.

\(^{49}\) Reference not included in order to maintain anonymity.
After describing the practice of risk notification in more detail, this chapter discusses some of the emotive debates and tensions raised by this practice, including notions of family, confidentiality, the right to be informed or not to be informed, and responsibility.

The Practice of Risk Notification
In the Unit, the process of risk notification began with a clinician (whoever was assigned to the client) asking their client to provide verbal and formal written consent for the service to collect information about and contact certain biogenetic relations. The client was asked to provide contact details for as many relatives as possible, of those who staff considered were at increased genetic risk of familial cancer. Known as a “referral by a relative”, this process allowed the service to advise particular biogenetic relations of the client, via a brief letter, that a genetic mutation in an unnamed “intervening relative” had been identified and that they were possibly at increased genetic risk of developing familial cancer. The letter further stated that genetic counselling and testing was available to clarify the risk.

The intervening relatives were the first and/or second-degree relatives (due to the basis of their position in the pedigree) of a client considered at high risk of familial cancer and/or identified as a “mutation carrier”. The age at which individuals were notified was determined by the condition, the age at which surveillance for known mutation carriers should commence, National guidelines and clinical judgement. Both suspecting and unsuspecting perceived biogenetic relations thus received a letter in the mail.\textsuperscript{50} The recipient was asked to return a slip (in a reply paid envelope provided to encourage

\textsuperscript{50} In the case of those aged under 16 years (the age of medical consent in the State), the letter was addressed to the parent or guardian.
its return) indicating their interest either in attending genetic counselling or in having no further contact.

In an effort to maintain confidentiality and ensure anonymity between relatives, the letter was purposely generic in character. It did not include the name of the client who referred the recipient, or any other information about the type of mutation, in whom the mutation was found, and which other relatives had been informed. The client who had provided the contact details to the clinicians was also not advised of the response received from the biogenetic relations who received a risk notification letter. Even though very little information was passed on, because risk notification contacted people who may not have wanted to be contacted, the practice was in contradiction to the confidentiality of medical information and notions of autonomy (Bottis 2000, pp. 173–183; Laurie 1999, pp. 124–129).

Details of the individuals that were risk notified were placed in the existing client’s card file, with the connection noted on FamilyTrace. When the individual who had been risk notified responded to the letter a new card file was created and they became a client in their own right. Those who sought further information were offered an appointment and if they were living interstate they were referred to their local genetic services. When the genetic relative who received the letter contacted the service with questions about the risk, they were informed that they would need to attend an appointment to discuss their possible risks and the possibility of genetic testing. More specific information about their risk was only available once they attended an appointment because it was only during genetic counselling appointment that they, as clients, could provide explicit informed consent for the clinicians to provide them with further information.
When the individual who had been risk notified agreed to attend an appointment, they went through the processes already discussed, becoming a new client of the Unit. However, unlike normal referrals, these referrals were fast tracked and so they only waited approximately a fortnight instead of the usual six months for an appointment. This fast tracking was related to the simpler interpretation of the already known mutation (known as a pathogenic variant) because the laboratory technicians knew in what “spot” to look for the mutation in subsequent DNA samples. The client had either inherited the specific mutation or not. The easier identification of these mutations supported the practice of undertaking risk notification as it resulted in more mutation identifications for the clinicians and the service.

When the biogenetic relation of the client attended a genetic counselling appointment and had genetic testing, a cycle of obtaining consent to risk notify other relatives and inviting them to seek genetic counselling and genetic testing was repeated. With every new mutation identified in another person, a snowball effect occurred, with the number of contacted relatives growing. In some cases the informing was a rapid process, while in others it took months or years. When hundreds of families with familial cancer were identified with cancer-related mutations, it resulted in thousands of living biogenetic relations considered at high risk of familial cancer having possibly inherited the mutation. All subsequent mutations were added to the original proband’s pedigrees, allowing the clinicians to map and view the identification of the mutation and/or its possible effects (particular experiences of cancer), as it spread across multiple families and generations.

Although the snowball effect resulted in growing numbers of people to risk notify, counsel and test, in the majority of cases the service did not receive any response from
those to whom they sent the risk notification letters. In the year of fieldwork, the service’s response rate to letters sent directly to clients’ genetic relatives of people indicating interest in attending (whether they came for an appointment or not) was approximately forty per cent. All attempts to risk notify were recorded in the referring client’s folder and online on their FamilyTrace database. These included declines, no responses, or situations where it was not possible to obtain appropriate referrals and contact addresses. Where a client refused to provide the contact details of biogenetic relations, the details were also required to be recorded in their file under “incident management”.

Risk Notification and Genetic Kinship

Notions of Family

Staff of the Unit, in particular the clinicians, emphasised to clients, their visitors and myself, that overall they were concerned with the familial cancer risk of the whole family. This reflected the commonly taught, held and shared belief that familial cancer was a family disease that had implications for both the individual and the family, because families shared the same genetic material. Genetic counselling focused on the individual client’s health, which was reliant on the health history of the biogenetic-related family (both distant and potential) while instilling the client with responsibility and obligation (Petersen 1998, p. 66). As argued in Chapter 4, families were largely considered by clinicians to be made up of individuals who shared genetic material. Terms such as “familial” cancer, involving and affecting “families” and “family members”, were regularly used, however biogenetic relationships were key to the practice of risk diagnosis and risk notification. The concentration on, and perpetuation of the genetic family, extended to the practice of risk notification and was specifically evident in relation to whom the service chose to risk notify and whom they did not. When the clinicians
encouraged risk notification with their clients they used common expressions such as:
“genes are about family, we recognise that genes belong to the family” and “a genetic test
on you is a genetic test on your family”. When relatives of clients were risk notified by
the service, it was because they were viewed as being at risk of familial cancer because
they were perceived as sharing biogenetic material with the proband client in particular
ways. However, outside of the context of the Unit, clients who considered related because
they shared similar genes did not necessarily consider themselves as family members.

Although familial cancer was considered a family disease, in practice there was a tension
between what constituted familial relationships (biogenetic relations) and what should
have (social relations). The clinicians were responsible for making the diagnosis of a
familial cancer disorder in a client after obtaining, recording and interpreting information
about the client’s health history and their biogenetic relation’s familial health history.
The clinicians felt a sense of responsibility to inform and identify if certain biogenetic
relations too were at high risk of familial cancer and if they carried the known mutation.

With risk notification, individuals related through non-biological forms of relationships,
(such as marriage and adoption) did not receive the information, because they were
(generally) not biogenetically related. On the other hand, individuals who may not have
considered the client as a family member, and/or been considered by the client as a family
member, were considered as “family” because they were suspected as sharing DNA in a
specific way. Some individuals risk notified may not have been biogenetically related to
the proband client, which they may have known, or not known (in circumstances where
there were known or unknown adoption, or different biological parentage than what was
understood). The practice of risk notification therefore re-drew the boundaries of familial
relationships and did not acknowledge that familial cancer affects all those considered by the client as family members, and could possibly reveal paternity and non-paternity by identifying those who were suspected as being, but were not, biogenetically related.

The concepts of identity, family and belonging have a complex history with different cultures defining these in different ways (as demonstrated by Romanucci-Ross, DeVos & Tsuda 2006, p. 233). Through reinforcing historical concepts of patriarchy, racism, and pseudo-scientific criteria for belonging to, or exclusion from a specific territory, it has been argued that DNA research supports the practice of “genetic genealogy” (Brodwin 2002, pp. 324, 326; Nash 2004, pp. 4, 10, 26; Nelkin & Lindee 2004, pp. xii, 158). Thus, the new genetics (through research and practice) has the potential to reinforce or undermine traditional definitions and concepts of identity, ethnicity, kinship, and spatial correlation (Mountain & Guelke 2008, pp. 53–173). Specific understandings, definitions and assumptions of the composition of individuals and families were embedded within the practices of genetic counselling and risk notification that I observed.

“Allies” of the Clinic

In the Unit, for the practice of risk notification to occur successfully, the clinicians enrolled their clients in the recruitment of particular biogenetic relations through providing their contact details. In addition to contributing personal and familial knowledge, health histories, genetic material and clinical knowledge, through the provision of relatives’ contact details, they were further enrolled and engaged as “allies” of the clinic (Latimer 2013a, pp. 18, 82, 179).
Genetic discourse privileged the heterosexual nuclear family form as the norm and reshaped understandings of relatedness and kinship with the help of clients (Crabb & Augoustinos 2008, p. 303). Through the practice of risk notification the genetic information was “stripped of the social relations in which it was produced” (Everett 2007, p. 377). As argued below, this was evident in the risk notification of certain genetically related individuals, which sought, informed and revealed the genetic relationships while concealing and withholding the actual social relations (see page 245).

The Right for People to be Informed and for Clients to Inform

Legal Considerations

The potential legal implication was another reason for the practice of risk notification. The clinicians of the Unit had to strike a balance between providing information that the service was required to by law, through a duty of care to provide important medical information, and not breaking confidentiality or privacy laws. Although the clinicians were under no legal obligation to risk notify relatives of clients, they held some concern over a legal possibility of being held responsible for failing to advise relatives of a cancer risk. A concern was expressed about being held legally liable for being aware of the identification of a cancer-related mutation in a particular individual/family and failing to notify at risk biogenetic relations. As argued further below, it was problematic that the assumption of an individual’s “right to know” was connected with the perceived duty of care to inform people of a potential increased risk of familial cancer.

There was also debate about what should happen when a proband refuses to provide contact details for family members or to inform them of the genetic condition – whether clinicians have the right or the obligation to override the decision.
The Federal Government of Australia amended the 1988 privacy legislation in 2006 so that a health practitioner can disclose genetic information to family members at risk, in certain cases without the consent of the proband, in order to lessen or prevent a serious threat to life or health (Bonython & Arnold 2014, p. 168; ComLaw 2014; Otlowski 2007, p. 398; Suthers et al. 2011, pp. 385–386). Here ethical principles of autonomy, confidentiality, privacy, the right to know or not to know, and to know what, are called into question.

The initial process of referral by a relative was undertaken in anticipation and avoidance of any possible liability, as it legally made the client the referrer – thus shifting any legal responsibility away from the service and onto the original client. The legal obligation of the service to inform their clients’ biogenetic relations was not clear because it had not been tested in an Australian court. Even less clear was the extent of their supposed responsibility. For example, how distant a relative could be notified was a matter being considered by the Australian Law Reform Commission at the time of this research.

Just as there was concern that the service could possibly be sued by someone who could argue that if they had been risk notified then they would have undertaken certain measures which could have prevented or minimised the impact of cancer on themselves, there was also the possibility that legal action could be taken against the service by someone risk notified who did not want to know that they were at risk. Although those who were risk notified could choose not to attend and find out further information about their risk, the fact could not be changed that they had been made aware of a possible high familial cancer risk. Malaby’s (2002, pp. 283, 293) work on risk and contingency argues how people “variously engage” with contingency rather than minimise or manage risk.
It was apparent in the Unit that the practice of risk notification was one of the ways in which the clinicians engaged with the uncertainty as they were not able to minimise its occurrence.

