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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Contents

Declaration ........................................................................................................................................... 3
Related Thesis Publications ................................................................................................................. 4
Acknowledgements ............................................................................................................................. 5
Abstract ............................................................................................................................................... 10
Important Notes ............................................................................................................................... 12

Chapter 1. Situating the Field: Introduction and Background ......................................................... 13
  “It’s all in the Genes”…. or not? ..................................................................................................... 13
Biomedicine and the Hospital ........................................................................................................... 17
  The New Genetics and Preventative Medicine ............................................................................. 22
  Implications of the New Genetics ............................................................................................... 26
Being at Risk and Embodying Risk .................................................................................................. 28
Overview of the Thesis: Knowledge, Uncertainty and Potentiality ................................................ 29
  Locating the Ethnography ............................................................................................................. 38

Chapter 2. Situating the Field: Fieldwork Conditions and Relations .......................................... 40
Revelations at Home ......................................................................................................................... 41
Coming to Be: Historical Background and Present Practices of the Unit ....................................... 43
  Significant Events Fuelling the Unit’s Establishment .................................................................. 44
  The Location of the Unit ................................................................................................................ 45
  Important Predictions .................................................................................................................... 47
Funding and “Cost-Savings” of the Unit .......................................................................................... 48
Fieldwork Methods .......................................................................................................................... 50
  A Desk to Sit and Observe From ................................................................................................. 51
  Fieldwork at Home ....................................................................................................................... 54
Field Relations – Access Not Membership ..................................................................................... 56
  The Ambiguity of Rapport and Acceptance ................................................................................. 58
“Procedural Ethics” Versus “Ethics in Practice” ............................................................................ 66
  The Negotiation of Field Access ................................................................................................. 67
“Procedural Ethics” – Pre Fieldwork ............................................................................................... 68
Anticipatory Research and the Negotiation of Research ................................................................. 71
Practising Ethics in the Field ........................................................................................................... 74
  Informed Consent in Practice ....................................................................................................... 76
Conclusions ......................................................................................................................................... 80
Chapter 5. Genetic Counselling, Knowledge and Power in the Clinic .......... 154

The Practices of Genetic Counselling ................................................................. 155
A History of Genetic Counselling ................................................................. 155
Global Practices ......................................................................................... 156
Australian and Local Practices ................................................................. 157
Clinic Spaces and Clinical Emphasis ............................................................. 160
The Power of Clinic Spaces ........................................................................ 160
Genetic Counselling Clinic Appointments .................................................. 165
The Pedigree as a Tool ................................................................................ 167
Educating the Client .................................................................................... 169
Content and Extent of Information Provision During Clinic ..................... 171
Being Informed of the Decision to Test for a Genetic Mutation ................. 175
Translating the Uncertainties and Implications of the Genetic Test .......... 177
Clinician’s Discretion and Power ................................................................. 178
“Get the Doctor” .......................................................................................... 180
Clinic Appointments for “Presymps”: Almost Having the Tiger by the Tail 182
Being at risk ................................................................................................. 189
Risk, Responsibility and Surveillance ....................................................... 191
Uncertainty Surrounding the Diagnosis of Familial Cancer Risk in Clinic ... 194
Conclusions: Extents of Knowing and Not Knowing ...................................... 198

Chapter 6. Post Clinic and Beyond: Mutation Identification ....................... 202

Genetic Testing and Result Management .................................................... 202
The Provision of Genetic Test Results ....................................................... 204
Meanings and Connotations of Mutations .............................................. 207
The Potential Value of Mutations: Usefulness of Information .................. 211
Familial Cancer Mutations: Confirmation, Access and the Possibility of Prevention .................................................................................. 211
Familial Cancer Mutation: A Paradox of Future Hope and Future Concern 214
Current and Future Uncertainties ............................................................... 217
Genetic Testing for Potentiality ................................................................. 217
Genetic Testing and Genetic Relatedness ............................................... 217
The Management of Genetic Material and Knowledge .......................... 219
Conclusions: Post Result Appointment and Summary ............................. 224
Chapter 7. The Practice of Risk Notification: Considerations and Implications. 225
The Practice of Risk Notification ................................................................. 227
Risk Notification and Genetic Kinship .......................................................... 230
   Notions of Family ..................................................................................... 230
The Right for People to be Informed and for Clients to Inform ...................... 233
   Legal Considerations .............................................................................. 233
Perceived Benefits of Risk Notification .......................................................... 235
   “Non-Responders” and the Right Not to be Informed .............................. 240
The Maintenance of Genetic Information as Confidential and Private .......... 243
   Figuring it Out Anyway ......................................................................... 245
Conclusions ................................................................................................. 247

Chapter 8. Scientific Competition, Discovery and Progress ......................... 249
Competition and Excitement Around the Discovery of Mutations and Advancing Knowledge .................................................................................................................. 252
   Competition through Betting In the Unit ................................................. 253
   Competition in the Human Genome Project ............................................ 258
Excitement and Disappointment of Mutation Identification ............................ 260
   Excitement, Objectification and Discovery in Genetics ......................... 270
Objectification and Commodification of Genetic Material .............................. 272
   Genetic Material Markets .................................................................... 274
Health and Wealth ....................................................................................... 275
   A Relationship Between Science, Biomedicine and Genetic Medicine .... 275
Peer Gathering: A Conference of Competitiveness, Enthusiasm and Advancement .277
   Peer Recognition through Rewards .......................................................... 279
   The Difference between Driving a Car and an Aeroplane ...................... 279
   The Biggest Fish and the One that Got Away ....................................... 282
   The Latest, Fastest and Best Equipment ................................................. 283
   A Public Display of Clinical Practice and Medical Emphasis .................. 285
Conclusions ................................................................................................. 286

Chapter 9. Conclusion: Potentialities of Familial Cancer Risk ....................... 288
   Future Research and Thesis Contributions ............................................... 293

References ..................................................................................................... 296
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).