**Perceived Benefits of Risk Notification**

The clinicians of the Unit considered risk notification as a pro-active strategy based on a duty of care, benefiting individuals, families, the community and the practice of genetic testing. It was considered important to identify people at increased risk of familial cancer while they were still healthy (not suffering from cancer) because of the risk reduction strategies and cancer surveillance methods they could undertake to facilitate early detection and prevention. As discussed earlier, such measures included precautionary surgery, increased surveillance, and reproduction considerations (Pasacreta 2003, p. 590; Ratnayake et al. 2011, p. 98).

The argued benefits of risk notification to the community (based on the increased numbers of people undergoing genetic counselling and genetic testing) included decreasing incidence and mortality rates by improving early detection and the prevention of familial cancer for those considered at high risk. For the clinicians, they considered the potential benefits of risk notification, namely the possibility of early detection, and the avoidance of possibly life threatening familial cancer as outweighing the negative aspect of violating a person’s right not to be informed (Stol et al. 2010, p. 393).

It was argued that another benefit of risk notification was not only in the reduced incidence and severity of familial cancer but where a person was excluded as carrying the mutation identified in their biogenetic relation, the individual was said to avoid
unnecessary investigation and ongoing cancer surveillance (beyond that recommended for
the general population). This resulted in a saving to the community and government and
demonstrated one way in which genetic materials are co-modified, valuable and
marketable (an argument extended in the next chapter).

One of the reasons for risk notification being practised by the Unit was said to be due to
the low response rate of relatives when the clinicians left the sharing of such information
up to clients. The practice was considered as removing the responsibility for clients in
contacting the appropriate genetic relatives which, it was argued, resulted in less pressure
for both the client to advise biogenetic relations and the advised relations who received
greater autonomy to decide what to do. Some clients felt a sense of responsibility to share
their knowledge, while others felt a sense of responsibility to not share. Regardless of the
willingness of clinicians and clients to share information with at risk relatives, increasing
research is identifying barriers that impede the transmission of information and the uptake
of genetic counselling. These barriers include perception of relevance, the complexity of
information, family estrangement, physical distance, and not wanting to upset relatives
(Ratnayake et al. 2011, p. 98; Roshanai et al. 2010, p. 670).

For clients of the Unit, the arguments and motives for attending genetic counselling after
being risk notified, and for informing relations of risk, related to the belief that the
information gained would help in identifying and taking control of an increased familial
cancer risk. Mothers in particular have been said to feel a responsibility and obligation to
undergo genetic testing to help their at risk children, a motive that minimises feelings of
A Sense of Responsibility and Missing a “Golden Opportunity”

For the clinicians of the Unit, encouraging the proband to risk notify relations was difficult and challenging. Whereas a responsibility was felt for offering to carry out risk notification and encouraging clients to do it, it was viewed as the client’s responsibility to provide the contact details, and for those risk-notified to take up the offer. For some of the clinicians however, a tension was evident between being aware that clients did not want to notify biogenetic relations, but of not understanding the decision.

The beginning of this chapter opened with a section taken from an interview with a genetic counsellor about risk notification. In part of the interview, the genetic counsellor said:

…it’s frustrating when you see there’s a potential lifesaving gene in the family that people need to know about, and I don’t understand families who don’t talk to each other….

Emotive issues were at play when clients, and those considered at risk, were viewed by the clinician as missing out on knowledge considered as “potentially lifesaving” because of problems with family communication. When identified, familial cancer mutations were thus considered as potentially lifesaving and not potentially cancer causing (unless they were left unidentified). In a subsequent section from the interview, the genetic counsellor expressed her difficulty in understanding why it was common that the families they dealt with “knowingly let someone not, not know” (by not sharing information).

When clients did not provide consent and did not pass on contact details to clinicians, meaning that risk notification could not occur, or when at risk relations chose to decline the offer of genetic counselling, they were viewed as failing to recognise the significance
or potential benefits of pre-symptomatic genetic testing. This frustrated the clinicians, particularly after clinicians had informed the client that something could be done to reduce or prevent familial cancer.

Similar frustrations were expressed by clinicians in understanding why individuals did not respond to their risk notification letters – why they seemingly did not want to be informed of a possible risk. In a newsletter sent to clients already considered at high risk of familial cancer, a paragraph read: “knowing you are at increased risk of cancer provides a golden opportunity for intervention through targeted surveillance and other risk reducing strategies” (Unit Newsletter 2009). Staff felt a greater sense of responsibility to inform those who they viewed as lacking information and as in need of information about potentially life savings genes and golden opportunities, than they felt to protect those who did not want to know. People’s right to know was therefore considered as greater than their right not to know. When risk notification could not be provided because their client either refused to provide consent and contact details, or when the offer for genetic counselling and pre-symptomatic genetic testing was not taken up, it frustrated the clinicians. The frustration stemmed from their confidence in the importance and potential of the information they wanted to provide. This confidence drove the service to fulfil this principle even though it was inconsistent with the beliefs and wishes of some of those it was aimed at helping.

This was evident when a genetic counsellor consulted fellow clinicians on what to do regarding her struggle to risk notify family members when a client would not return her phone calls and provide the details of her biogenetic relations. The genetic counsellor explained how the client was dealing with a lot, having endometrial cancer and a child
with a brain tumour, but then exclaimed: “we have a job to do”. The genetic counsellor
was informed to send a final letter to the client reminding her about the benefits of risk
notification and to then sign off in FamilyTrace so that actions were not pending.

One of the clinical geneticists made a comment that the people who chose not to attend
were “ignorant of its importance”. This statement displays how potential clients who did
not attend the Unit to be informed of their risks, were viewed as not wanting to know
purely because they were not aware of the importance of the information received during
genetic counselling. When a potential client did not want information it may not have
been because they were unaware of the perceived benefits but it could also have been an
active rejection of the explanation of the illness offered – because the Unit’s explanatory
models of illness (cancer caused by the inheritance of a genetic mutation) did not match
those of the person risk notified. However, the likelihood that their clients were in fact
knowledgeable about the information and that was why they rejected the offer and chose
not to follow up was also not expressed as a possibility by staff. 51

In Chapters 5 and 6, the varying and often ambiguous meanings of communicating risk
were highlighted. Risk theory is important to consider here because the practice of risk
notification, which aimed to get potential at risk clients to attend familial cancer genetic
counselling, relied on it. Informing those that they could be at risk, shifted their fate from
diverse aetiological theories of disease to the notion of a mathematical probability which
they could possibly control the outcome by taking up the recommendations.
The client and their family were then allocated the responsibility for risk reduction (see

51 Nor were other possible factors such as the wrong address, family estrangement, fear, too busy.

“Non-Responders” and the Right Not to be Informed

Biogenetic relations who were informed that they may be at genetically increased risk of familial cancer and did not take up genetic counselling, were termed “non-responders”. The reasons provided by clinicians as to why they had non-responders related to the individuals’ confusion about the significance of the information. Due to respecting confidentiality, however, the Unit was not able to contact non-responders and ask why they did not take up genetic counselling and pre-symptomatic genetic testing. Recent literature has investigated reasons why relations choose not to attend. Relevant factors suggested have included the invasion of privacy, a lack of understanding or awareness of risk despite receiving information about it, the lack of importance or the acceptability of preventative options, fear of genetic discrimination, health insurance consequences, the psychological impact of test results, possible costs, and relationship and communication problems between family members (Gilbar 2007, p. 390–391; Kieran, Loescher & Lim 2007, pp. 101–102; Pasacreta 2003, pp. 590, 596–597; Ratnayake et al. 2011, pp. 98–99; Stol et al. 2010, p. 394).

Regardless of the cause, potential clients’ actions of rejection, of not answering the risk notification letter and/or not choosing to attend an appointment, was a deviation from the clinicians’ medical recommendations which they believed could have led to the prevention of future cancers through the knowing and control of the risk. This view reflected the confidence the clinicians held in relation to the concept of risk. By choosing not to go to the Unit to have a genetic counselling appointment and find out information
offered by the Unit, potentially at risk clients displayed their agency in choosing not to know further details. Taussig (1980, pp. 4, 10, 12, 13) wrote that the physician’s ability to explain and cure is linked with the power to control and when patients are unwilling for diagnosis or treatment they challenge the physician’s authority, both personally and their role socially.

Various practices and debates surround clinicians contacting the biogenetic relations of a client in order to maximise the potential for reduction of familial cancer by ensuring communication of information about the disease (Godard et al. 2006, pp. 103–110; Gold 2004, pp. 72–78). Much of the debate has been about people’s right not to know by choice – which is difficult given that people must know there is information available in order to choose not to know (Adorno 2004, p. 435). The reasons for informing relations, and for wanting to be informed, elicit strong views and emotions; likewise there are strong arguments against the practice of informing, and of being informed of one’s risk without consent, including autonomy, confidentiality and ownership of genetic information.

Published literature by similar genetic counselling services both interstate and worldwide that have contemplated and evaluated the use of risk notification, using surveys of the general population regarding views about risk notification and/or of short-term risk notification trials (including Ratnayake et al. 2011, p. 99; Roshanai et al. 2010, p. 670; Stol et al. 2010, p. 392). The decisions by similar services not to implement risk notification were ultimately because of the possible greater psychosocial, ethical and legal implications of cancer familial risk notification. The disclosure to an individual who did not actively seek the information and therefore had no choice of being informed that they
may be at risk could be considered as detrimental to the individual by changing their perception of risk, sense of privacy, psychosocial well-being and their family relationships.

Not being able to make a choice about knowing or not knowing medical information raises issues of privacy or autonomy and is debated in literature (Adorno 2004, p. 435; Bottis 2000, pp. 173–183; Laurie 1999, pp. 119–130). When a person receives a letter informing them that a familial cancer mutation had been found in an intervening relative and that they may be at increased risk, the knowledge of being potentially at risk cannot be undone. This demonstrates how being risk notified is complex because the knowledge of being at risk carries certain responsibilities (Stol et al. 2010, p. 393). The person risk notified needs to decide whether to accept the offer of genetic counselling and genetic testing, to learn more about their possible risk, or to reject the offer. Where a person is informed that they are at increased risk of familial cancer, they need to decide if, and to what extent to act on the risk and the associated risk-reduction measures provided.

For the possibly unsuspecting and unwilling biogenetic relations who receive risk notification in the mail, it may violate their interests by not having a choice to act on their right to not know. The imparting of familial cancer risk information in the home meant that the power of the clinic was extended to the home of the recipient – no longer bounded in the physical clinic and sought by individuals. For those risk notified by the Unit they had no choice in being made aware of a possible high familial cancer risk and possibly had no prior knowledge of this risk – this demonstrated the power the clinicians had in informing certain individuals.
The Maintenance of Genetic Information as Confidential and Private

Although clinicians considered familial cancer as a family disease, which had implications for both the individual and the family, there were clearly defined limits to the information passed on. Much of the information the service held about the families was considered private and a breach of confidentiality if passed on. Due to this, only the details about the possible risk of the person risk-notified were provided. Staff would not provide information to relations of other clients in regards to which family members they had seen, and any other details to do with their case (such as who had testing and what the results were). Even so, the power of clinicians to share genetic test results at their own discretion can be considered as undermining the confidentiality on which the health care practitioner and patient relationship is based.

In the clinic, medical discretion extended to the concealment of information about relations. The impact of not disclosing particular information was made apparent during one clinic appointment. In the appointment, after the client had asked about whether their cousins had attended and were told by the clinician that they could not pass on the information, the client replied how she wanted to know who had been notified so they knew whom they should try to contact. The client further said: “we will just ask”, demonstrating how, in this case, the client had the ability to by-pass the clinician and contact the cousins directly to find out the information.

In another instance, during an official debrief at a Monday clinical meeting, a genetic counsellor relayed to fellow staff information about a client’s clinic appointment. This client had attended clinic after receiving a letter saying that an “intervening relative” had been found with a mutation. The client’s father had died when she was eight and she
had little knowledge of her family history. This was evident in the family pedigree for which she had provided information – which only displayed the name and dates of birth of herself, her father and grandfather. The genetic counsellor informed fellow staff that she “had to be careful” because the client was not aware that she “belonged”, by being genetically related, to the large Gilmore family. Although the majority of the “family” members lived interstate, the clinicians saw various clients belonging to the Gilmore family in their state and this was why they had a copy of their large family history book in the Unit.

Upon my interviewing the genetic counsellor later, it was explained that “being careful” meant that she took certain precautions with this client. These precautions not only involved not providing details about the mutation and the intervening relation identified with the mutation, but also by not providing any other details about the knowledge, and existence, of the interstate family to which she was said to belong, even if the lack of familial information caused the client distress. The genetic counsellor stipulated that the service was there to provide information and advice in relation to a client’s risk of familial cancer and not to inform them of any other familial related information. This demonstrated a few key things: how clients only had the right to be informed about their own familial cancer risk but not about biogenetic relations; that the service held knowledge of biogenetic connections between people, which they termed families; but that the staff owned the knowledge and protected this knowledge.

Clients were informed that they could not be told of the specifics around which family members had mutations identified, the type of mutation identified, and which family

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52 As described in Chapter 4.
members had been informed of the potential risk. This demonstrated how there were attempts made to try and separate biogenetic information from individuals, and their social and biogenetic relations and vice versa. It became apparent that it was not possible to entirely cut the ties linking the person, relations and the familial information (see below). There are deep implications for individuals, families, genetic services, for notions of kinship and relatedness in the practices of risk notification because of the inseparable connections between the individual, family and information.

**Figuring it Out Anyway**

As discussed, for those risk-notified who wanted more information about other relations, they were informed that the service could not provide them with that information. Although the protection of what was considered private information was fundamental to the service, in practice the clinicians were not always able to protect biogenetic information. In clinic, the clinicians’ subsequent explanations to the clients regarding their risk level and/or per cent, and how they measured their risk often revealed certain clues about the side of the family, generation and gender of the person in whom the mutation was found.

During one clinic appointment with sisters Mary and Kim (mentioned previously), they explained to the genetic counsellor that while they were waiting for their appointment they had received a letter which informed them of the finding of a BRCA1 mutation in a biogenetic relation considered four generations removed. The risk notification letter had been sent from a registry involved with a national study on ovarian cancer that had collected the information from the relative identified with the BRCA1 mutation through a genetics service. Mary asked Ann (the genetic counsellor) which other family members
had familial cancer related mutations identified. The genetic counsellor replied that they could confirm that there was a mutation in that part of Mary’s family, but:

we’re quite often in a position where we know sometimes more about your family than what you might, and due to confidentiality and privacy I can’t give you the specifics.

Although the sisters were informed that the service could not provide them with specifics around which other family members had had mutations identified, the subsequent explanation of the sisters’ risk revealed certain clues. They were informed that the risk of Mary and Kim experiencing another cancer was “around 12.5 per cent”. Ann explained that in reality, their experience of ovarian and breast cancer meant their chances of carrying the mutation could be much higher. The number was statistically low because they (the service) generated the information on what had been found and because a BRCA1 mutation had been identified “four generations away”, they (the service) did not know if the sisters’ “intervening relatives” carried the BRCA1 gene. The number would change when the clinicians found out if the mutation occurred in genetic relatives considered “closer” to the sisters. It was further explained to the sisters that if they were found to have the BRCA1 mutation then their risk of familial breast and ovarian cancer would be statistically at the highest number.

In relation to risk notification and notions of privacy and confidentiality, this example demonstrated the complications in managing shared genetic information. The calculation and explanation of a client’s risk per cent revealed information about which “side” and “generations” of relatives were and were not involved in the risk calculation. The sisters gleaned from this information who the BRCA1 carrier was. Subsequent genetic testing of the sisters revealed that they both carried a BRCA1 mutation but that it was different from the particular BRCA1 mutation that had been identified in the biogenetic relation
four generations removed. This resulted in genetic testing being sought from the sisters’
mother to see if the mutation had not passed through the paternal side (as suspected) but
instead had passed from the maternal side. The clinicians aimed to identify and map the
mutations as they passed through generations of biogenetic relations so the clinicians
could more accurately provide familial cancer risk notification and prediction.

Conclusions
The formal practice of risk notification as performed in the Unit, required the clinicians to
enrol clients in providing consent and provision of contact details, in order for the
practice to be considered as adhering to autonomy and confidentiality. The clinicians
provided information about cancer mutations and the risk of familial cancer among
certain biogenetic relations in a particular clinical manner – through a letter, with de-
identified information and the offer of genetic counselling and pre-symptomatic testing.
The clinicians of the Unit felt a sense of responsibility to inform those possibly at risk,
and to undertake genetic testing to identify familial cancer mutations. Risk notification
was considered to be about identifying clients at high risk in order to inform them,
provide access and encourage to reduce or prevent the risk of familial cancer which
would result in economic savings, and to protect the clinicians and service from potential
liabilities. Although the clinicians held knowledge of biogenetic connections between
people, which they termed families, the clients only had the right to be informed about
their own familial cancer risk and not about biogenetic relations. With the clinicians
owning and protecting this knowledge, the practice demonstrates the amount and power
held and perpetuated by the clinicians.
By increasing the net to identify more people with mutations, the practice was also about the need to identify, authenticate and progress the number of people identified with familial cancer mutations. This need also related to the value placed on the discovery and progression of knowledge surrounding the science of genetics, a topic which is discussed further in the next chapter. I argue that being excited, betting and competing over the prediction, finding and identification of mutation, was not only a way in which the clinicians engaged with uncertainty. It also displayed value and was encouraged because it held both individual, group and community benefits tied to the potential and the advancement of scientific knowledge and progression.
Chapter 8. Scientific Competition, Discovery and Progress

Betting and the Chocolate Frog Accolade

It was during one Monday clinical meeting after discussing a client’s case that two genetic counsellors bet a chocolate frog against the clinical geneticist, that a mutation was going to be identified in the client. Using a red marker, the words “BIG CHOC” were written on the top right hand corner of the meeting room’s white board in capital letters followed by the client’s file number, the initials of the staff members involved in the bet, and “No” and “Yes” to indicate their prediction.

The act of documenting the bet on the board served as a reminder to those involved in the bet as to who predicted what outcome, and it made the bet visible to all staff members (including those not involved). Just over two months later and during lunch one of the genetic counsellors announced to the group present that the client was negative for the mutation. A large milk chocolate frog measuring fifteen centimetres across and wrapped in gold foil had been bought from the State’s leading maker of fine chocolates and handed over to the clinical geneticist who won the bet. The winning clinical geneticist held up the chocolate frog and asked if I wanted to take a picture.

This prompted one of the genetic counsellors to ask what I, as an anthropologist, thought of their betting. Another genetic counsellor quickly replied that the bets did not impact on the outcome, that it was the laboratory who looked for the mutations. At this time, nothing more was said, and the winner broke up the frog into chunks and left it on the meeting room bench informing all the staff in the Unit that it was there to be shared.
Even though the frog was too large for one person to eat, the decision not to take it home, but rather to allow fellow staff members to share in the winnings, displayed generosity and fostered a good winning team spirit and staff comradery, while reminding staff of the winner and showing that the bet was not taken seriously.

It was during lunch the next day, after one particular genetic counsellor had taken time to reflect on the bet, and my presence of documenting the bet, that concerns were raised about how bad the practice would look if documented in my thesis, saying: “what would they [the clients] think”. Other staff present responded that it was not bad, with one saying: “all work places do it...use black humour as a way of coping and getting the mind off of it”. The concerned genetic counsellor said: “it looks like we are betting for him to get cancer” referring to the client they had bet about. Another genetic counsellor responded: “we don’t mean anything by it, it’s not personal”. This conversation demonstrated ways in which my presence caused some staff to reflect on the meaning and appropriateness of their betting practices. For other staff, the practice of betting was not related to the individual client as it could not impact on whether a mutation was or was not found, and because they used betting to incorporate humour as work coping mechanism.

This case raises questions about the type of enthusiasm shown for finding mutations and its role in the practice of genetics. This chapter aims to place particular practices in the Unit within the broader context of scientific competition, discovery and progress. The first section reveals cases of betting in the Unit and its role in the Unit and links them more broadly to the competitive practices undertaken during the human gnome project. I will explore the ways that staff of the Unit regularly exercised their competitive nature
and expressed excitement. The chapter then analyses the factors influencing the excitement and disappointment expressed by staff of the Unit involved with the discovery of mutations and advancing knowledge.

In order to explain why the clinicians of the Unit displayed enthusiasm for mutations and competed through betting, it is important to explore some of the global practices and competitions held in order to situate clinicians’ practice in the broader context of scientific discovery and progress. One form of client objectification and commodification was visible in the practice of competing and betting over the discovery of mutations. The section which follows introduces and discusses the objectification, ownership and commodification of genetic material and knowledge by scientists, clinicians and companies involved in research and clinical practice of the new genetics on a global scale. It discusses the importance of the pursuit, prediction and finding of knowledge and the identification of scientists and clinicians as most accurate. In the Unit the objectification and commodification of genetic material was evident in numerous ways.

The final section combines the two worlds, the Unit and wider new frontier practices, describing a conference where likeminded peers (including some staff of the Unit) gathered to share their local genetic knowledge and clinical practices in particular ways. Through meeting, sharing and debating, the conference and its attendees fostered competition, enthusiasm and advancement within the genetics community. The chapter demonstrates how the practices of the Unit reflected, and contributed to, the global scientific competitions and betting practised as part of the new genetics as situated within the broader pursuit of knowledge and certainty.
Competition and Excitement Around the Discovery of Mutations and Advancing Knowledge

Magic (of which gambling is a functional equivalent), science, and religion are a “three-cornered constellation”. They are distinct but interconnected modes of adjustment which enable men to meet uncertainty, attain rational mastery of their environment, and deal with problems of meaning respectively. (Fox 1959, p. 232)\(^{53}\)

In 1959, a classic ethnography, *Physicians and Patients Facing the Unknown*, was published by Renée Fox. Part of the ethnography describes the behaviour of physicians in a metabolic ward when it came to betting. Fox described how physicians played “the game of chance” involving “guessing and wagering behaviour” that was always undertaken with banter and joking (Fox 1959, p. 82). This joking partly reflected the humour of the group but it also was how the physicians acted out the uncertainties and limitations that they faced (Fox 1959, p. 84). The physicians likened some of their scientific endeavours to lotteries because of their inability to predict, control and explain (Fox 1959, p. 84). Through placing bets on the possible outcome of tests, through gambling jargon and humour, group wagering was considered as having several functions (Fox 1959, p. 83). It was an organised way of dealing with clinical and research problems and formulating medical prediction. It also served as a form of “friendly competition”, which they hoped would confirm their predictions or reveal new knowledge which would increase certainty and control (Fox 1959, p. 85). Lastly, Fox argues that betting was a form of assertion: in spite of the fact that their knowledge was not sufficient, they did not let it deter them and proceeded anyway (Fox 1959, p. 84).

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\(^{53}\) A connection between magic, science and religion was also made by Emily Derkheim “The Elementary Forms of the Religious Life” (1947), and Bronislaw Malinowski “Magic, Science and Religion” (1948).
Competition through Betting In the Unit

The objectification and commodification around the new genetics extended to the practices of competing and betting (both physical and verbal) over the discovery of new information and mutations in the Unit. These practices took place and surrounded the discovery of mutations by the clinicians. Physical betting incorporated a prize to be won, and verbal betting, which occurred more frequently, involved only a verbal exchange, illustrating how the practices were to gain status, and associated social capital, rather than for financial or physical gain. As demonstrated with examples below, excitement surrounding the discovery of mutations and the competitive practices shown and expressed by staff were indicative of the competition that exists within the genetic community, and biomedicine, serving different roles and functions depending on the contexts.

Being the “Queen Of Mutations”

Early in December 2010, the staff of the Unit held a Christmas breakfast celebration. By 9:00am, what was normally the meeting and lunchroom had been transformed into a space of celebration with the addition of Christmas food, decorations and presents in the corner ready for the Unit’s annual “Kris Kringle” (see Figure 11, page 113).54 While staff ate, an A3 sized pie chart was placed on the white board. The black and white pie chart had been divided into four sections and filled in with a black marker with numbers and mutation acronyms. The paper read, “2010 Mutations – Guess The Counsellor” and had a gold sheriff’s badge attached (see Figure 16, next page).

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54 Kris Kringle involved each staff member picking out of a hat the name of a co-worker and then buying them a gift. The receiver was not meant to know who the giver was even after the exchange of gifts had taken place. The game that required the exchange of gifts between staff encouraged fun and the helped cement social bonds.
One of the genetic counsellors announced a game for staff to play. The game involved staff members guessing the correct order of the genetic counsellor with the highest “hit-rate”, or the most success in finding mutations in the clients they had ordered testing for in last year, down to the genetic counsellor with the lowest “hit-rate”.55 A total of 23 mutations had been found for the whole year, which demonstrated the rarity of mutation identification. All staff members guessed correctly the winner, a genetic counsellor frequently called “the queen of mutations”. This label was used often by staff to refer to this genetic counsellor, reflecting the frequent mutation discoveries in her clients compared to her fellow clinicians. As a form of praise, the label was predominantly used.

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55 The hit-rate had calculated a percent based on mutations identified compared to the number of genetic tests ordered.
by the clinicians – those who also saw, selected and tested clients, and so it indicated that
the charting and recognition of such results was important to them.

While the winner was guessed correctly, no staff member guessed the overall correct
order. When the order was revealed surprise was expressed over the longest standing
genetic counsellor of the Unit having the lowest hit-rate. Questions were asked of the
genetic counsellors who had made the game, including if they had calculated the hit-rate
against the number of mutations tested for. The surprise reflected the belief that the
clinicians played a role in deciding and selecting to which clients they offered genetic
testing to identify a mutation and to whom they did not – hence the belief that they could
influence the hit-rate through the use of their clinical judgement which, in this case, was
presumed to be linked to the number of years’ experience. With the lowest hit-rate
achieved by the longest serving genetic counsellor, the findings were contrary to their
expectations which caused confusion and reflection about how their clients were selected
for genetic testing.

With only the genetic counsellors’ scores on the game, there was also disappointment
expressed about the absence of the clinical geneticists’ scores because they too wanted to
see their results. They wanted the recognition that they too saw clients and ordered
genetic testing and so they wanted to see how they, as expert doctors and clinical
geneticists, compared to each other and with the genetic counsellors. The participants and
winner of the game did not receive any physical present or gift because the prize was the
verbal declaration to be known by all staff. This demonstrated how the game was about
peer recognition and reflected the competition and co-operation of scientific endeavour.
Learning through Betting

Verbal betting also took place when staff contributed their opinions on the possible test results of other staff members’ clients. When one verbal bet held between the two clinical geneticists resulted in an “equivocal”, or inconclusive result, the result was used to learn from. As discussed earlier, an equivocal result meant that the lab did not find, but could not rule out, the presence of a mutation. In this bet, the equivocal result meant that a winner could not be declared, because technically they were both wrong having nominated a “yes” or “no” result. Learning from this, it was agreed that betting, from that time forth, would include the option of an “equivocal” result along with “yes” or “no”. The change in their betting behaviour and the rules of the game not only reflected the nature of the mutation test results but the importance of undertaking such a game – that it could be learned from. Such a practice is not only a reflection of the idea that scientific experiments or research promotes learning, discovery and engagement with the scientific field but that it sometimes occurs in unconventional or unexpected forms. Learning through betting, by testing knowledge and incorporating new knowledge to change practices along the way, led to the gradual growth of the clinician’s medical and genetic knowledge and clinical judgement.

The Role of Betting

From the physical and verbal betting examples provided, it is evident how in this particular Unit, betting was not practised to obtain physical prizes; rather like Fox’s (1959) findings discussed above, betting fulfilled many functions and provided benefits. It was an ongoing practice that selected staff undertook as a form of recreation and fun that, at the same time, promoted learning and created a break from the mundane work environment, enabling a sort of “time out”, which separated their work practices from the
reality of the situation. Thus betting was used as a coping mechanism and a form of objectification, separating genetic outcomes from both the client and clinician.

The practice of balancing risk taking versus return is evident in betting as the practice of betting and competing suggests there is some predictability. These are forms of interpersonal engagement and engagement with the future random events. Predicting genetic test outcomes reflected more widely on the nature of the clinicians’ job roles – the provision of risk estimates to their clients on a daily basis. Through betting, the social capital to be gained was more important than physical capital or reward. All staff members who played shared in the glory of winning – winning through being right. Betting provided another way in which staff expressed their expert knowledge in the prediction of their clients’ risk amongst their fellow co-workers and, by doing so, it provided a platform in which each other’s achievements could be recognised and dominance could be asserted. It was also about highlighting which staff members knew what. Although clinicians were not able to determine whether a mutation was found, they chose the clients to have testing and betting was about trying to predict what was not yet known. It was important to the clinicians because it enabled them to compete and see where each staff member stood in relation to one another and enabled the testing of their clinical judgement – whether they played a role by choosing which clients could undertake genetic testing and have mutations discovered. Betting displayed and acknowledged the vested interests of the Unit’s staff in outcomes, which is similar to the competitive betting practices undertaken in the broader genetic field.

Although there were never any explicit rules made about who could play, the betting only ever involved the clinicians (those who directly saw clients). The remaining staff either observed or expressed their encouragement by taking sides and/or following up on the results.
Competition in the Human Genome Project

Competition in medicine and genetics is viewed necessary in order to progress the science, with a deep seated desire amongst scientists to understand the human body by breaking down the body into miniscule fragments or genetic material – evident in the HGP and its “mapping” on a global scale (Sharp 2000, p. 314). Reductionism, the process of reducing people into their smallest parts (genes, cells and atoms), is a common practice in science and said to be most evident and extreme in the HGP (Hubbard & Wald 1999, p. 3). One of the primary goals of the three billion dollar HGP when it began in 1989 was said to be in the identification of the genetic causes of common diseases (such as cancers) for which medicines could be generated. The developers were so confident in the project that Bill Clinton (the American president at the time) was encouraged to describe how it would: “revolutionize the diagnosis, prevention and treatment of most, if not all, human diseases” (Wade 2010, p. 1).

A timeline of ten years was given to complete the genome, with treatments expected five years after that – however complexities around the genetics of disease were not anticipated. Both those within the community (scientists, geneticists, doctors, clinical researchers and other workers) and those outside (the public, media and psychosocial researchers) have commented, not only about the elusiveness of the project and how the goal or “race” still remains, but also about how new information is causing changes to what was known and causing a “major paradigm shift in human genetics” (Gibson 2010; Wade 2010, p. 3). The year 2010 marked the ten-year anniversary of the completion of the HGP which generated much discussion about the progress that had been made and the discoveries still to be found. As part of this, there was a dedicated special edition produced for the highly distinguished scientific journal Nature. In one article, the author,
Alison Abbott, conducted interviews with the major players of two teams of main “competitors” involved in the HGP creation on what it was like for them to take part in the “fastest, fiercest research race in biology”, a race in which they officially tied (Abbott 2010, p. 668). The article highlighted the range of tactics, obstacles and frustrations faced, alongside the advances and discoveries made (Abbott 2010, pp. 668–669). This example demonstrates how the practice of competing in this area was not only set up and practised, but deeply ingrained and an expected part of the science of discovery, progress and the production of genetic knowledge.

*Betting Over the Number of Human Genes*

During the HGP when the human genome was sequenced, the number of genes it contained was to be calculated. It has been documented that races took place and bets were also made, as to how many genes would be discovered (Gannett 2010; Normile & Pennisi 2002, pp. 32, 34; Pennisi 2003, p. 1484). An informal contest termed “GeneSweep” was organized at a genome meeting and involved researchers predicting how many protein-coding sequences it took to make a human. Most estimates insisted humans had around 100,000 genes, and when results found the number to be between 25,000 and 30,000, the researchers were extremely surprised at the low number; the result meant that even the rice genome had more genes than humans. With only a few researchers having bet that the number would be that low, a sequencer and two of her colleagues were declared winners of the pool having predicted 25,947 genes in 2001. Their prize was half of a $1200 pool (Pennisi 2003, p. 1484). With such a low monetary value awarded, it demonstrates that the act of betting, in this case, was more about the

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57 The two main opposing competitors were Francis Collins and Craig Venter. The official tie was announced by Bill Clinton at a ceremony at the White House in 2000 (Gibson 2010).
prestige and the social capital gained from winning by closest prediction. Betting demonstrated one particular way in which the clinicians displayed interest in the outcomes of genetic mutation testing, another way was shown through displays of excitement and disappointment.

**Excitement and Disappointment of Mutation Identification**

In the Unit, excitement was expressed, discussed and displayed across many tasks and contexts, occurring prior to, during, and after a mutation had been identified. As highlighted with examples below, excitement involved various emotional responses that were displayed and attached to the search for mutations. Sometimes the clinicians expressed their enthusiasm explicitly by saying bluntly how they were “very excited” about a mutation being found. While another time, the possibility of obtaining a mutation was expressed in a different form when a genetic counsellor working on a client’s pedigree on her computer screen said out loud “2, 4, 6, 10, 12, 15… yum, yum, yum”. The higher the total number on the client’s pedigree, the higher the client’s risk and chance of finding a mutation. This occurrence displayed how this particular clinician expressed enthusiasm over the possibility of finding a mutation while assessing and calculating a client’s risk. The range of excitement shown was also dependent on how long it had been since a mutation had been found by the Unit and the type of mutation found, with the rarer mutations resulting in more excitement shown by staff. Further examples are discussed below, followed by analysis on the function of such excitement.
In Chapter 4, I detailed the general sequence of the Monday clinical meetings held in the Unit. I explained how one of the first agenda questions asked if any new mutations had been found, while a vigorous drumroll on the table built anticipation amongst all staff members and signalled the approach of possibly exciting news that a mutation had been found. This regular weekly practice, a ritual, demonstrated a sense of team social bonding and excitement in the environment. The majority of the time, no mutations had been found, reflecting how four out of every five genetic tests came back as negative or equivocal (meaning that the results were unclear). Regardless of the number of mutations found, the weekly ritual remained, which demonstrated a commitment to future diagnostic certainty.

When a mutation was found, details about the type of mutation or about the client and/or particular relations were shared. All mutations including common mutations (e.g. BRCA1) were greeted with cheer or applause in recognition of the find, while with rare mutations staff responded with increased and/or louder cheers of excitement. As discussed below, increased excitement was shown towards cases involving: first or rare mutations, the finding of a mutation in a client that they did not expect, or if a client or their family members who had been awaiting results for a long time.

First and Rare Mutations

In one instance Joy, a genetic counsellor, informed the group that they had found their first “folliculin mutation” and that they had actually found two because both the father and his son were identified with the mutation. After everyone had cheered, Joy stated: “we are very excited, I don’t think the family will be, but that doesn’t matter”.

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This statement demonstrated how Joy was upfront in disclosing how although the family was not going to be happy with news about the mutation (which she had found out during their appointment), this did not matter. In this case, the reward of finding a mutation was not about the clinicians being able to make an emotional empathetic connection with their client in order to help the family. Rather, it was about being the first to find, to discover, a rare mutation – an achievement that reflected a goal that surrounds scientific/genetic discovery and progress. The excitement over the discovery of a new mutation for their service was also related to there being little knowledge surrounding it, and so the discovery and information to be gained held both personal and broader benefits. This is discussed further below and is linked to broader concerns and aims in genetics and science.

Engaging and Enraging Clients

Most displays of excitement were observed “in-house” and between staff, however there was an instance where a debrief provided insight about its display with clients. Upon the return of the genetic counsellors from a high-risk clinic (see Chapter 4), Sue (one of the genetic counsellors) explained that because the doctor did not arrive, they spent the session talking with the clients. During this time the clinicians spoke about their excitement over finding mutations, in which one client, reportedly upset, responded: “why are you getting excited about bad things?” I asked how Sue had responded and she explained that she informed the client that the clinicians were excited over “the usefulness” of the information. Joy, the other genetic counsellor present, said that she thought the clients wanted to know they had mutations too but they did not get excited about them saying: “they know it is useful and not knowing is bad and not helpful”. This is an example of a schism; how Sue and Joy were excited to discover mutations for
their usefulness and to promote and share selective knowledge to engage the interest of certain clients. The clients also wanted mutations to be found, but this was for the certainty and knowledge they believed it would bring and not because they were exciting to discover.

**Earlier Age Onset = Better Chance of Mutation Discovery**

Although staff’s emotional reactions reflected beliefs held about the positive nature of the information gained from mutation discovery, sometimes the clinicians were openly reflexive about their reactions. One such instance occurred during one of the Monday clinical meetings, when one of the clinical geneticists was figuring out a risk assessment for a client by looking at the family pedigree in front of her. The clinical geneticist said that the client’s mother had bowel cancer at age thirty-five. Sheryl, the genetic counsellor who was going to be involved in the case cheered “yeah!!”, while pumping her fist in the air. The clinical geneticist then corrected herself saying it was at age seventy-five, while laughing at Sheryl. Sheryl laughed and looked at me saying: “you must think we are sickos, getting excited by early onset cancer”. This demonstrated how my presence impacted on how Sheryl reflected on her own reaction of getting excited over a mistaken case of early onset cancer, even though her reaction reflected their professional knowledge and beliefs; knowledge and beliefs that connected earlier experiences of cancer with a better chance of finding a mutation in the client.

**Given Time Mutations Would Be Revealed**

The length of time it took to find a mutation also impacted on the degree of importance placed on the find: the longer the time, the more precious the positive mutation results. An example of this involved Joy, saying that she was able to retire because a “PTEN
mutation” had been found in one of the families she had “hoped to get answered”. Joy explained how it was important to her that something was found in this particular family because they had had “ongoing” investigations for the twelve years that she had been working in the Unit. Ongoing investigations referred to the variety of genetic tests that had been conducted on the blood collected from different relations throughout the past twelve years, and the ongoing communication Joy had had with the client and their family members as more cancers and tumours were discovered. Joy’s explanations illustrated how different clients and families were bestowed with different levels of hope in finding mutations. In this example, Joy’s clients were not equal – those that had waited longer for a result deserved to have a mutation found, more than those who had not waited as long. The finding of an answer provided closure and hope for Joy and the service.

The case also raised concern about the twelve years of negative mutation identification and a question about how many other families were in the same position, after also being found to have tested negative for a BRCA mutation. As explained earlier, when no mutation was found, cases were closed, meaning no more work was done on them, unless new information prompted the clinicians to reconsider the clients. The clinicians agreed that there could be many clients in similar positions, but that there were too many for the service to organise retesting of them all. They would have to wait until further experiences of cancers warranted further investigations. This example demonstrated that even though the clinicians admitted that they could not retest families with similar

58 The PTEN gene made of phosphatase and tensin homolog protein, results in a large number of cancers and/or non-cancer tumours both at high frequency.
negative or inconclusive results, there was a sense that, like the family above, mutations would be revealed in time.

*Disappointment and Unpredictable Results*

Although staff had no way of manipulating the outcome of the test results, the staff expressed joy when they thought their client had a strong chance of having a mutation found and, on the contrary, they also expressed disappointment when they found out that they did not. Disappointment was expressed when the genetic counsellors felt they had missed an opportunity to be in charge of a case where they were sure a mutation would be identified, and also when results did not go as they had predicted. Sometimes their disappointment in not finding a mutation, regardless of employing all possible attempts, was expressed. In one instance, the clinicians stated: “we have thrown the book at them and not found anything”. The “throwing of the book” analogy meant that even though the laboratory (with the prompting of the clinicians) had undertaken all possible genetic tests, no information was gained to help the clinicians understand, interpret or explain why particular clients with extremely high numbers of cancer diagnoses in their families were not found to carry familial cancer related mutations.

Just as excitement was encouraged, the display of negative attitudes was discouraged because it was not in keeping with the optimism of the Unit. It was also not in keeping with the optimism that exists in the wider genetic community (Reuter & Neves-Graca, 2007, p. 135). Often when a clinician informed their colleagues of there being a strong chance of finding a mutation and another disagreed, arguing that they would not find a mutation, they were told not to be “cynical” or “negative”. A genetic counsellor, who was particularly known for being cynical and negative, once stated that her negativity helped
fellow clinicians have mutations found in their clients. Upon further explanation, she described how when they believed that a mutation would be found, that was when one was not, and conversely when clinicians thought they would not find a mutation, that was when a mutation was found. I had heard other similar statements made by the genetic counsellors, including that unexpected outcomes were “the way of the world”. This indicated that a belief among some of staff existed around the view that scientific results did not always occur in expected ways – that science was unpredictable. This idea is supported by Marcum (2010, p. 202), who purports that experiments in science may inadvertently generate results that are unpredicted and that it represents the extent of theories to account fully for the results. The reality of genetic testing for familial cancer meant that no mutations were found the majority of time. As the clinicians could not explain, using their current knowledge and theories, why their mutation test results did not go as predicted, they sometimes instead expressed a superstitious belief that universal fate was at play. This view considered it possible to cause bad luck for themselves (the opposite result to that which was predicted) by talking too confidently about predicting a mutation find and that this could be counteracted by speaking pessimistically about the chance of finding a mutation.

The various roles and functions of these behaviours can be categorised as both reflecting and resulting in four main things: first, coping with uncertainty through competition and separation; second, individual and collective capital; the third, learning, predicting and testing theories; and fourth, the possibility of minimising and/or preventing of familial cancer. I will discuss each of these aspects in turn.
Coping With Uncertainty Through Competition and Separation

Finding any mutation was seen as significant and a cause for excitement because of the rareness involved in finding a mutation. The majority of the time, genetic tests were performed on individuals who met the criteria and in whom staff believed a mutation would be found. One of the clinical geneticists once explained to a medical student (on placement in the Unit) the difference between the Unit and the general genetics unit. The clinical geneticist emphasised that the general genetics service was “different” because it focused on diagnosis, whereas they (the Unit): “do not expect to find a mutation in our families, we expect not to find a mutation in our families”. This statement demonstrated how there was an expectation by the clinicians that they would not find a mutation. Therefore, when they found a mutation, it was particularly special because overall it was relatively rare. Excitement in the Unit was therefore influenced by the nature of the difficulties and the uncertainties inherent in this particular area of familial cancer genetics. The regular practices, the rituals, demonstrated their commitment to future diagnostic certainty. Another way that the clinicians coped with the uncertainty surrounding mutation results involved them separating the gene from the individual via competition. As stated earlier, all individuals were considered as being made up of genes, yet genes were separated and separable from the individual client, and so competing and betting over a mutation, did not represent the individual human being.

Individual and Collective Capital

I was informed by one of the clinical geneticists that a mutation positive result was “exciting” because it provided recognition that they, as clinicians, were “selecting people properly”. The betting and competing fostered team spirit and staff comradery which contributed to the collective capital of the group. Individuals also gained capital through
peer recognition of their achievements, while asserting knowledge and dominance. With every bet and competition, the identification of which clinician won also reflected the teams and hierarchy of the Unit – although all staff members could participate by taking sides, only the clinicians obtained value and reward through participation in the game. The results of the mutation tests informed all staff, not just the clinician involved, if a mutation was, or was not found, and who was right and wrong. Winning demonstrated who was more able to select and predict accurately, using their clinical judgement, in a game involving uncertainty.

*Learning, Predicting and Testing Theories*

With no staff member of the Unit undertaking any genetic testing in the laboratory, practices of betting and competing over mutations could be viewed as a way of the clinicians being able to predict, and test their theories and justify their scientific knowledge. The finding of mutations was exclusive and empowering for the clinicians as they were at the forefront of the discovery process having selected which clients could have genetic testing. As explained, although all mutation discoveries were valued by the clinicians, the rare, the complicated and the deadliest mutations were most valued.

The clinicians’ work was based upon and composed of theories and hypotheses developed and proposed by past mentors and other members of the scientific/genetic community (Marcum 2010, p. 202). The attribution of different values can be linked to what was discussed previously in regards to the excitement that revolved around the scientific discovery of new knowledge to be found in the context of science and medicine. By discovering and accumulating new, rare and unique mutations, the clinicians were accounting for a condition, part of building upon those theories and collaborating with
other esteemed members of the scientific/genetic community through publications or presentations. It was about being part of an internationally expert team in identifying this new mutation and publishing it first – thus being at the forefront of the discovery process and making the unknown known. By identifying, publishing and sharing the information obtained, they were part of the horizon for scientific progress as they moved the science from one state of knowledge to another, a concept said to be critical for the extension of the field and for future understanding and scientific progress (Marcum 2010, p. 205).

The Possibility of Minimising or Preventing Familial Cancer

Discovery of mutations was not only about the achievement of finding another mutation, or the possibility of uncovering something new and/or rare. As discussed in Chapter 6, the clinicians, in their view, also used the results to possibly reduce or prevent more cancers, thus improving and/or saving the lives of their client and/or the client’s biogenetic relations. The finding of a mutation in their clients and/or their biogenetic relations provided clinicians with a sense of achievement because it offered greater knowledge, access to services and closure. This view reflected the broader understanding within genetic discourse – that genetics holds the key to solving and/or eradicating a range of human problems (Reuter & Neves-Graca 2007, p. 134).

In Chapter 5, I wrote about how clients were said to have wanted a simple answer as to why they experienced cancer in them or their family members; asking the clinician whether it was due to the HRT they took, an accident, exposure to chemicals or stress. The genetic counsellors informed their clients that their experience of familial cancer was beyond what they (the client) did. Staff of the Unit enjoyed finding mutations, as through the identification of a mutation they were able to provide, to the client, an explanation
backed by proof of a mutation diagnosis. The mutation provided physical evidence of a hereditary explanation for the experience of familial cancer (using biomedical genetics knowledge), in an area that is acknowledged as ever-changing and fraught with uncertainty and unknowns.

**Excitement, Objectification and Discovery in Genetics**

As explained throughout the thesis, the prediction, finding and sharing of mutation discovery was seen as significant and the cause for excitement because it was understood to be empowering for the individual client and the clinicians involved. It was empowering because mutation identification equalled greater confirmed knowledge and justified access to services. Excitement over the identification of mutations was wrapped up not only in the possibility of helping clients but in excitement around the process of scientific discovery and advancement. Various examples have been provided above, demonstrating the different practices, roles and functions of betting, competing, and displays of excitement surrounding the discovery of mutations.

The new genetics, built upon the HGP, re-produces the idea that science and medicine have no boundaries and that, with time, human suffering will be dealt with and solved by scientific and medical advances (Leslie 1980, p. 191). The new genetics is believed by some to hold the key to solving and/or eradicating a range of human problems, not only involving the identification and eradication of minor and major disease (such as cancer). Research involving the genetic basis of disease is being used to explain, with the view of eradicating, everything from health problems (Petersen 2006, p. 481), criminal and deviant behaviours (such as alcoholism) (Reuter & Neves-Graca 2007, p. 134; Spallone

The promises and claims made about the mapping of the human genome have been argued as highlighting a utopian view of health (Hubbard & Wald 1999, p. 164; Lock 2002, pp. 240, 241). The idea that civilization has moved, and is progressing, in a desirable direction through the manipulation of knowledge has been termed the “myth of progress” (Gray 1999, p. 27; Niiniluoto 2011, section. 2.1). The notion of extending knowledge and progressing scientific and medical knowledge, with its associated advances, through the discovery of new mutations, was expressed by the clinicians of the Unit. This was reflective of, and contributed to, practices in the wider genetics community.

*Horizon for Scientific Practice*

James Marcum (2010, pp. 187, 191) applied the notion of Horizon for Scientific Practice (HSP) to account for the justification process and the scientific practices employed to facilitate scientific discoveries and progress science. The notion of the horizon refers to the boundary between the land and sky and involves two areas. The first area constrained by the horizon’s boundary is made up of views and objects that are mostly known and familiar to the viewer, while the second is the new area that opens up as the viewer moves towards the horizon and its boundary shifts. This new second area, although connected to the first, mostly contains the unknown and unfamiliar and holds the promise of more new views as the viewer moves towards the horizon (Marcum 2010, p. 191). In drawing connections to scientific experiment and discovery, the metaphor of the horizon is the boundary where scientists investigate and understand the world, and it signifies a
potential for more research and understanding (Marcum 2010, p. 194). The goal of scientists when conducting their experimental or theoretical activities on a day to day basis is to make the “unknown known” and the “unfamiliar familiar” so that they can understand, explain, manipulate and control the findings (Marcum 2010, pp. 191, 192). By doing so scientists not only advance their understanding of the science (as the horizon recedes) but each activity undertaken potentially holds the possibility of major discovery and progress (Marcum 2010, p. 194).

Using Marcum’s metaphor of the horizon, after scientists observed, recorded and/or collected the results of experiments, they either formulated new theories or revised their current held ones, thereby increasing the base exposed as the horizon recedes (2010, p. 198). In a similar manner, the clinicians of the Unit used the results of the genetic tests to inform them of whose clinical judgements were correct and who needed to modify their clinical judgements. On a day-to-day basis, the clinicians incorporated knowledge and theories along the way, which led to the gradual growth of their medical and genetic knowledge. Through betting, horizons were shifted as scientific progress was made by replacing their existing knowledge and/or incorporating new knowledge as a result of the outcome of the game. This goes part of the way to explain the excitement demonstrated by staff of the unit. Yet there seemed to be some level of objectification of clients rather than empathetic connection, which relates to broader issues in bio-medical culture.

Objectification and Commodification of Genetic Material

Research, predominantly by anthropologists, sociologists and psychologists, discusses how bio-medical culture, in general and specific specialty areas, objectifies patients. Building upon a Cartesian derived perspective of mind and body, as outlined in the
introduction of this thesis, clinicians are primarily concerned with the physical body rather than the embodied patient as an experiencing individual (Barrett 1996, pp. 12, 85, 242; Bourdieu 1977, pp. 91, 218; Chambliss 1996, pp. 21, 26; Foucault 1973, p. 5; Jaye 2004, p. 41; Merleau-Ponty 1962, pp. 66–91). Scientists and clinicians working in the area of the new genetics focus on the genetic material, the DNA, and the genes and mutations said to exist in all people. Through their work there is a separation between the DNA, the gene and the person. Advances in biotechnology subject the body to fragmentation, and so, with new forms of property and property relations being formed, these advances raise debates about medical privacy (Rabinow 1992, pp. 169–187; Strathern 1998, pp. 214–232; Sunder 2006, pp. 12, 67, 153).

Commodities can be simply defined as objects of economic value (Appadurai 1986a, p. 3). A broader concept of commodification defines it as encompassing all capitalized economic relations between humans in which human bodies are the token of economic exchanges that are often masked as something else – love, altruism, pleasure and kindness (Scheper-Hughes 2002, p. 2). Commodification of the body and its parts requires objectification in some form, transforming persons and their bodies from a human category into objects of economic desire (Sharp 2000, p. 293). In Sharp’s examination of anthropological discussions and understandings of body commodification, it is argued that the human body, and its parts, have historically been a target for commodification across various cultural settings. These include oppressive labour practices like slavery to female reproduction. It is argued that a paradigmatic shift in the anthropological understandings of the commodified, fragmented body, is occurring in the application of biotechnologies and other related scientific arenas of reproductive technologies, organ transplantation, cosmetic and transsexual surgeries, genetics and immunology, and the
Pulling the above arguments together, the objectification and commodification of human genes is economically valuable, and is clearly evident in the practice of storing, immortalizing, reproducing and engineering which are masked in the guise of medical education.

**Genetic Material Markets**

In Chapter 6, I discussed the uncertainties regarding the current and future ownership of genetic material. Andrews and Nelkin expose the global debates found in lay and professional writings over the ever expanding markets for the taking, use, distribution, genetic testing and patenting of human tissue. It is in these markets where the body is reduced to a “source of raw material for salable products” (1998, p. 53). The locating of relevant mutations on the human genome are often directly linked to biocapital (Lock 2012, p. 143). It has been argued that the commonplace practices of patenting genes and living organisms, the production of new classes of drugs, billion dollar endorsements, and the commodification of biological materials as valuable and marketable are ruled by the global economic growth of the billion dollar biotechnology business (Nelkin & Lindee 2004, pp. xv, xxiii–xxv; Palsson 2007, pp. 150–175). Termed the “human genome industry” (Palsson & Rabinow 1999, p. 14), the most efficient means of advancing knowledge, particularly with genetic sequencing, is through the partnerships between the market and biotechnology (Palsson & Rabinow 1999, p. 18).

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59 According to Sharp (2000, p. 311) a cyborg is “an amalgamation of human, animal, and technological parts”.

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Health and Wealth

Through a relationship of health and wealth, medicine has an alliance with government which is distributed across multiple sites, found in the clinic, home, health and social policy, the law and the Internet (Latimer 2013a, p. 40). Both private and public funding is dependent on the partnership where private and public health agendas are, or can be, shown to be addressed. Such agendas require the pursuit of knowledge that will be useful in improving health (Latimer 2013a, p. 41). It was apparent, in the Unit, that the value in the commodification of biogenetic materials also occurred at the local level. As identified when I discussed “Coming to Be: Historical Background and Present Practices of the Unit” (Chapter 2), the predicted economic savings of the service were a driving factor for the service’s establishment and continual funding, with the costs involved with the running of the Unit being offset by the potential savings the Unit could generate through its services.

A Relationship Between Science, Biomedicine and Genetic Medicine

The term “medico-clinical” has been used to describe and emphasize the “inseparable relationship that exists between contemporary cosmopolitan, biomedical practices and associated forms of scientific research and knowledge” (Sharp 2000, p. 288). Assuming a range of forms, the “medico-clinical fragments” transform living bodies into scientific work objects (Sharp 2000, p. 298). The medico-clinical realms require a range of human body parts including cadavers, skeletons, blood, organs, transplantable tissues, microscopic ova, sperm and genetic material (Sharp 2000, p. 289).

Niiniluoto (2011, section. 2.1) describes science as a complex multi-layered system involving a community of people (scientists) who use scientific methods to produce new
knowledge. The notion of science can refer to various aspects: a social institution, the researchers, the research process, the method of inquiry, and scientific knowledge. Within each of these aspects, different types of progress can be distinguished including: economic (increased funding of the research), professional (increased position of the scientists and their host institutions), educational (increased skill and expertise), methodical (new methods or the refinement of research), cognitive (advancement in scientific knowledge), technological (increased effectiveness of instruments and techniques), and social progress (increased economic wealth and/or quality of life).

A number of popular theories of scientific progress or advancements view science as either a “backward-looking or forward-looking” goal-related concept (Niiniluoto 2011, section. 2.4), as always including truth and information (Levi 1967, p. viii; Popper 1959, p. xxii; Popper 2002, pp. 6, 7), as having explanatory and predictive power (Hempel 1965, p. 117) and including accuracy, consistency, scope, simplicity, and fruitfulness (Kuhn 1977, pp. 329, 332, 339). As part of an analysis of science, philosophers have also made various distinctions between the discovery of scientific hypotheses or theories and their justification (Marcum 2010, p. 188). For Allan Franklin (1993, pp. 2, 127), this involved examining not only the role of experiments in the process of scientific progress or discovery but the justification of the experiment.

There are some theorists (including Rose 2007, pp. 10, 11, 50) that consider science and technology as posing challenges to medical authority and diminishing the power of doctors, because of patient consumerism, the widening access to medical information and knowledge, and the abundance of technological and scientific knowledge. Counter to the argument which views the new genetics as eroding medical dominance, Latimer (2013a,
p. 45) considers medicine as “medicalizing science”, arguing that medicine and science interact and have a relationship that is both co-constitutive and interdependent. Instead of considering these domains as competing technologies that are undermining clinical authority, Latimer considers them as “medicine’s intermittent allies”, magnifying, enhancing and reproducing its dominant position (Latimer 2013a, p. 204). Medicine’s power is extended in the clinic, by using science (genetic medicine) to intensify its medical authority, its power, so that it is both biological and social (Latimer 2013a, pp. 196, 197). The power and dominance of genetic medicine is displayed when medicine and science work together on the unknown, sometimes classifying and sometimes deferring on genetic diagnostic categories (Latimer 2013a, p. 191). One example provided by Latimer explains how DNA testing results required clinical interpretation as to whether or not the test results were accurate and what they meant. Clinical opinion was required by people, insurance companies and employers. Genetic medicine stems from the work of both science and medicine and is considered an ambiguous new frontier where the unknown, uncertain and non-standardised, works alongside a firm commitment to future diagnostic certainty (Latimer 2013a, p. 202). Where there was uncertainty this legitimated the need for more investigations and more clinical judgement in the future (Latimer 2013a, p. 201), a practice evident in the work of the Unit. This concept of new frontiers and future diagnostic certainty was also clearly demonstrated in my fieldwork through a major familial cancer conference.

Peer Gathering: A Conference of Competitiveness, Enthusiasm and Advancement
As previously mentioned, competitiveness and enthusiasm over the discovery of mutations also existed outside of the Unit, and was propagated and shared by colleagues who made up a larger social word of the familial cancer genetics community.
I attended an annual four-day interstate conference on familial cancer research practice held in a secluded beachside hotel, which essentially required attendees to stay in the same hotel. The conference focused on research and practice within the area of familial cancer, and involved the gathering of a likeminded “close knit” group (a few hundred) made up of geneticists, clinicians, surgeons, biologists, genetic counsellors, nurses, psychologists, oncologists, pathologists and epidemiologists who were predominantly from Australia. From the Unit, the two clinicians, three of the four genetic counsellors and the two research nurses attended. With presentations held throughout the days and dinners held throughout the evenings, the conference was a chance for all attendees to mingle and network.

While attending the conference, competitiveness, enthusiasm and excitement over mutations were displayed in various ways and forms. The conference largely involved presentations that were held throughout the day, focussed on describing the “ground-breaking research” that was being conducted in order to discover more mutations. The majority of the presentations revolved around genetic research and advancements and were presented by geneticists, scientists and specialists. Fewer presentations were defined as “psychosocial research”, and were presented by research nurses, genetic counsellors and psychologists held at the same time as the clinical non-defined presentations. Being defined, on a smaller scale and in competition to “scientific” presentations, it separated the doctors and scientists from other health workers, and reflected and propagated the imbalance of psychosocial research in the area of genetic medicine.
Peer Recognition through Rewards

The conference committee provided a large trophy (adorned with a microscope) to two separate male presenters who were awarded for the significance of their work and contribution to the scientific community. The first awardee presented research on a large case involving FAP mutations. With the mutation originally discovered in a Utah family, the family was made up of 5,000 members and connected to mutations found in families across America. The family also extended seven generations (going back to England in the 1600s) and was said to be responsible for one per cent of all known FAP mutations. A photograph was shown which depicted staff laying out hundreds of pedigrees printed out in a line and stuck together along the hallway of their offices in order to indicate just how long the family connections were that they had pieced together. Upon seeing this photograph, a number of attendees collectively gasped, which conveyed how impressed they were at the scale and size of the operation. The award and reactions demonstrated that value was ascribed and rewarded for persistence, thoroughness, tracing and tracking of mutations across families and generations, through time and places.

The Difference between Driving a Car and an Aeroplane

It is clear as noon-day, that man, by his industry, changes the forms of the materials furnished by Nature, in such a way as to make them useful to him. (Marx 1967, p. 319)

The second presenter awarded and was promoted by conference delegates as being a world-renowned genetic pathologist that attendees were fortunate to observe. After being presented with an award for his contributions to science, the presenter spoke confidently about the “new era” of using Next Generation Sequencing (NGS) machines to find mutations. A belief in the power of the genomic sequencing machines stems from recent developments where it is believed that such machines will extend the horizon of genomic
biology, discovering new genes and their interactions in developmental and pathological processes (Gene Ontology Consortium 2000). Traditional cancer genomics was compared to the improved way of obtaining results through the new NGS models, saying it was the: “difference between driving a car and an aeroplane”. This reflected the increased complexities and capabilities of the NGS machines compared to the traditional practice of cancer genomics. The presentation was a mixture of things, displaying complicated technical graphs and using scientific explanations, aimed at those with the most extensive training and knowledge in the area, alongside lay, simple language and examples that someone completely new to the area could follow.

The focus of the presentation was to convey and encourage advancement in genetic knowledge via the latest NGS machines. It was described how too many researchers in the area were publishing and arguing that the NGS machines had little clinical use because they were producing information that was not able to be translated. The presenter adamantly argued that because cancer is complex, the interpretation of results was like “wolves in sheep’s clothing” and that “we need to be able to translate the results of NGS and not be afraid to use them”. NGS, it was said, was going to provide “a new lens on cancer” and would be like “the invention of the telescope”. Various pictures were displayed on a large screen during the presentation and one depicted an iPod with a genome sequence on the screen. It was explained how the next step would be to incorporate one’s personal genomic information into handheld technology, with the argument that patients would soon demand access to this technology. This argument represented the speaker, as a scientist, pushing for his fellow colleagues to interpret the currently uninterpretable information produced from the NGS machines, and pushing for it to be done in order to meet future consumer trends.
Upon completion of the presentation, the audience was asked for questions. John, from the Unit, was first to question the presenter over the usefulness of the NGS because of John’s reservations about the ability of clinicians to interpret the results. John stated that NGS remains an insufficient technology due to misinterpretation, and that they (as specialists) “risk being bamboozled by the power of proteomics”.60 The speaker responded that they (as clinicians) should not view the current interpretation issues as a negative feature and that the reason they all got into the profession as educators and specialists was to help people, before saying that it was “time to take some risk and walk the plank as the others will lie the next plank in front”. The presentation by a leader in the field, who admitted the technological limitations but encouraged advancement, demonstrates Latimer’s argument of a commitment to future diagnostic certainty (2013a, p. 202). This reflected the overarching system of belief that genetics should be progressive, and displayed the power, recklessness, encouragement and hope that is ever present in the field of mutation identification. Thus it also supported the notion of horizon progression, with the belief that there will be another plank, another horizon to light the way.

Other questions posed conveyed anxiety over the NGS technological advancements and the increase in affordability of the NGS machines. Some clinicians felt the use of such machines would increase the by-pass of genetics services, where the public could prepay and order more detailed information (already available at the time of this research) about their whole genome online and receive back genetic information that had not been

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60 Proteomics is the research into protein structure and function (National Collaborative Research Infrastructure Strategy (NCRIS 2011)).
interpreted to the user. This practice would take away clinicians’ power over deciding who gets tested for what and the opportunity to test their own clinical capacities, while also removing the ability of clinicians to relay the possible impact of the information and choices to the purchaser. This demonstrated a contradiction for clinicians who, on the one hand, wanted to advance the knowledge surrounding genetics and share that knowledge but, on the other hand, were wary of losing the ability and the power to be in control of information interpretation and provision. There is a current argument that views the demise of medicine’s gaze and authority with the expansion of techno-scientific medicine (Rose 2007, p. 11). Latimer points out that genetic results from online do not really matter to insurance agencies or help explain work absences because genetic tests require the sanction of clinical interpretation and opinion. The clinical interpretation and opinion is medicine’s authority and so rather than science and technology taking over medicine, the new genetics is arguably reinstalling medical dominance in the clinic, as the location of discovery (Latimer 2013a, pp. 196, 197).

The Biggest Fish and the One that Got Away

Group discussions held by conference delegates after seminars, during morning and afternoon tea, and over lunch and dinners, involved descriptions of one another’s work, and comparisons with each other’s work. Stories that were shared incorporated the discoveries that had been made and predicted discoveries that were soon to be made. From my observation, some stories were reminiscent of the retelling of fishing tales, but instead of emphasis being placed on the size of the fish caught, the type of mutation and the size of the families it affected were emphasised. There were also stories shared about years of research having disappointingly not produced the answers they were hoping for and these were reminiscent of fishing tales about the “one that got away”. Regardless of
the outcome, hope and optimism was apparent as they had nonetheless learnt in the process. This reflected a practice and belief that familial cancer genetics was about teetering on the edge, a part of practices where the next major discovery was sure to come.

The Latest, Fastest and Best Equipment

As explained earlier, genetic research is driven by money. The conference provided a chance for the newest and best equipment to be spruiked and sold. Representatives of the latest NGS machines had displays advertising their products lining the entrance hall to the conference rooms (see Figure 17, next page). The displays contained information posters and brochures, and attendees were encouraged to attend related presentations to receive a free gift (for example, in one presentation, the gift was a drink bottle embellished with the company’s name). During the presentations, representatives from the companies emphasised how their machines were better and faster in their ability to sequence whole genomes and to find mutations. They stressed that genetics could not progress without them, thus encouraging their purchase and commodification.
Figure 17. Photographs of company displays advertising new technologies and equipment.
A Public Display of Clinical Practice and Medical Emphasis

As part of the conference, a two-hour poster session evening was held in the hotel’s public main foyer (see Figure 18). The evening involved five-minute discussions held in front of eight of the 46 posters, with the theme title: “What is being practised in the clinic”.

Waiters weaved in and out of the crowd, offering free wine and cheese to attendees who read about and discussed the latest practices. The visual and verbal display of knowledge during the poster session, and throughout the conference, enabled the propagation, continuation and celebration of science and genetics amongst likeminded insiders – made up of clinicians and researchers. The purpose of the posters was to inform and update each attendee in what was being practised, researched and achieved in genetic clinics.
Just as anyone with Internet access can view the complete sequence of the human genome online at http://www.ncbi.nlm.nih.gov/genome/guide/human/ (National Center for Biotechnology Information (NCBI)), 2002 it did not seem to matter that the posters were on display (and had been the whole day), in a public place where other hotel guests walked through the lobby. It did not matter because the posters contained information using specific language (the same language used in the medical texts in the bookshelf of the Unit), requiring specific education – insider knowledge to understand and decipher. With genetic language and diagrams used on the posters to describe the practice and results of genetic clinics and research companies, the clients and families, whom the information was drawn from, remained absent. This illustrated the objectification and a separation between the DNA, the gene and the person.

Conclusions
Local and global competitions and practices were drawn upon in this chapter in order to situate the Unit’s practices of competing through betting, and being excited over mutations, in the broader context of scientific discovery and progress. Competition through the practice of betting and excitement over the finding and identification of mutations was regularly displayed by staff of the Unit, expressed in numerous ways that were sometimes explicit and other times implied. I have demonstrated that the purpose of the practices and how they were expressed varied depending on the context, time and space. The clinicians of the Unit regularly exercised their competitive nature and expressed excitement for various reasons, which included: gaining individual prestige, as a form of recreation, to display power and knowledge, as a team building exercise, as a coping mechanism to manage the stress of their work, and to reflect and/or engage the interest of their clients.
I have argued that there was some level of objectification and commodification surrounding a client’s genetic material and knowledge. This was largely expressed through competition and betting which, whether occurring in the Unit or on a wider global scale, was both collegial and competitive, serving numerous purposes. I argued that the ownership, sharing and competition surrounding genetic material and information did not represent the individual because the genetic material and information obtained had been physically removed and emotionally separated by such practices. Selected examples from a conference involving the gathering of a “community” of medical clinicians and scientific researchers were also analysed. Such community gatherings functioned to increase collaborations and connections towards common goals, but they also fostered rivalry and competition – all of which facilitated building knowledge, as well as identifying what was individually and collectively known and unknown. Locally, the work of the service functioned in a similar fashion, but the clinicians of the Unit used and shared selected knowledge, competition and uncertainties with their clients in clinic and beyond the clinic. This demonstrated some of the ways in which scientific discoveries and, in particular, knowledge about familial cancer risk and mutations (including the possible implications), were shared in the Unit and more broadly, in contemporary western biomedical societies. In the concluding chapter of this thesis, the key findings and arguments from the research are presented, along with the important implications and considerations.
Chapter 9. Conclusion: Potentialities of Familial Cancer Risk

In biomedical practices, potentiality indexes a gap between what is and what might, could, or even should be. (Taussig, Hoeyer & Helmreich 2013, p. 5)

This ethnographic thesis has demonstrated and discussed contemporary familial cancer risk production, dissemination and prediction in a familial cancer genetic counselling and testing Unit. The Unit, through its complex work of familial cancer risk identification and classification, emerged as a site of clinical biomedical prediction, intervention, and knowledge production and dissemination. All of the staff members of the Unit were united in the belief that all individuals were made up of things called “genes” and “mutations” which were considered, on the one hand, as a defining and unique part of the individual, and, on the other hand, as shared with the individual’s biogenetic relations. Certain mutations in genes believed to be responsible for an increased risk of familial cancer were also considered to exist both in an individual’s genes and in the genes of particular biogenetic relations, which resulted in various complexities, considerations and negotiations by the clinicians and clients.

With the majority of my time spent in the Unit, observing staff members, I was very fortunate to observe and learn about an area that was not normally accessible to outsiders. During fieldwork, I largely observed and heard about the benefits of identifying familial cancer risk. Terms such as “familial” cancer, involving and affecting “families” and “family members”, were regularly used. However, genetic relationships were key to the practice of familial cancer risk diagnosis and risk notification despite genetic relationships not necessarily constituting “family” for many people. I have demonstrated the different ways in which the prediction, explanation and communication of familial
cancer risk, including the practice of risk notification, was complex and uncertain, which required and resulted in ongoing judgement and negotiation by staff, and in particular the clinicians. Their ongoing work with uncertainty demonstrated a continued commitment to developing the science and the potential for certainty, an argument also described by Latimer (2013a, p. 200).

Medical anthropologists such as Good (2001, pp. 400, 403, 407), Lee (2013, pp. 78, 84) and Mattingly (2010, pp. 3, 4, 5, 6) have commented on various ways in which uncertainties produce hope for both patients and for their treating clinicians. This thesis adds to this work by demonstrating how information about familial cancer was sought and imparted by the clinicians and their clients, for its perceived usefulness – for its potentiality. The clinicians provided all of their clients with information about their familial cancer risk and, more importantly, about what could be done to reduce or prevent their risk of familial cancer. The clinicians viewed this information provision as potentially enabling the reduction of the risk or severity of familial cancer-related diseases, through genetic testing and risk-reducing and/or preventative approaches while also saving the government money and avoiding possible legal responsibility.

The clinicians, and some clients, considered and emphasised that the information was potentially lifesaving, a “golden opportunity” rather than the information being a threat. However, there was often a schism evident between clinicians’ needs, and their clients’ needs, for mutation knowledge. The clinicians hoped for better understanding of familial cancer and of health and disease, in order to help in the prediction, reduction or prevention of familial cancer. Clinicians also hoped the information would help contribute to finding the eventual cure for cancer, through personalised medicine which,
they assured themselves and their clients, would be coming. The clinicians sought to
discover a mutation as part of their striving for scientific advancement and progression of
knowledge. They then shared the value of their persistence and thoroughness with each
other and their clients, which they considered as having personal, individual, familial and
societal benefits.

Clients and families also wanted to explain and prevent the experiences of cancer in
themselves and their family members. The majority of clients, however, sought certainty
and explanation through genetic testing, wanting to know if the cancers experienced in
them or their relations were due to an inherited mutation. For clients, the need to know
their future risk of familial cancer, including when, where and in whom it might strike,
and whether and to what extent they should engage in risk-reduction measures, was tied
to mutation confirmation. Clients wanted certainty, but even when a mutation was
identified, confirmation and certainty about whether a client would experience familial
cancer was not something the clinicians of the Unit could provide. Rather, the clinicians
provided a risk estimate – an ambivalent notion that caused and supported ongoing
negotiation about what it could potentially mean or not mean.

Other concerned stakeholders, such as governments, insurers and employers, sought to
predict and prevent future liabilities and to predict. My research revolved also around the
potentialities of my research, as my scholarship was awarded because of my potential
contribution to the area. In terms of familial cancer risk, the Unit was funded and the
clinicians worked to identify familial cancer risk, and associated mutations, because of
the positive potential for cancer reduction or avoidance and, thus, economic savings.
However, as demonstrated in this thesis, there was, and are, other potentialities stemming
from this practice. As Taussig, Hoeyer and Helmreich (2013, p. 8) argue, potentiality is ambiguous “suggesting possibility, danger, and desire all at once (or in turn)”.

Familial cancer risk estimates and the genetic mutation results only offered information about probable future risk, resulting in problematic knowledge. Genetic testing and genetic knowledge involves “pragmatics of uncertainty” (Konrad 2005b, p. 145). On the one hand, the clinicians of the Unit appeared comfortable with the uncertainty surrounding familial cancer, practising regardless of the unknowns because of a belief in the benefits of what was known. On the flip side, the clinicians worked to gain certainty, demonstrating power by managing the uncertainties by predicting, betting, debriefing, and sharing the unknowns and complexities with some clients to varying extents.

I argued that the local competition reflected the uncertainty and propagated similar practices as found in the Human Genome project (HGP) and medical research more broadly. For the clinicians of the Unit, the identification of a mutation, and in particular, a mutation considered unique or rare, was exciting because it had both personal and broader potentialities. The clinicians were enthusiastic to be involved in the prediction of familial cancer risk and the discovery of a mutation because it equalled new knowledge and opportunity. The competition and excitement displayed by clinicians was linked with the rarity of finding a mutation and the importance of contributing to and progressing the individual client’s case, the familial case, and knowledge of familial cancer and genetics more broadly. The regular friendly competitions were a way in which the clinicians handled the uncertainties of their work, while reflecting and contributing to staff comradery, humour and the identification of the clinician with the most accurate clinical judgement and prediction.
I have argued that regardless of the preconceived knowledge about genes and familial cancer risk that a client had prior to undertaking genetic counselling, clients were impacted by which clinician did the informing and the extent and way in which the clinicians informed their clients about what was known and what was not. These factors impacted on how the client experienced genetic counselling and, ultimately, how they considered themselves as at risk of familial cancer, or not, and the associated physical, emotional, psychological, social and economic implications of such knowledge.

My participants held many varied beliefs. Some anthropologists have argued that although beliefs are real phenomena, they are held in the minds of our participants and are thus inaccessible to anthropologists; ignoring this leads to a crisis in representation (see Dein 2003, p. 149; Dein 2007, p. 49; Good 1994, pp. 25, 45; Needham 1972, pp. 2–6). Dein (2007, p. 44) suggests “that it is the expert who oversystematizes a society’s way of life”, that people hold bits of information, which may be conflicting, and incoherent explanations of illness, or belief systems which are oversimplified and presented in some ethnographies. Explanations do not function as a whole but in bits (Bloch 1998). As an anthropologist, I experienced participants’ articulated and displayed beliefs and knowledges sometimes coherently, sometimes as fragmented, and sometimes as contradictory and confusing. Thus, it would be inaccurate on my part to present participants’ belief systems as uniform and always logical. The knowledges and beliefs they displayed were contextual and often contradictory, with staff and clients believing in one thing in one context and not necessary believing in the same thing in a different context. Sometimes the clinicians performed certainty, and at other times uncertainty when categorising familial cancer risk or interpreting results, a finding consistent with that of Latimer (2013a, pp. 8, 198) and of Seale, Pattison and Davey (2001, pp. 60, 77).
Future Research and Thesis Contributions
Genetics is changing medicine and changing society but it is not a unidirectional relationship as society also impacts on the acceptance of genetics and its associated practices. The promise and hype surrounding the new genetics is enormous. Lock challenges medical anthropologists’ to “confront and critique the obvious limits of narrowly defined deterministic genetics and epigenetics” (2012, p. 160). With the production of faster, greater and more assessable NGS machines, along with consumer demand, the practice of genetic testing is expanding. As a result of the expansion of genetic testing, biological explanations of illness have become further entrenched in comparison to cultural, psychological and social conceptualisations of illness (Conrad & Barker 2010, pp. 67, 77). However, instead of genes being considered as “absolute predictors”, they should be considered as embedded in complex biological, social and economic networks and relationships (Hubbard & Wald 1999, p. 12).

In this thesis, I have shown that clients of the Unit considered at increased risk of familial cancer were understood or constructed as ill via the object of medical attention, which required them to fight against the possibility of cancer. Jacob (1998, p. 102) states “we are all virtual carriers of some illness because of our predispositions and our life habits”. In building upon this thesis, although familial cancer risk is only being identified in a small number of members of the population, the new genetics will only increase in its grasp of genetic disease prediction, which will affect us all – some sooner than later. Whether people should be informed about a prediction of disease should be more openly and widely discussed and debated now. The identification, provision and communication of familial cancer risk will continue to have important cultural, ethical, social and psychological implications. I have shown that there are difficulties in, and arguments for
and against, knowing and not knowing. People have the right to be informed of being possible carriers of a mutation that predisposes them to familial cancer and, similarly, individuals have the right not to be informed about this. This thesis has illustrated the difficulties in achieving the fundamental biomedical ethical principles of autonomy, beneficence, justice, privacy and confidentiality. As discussed in the thesis, the sharing of such information relates to the commodity and property of biological materials and information. Everett (2007, p. 383) argues that a tension exists between the “collective and individual rights to things”. Such a discussion should center around who does, and should, own and share biological materials and the information derived from them.

This thesis has critically analysed the social understandings, the practice and implications of familial cancer risk, which directly reflects the present and continual ambiguity surrounding the issues. Genetically testing those whom have not suffered from cancer, are not sick with familial cancer, and may never be sick, in order to establish their risk, raises important questions about individual, family, community and socio-cultural rights and responsibilities. The opportunity, limitations, possible and actual implications, and the ambiguities of familial cancer diagnosis need to be openly accessible, shared, discussed by all community members, and used to inform guidelines and policy. Overall, further ethnographic research on micro and macro levels needs to question the role of genetic technology and the determinative nature of genetic knowledge. In relation to new technology, such as NGS machines, research should focus on the social implications of their application and results, how they are incorporated, understood, managed, provided and explained by clinicians in the clinic and how clients manage receiving multiple variable predictions for different illnesses. Future research on the determinative nature of genetic knowledge should follow and document how uncertainties, tensions and interests
are understood, expressed and negotiated, which would lead to better understandings of
the cultural, economic and legal implications of contemporary genetic knowledges and
practice.
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