HOLDING YOUR BREATH: 
PREDICTIVE GENETIC TESTING 
IN YOUNG PEOPLE

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Abstract

A clash in perception is taking place. Some perceive predictive genetic testing in young people to be too potentially harmful to allow. Others perceive it to be an opportunity for benefit, even an opportunity for the prevention of harm. In this thesis I consider the issue of potential harm to mature young people who seek predictive genetic tests.

There are two parts to this thesis. In part one (chapters 1-4) I provide a background to the current debate. I describe the prohibitive stance purported within current guidelines, the arguments used to justify this stance and the opposition that has arisen in response. I discuss the psychological and social ways in which young people differ from adults, arguing that it is likely young people will react differently from adults in response to predictive genetic tests. However, I conclude that the lack of empirical evidence means we are unable to determine if these differences will confer a greater potential for harm or benefit when young people are tested. Finally, I present a discussion of two fundamental gaps in our knowledge about testing in young people: a lack of knowledge about current practice and a lack of first-hand evidence about the effects of testing. I argue that empirical research is required.

In part two of this thesis (chapters 5-7) I present the findings of my own empirical research. Firstly, I describe the findings of an international survey of clinical geneticists. Secondly, I describe the outcomes of 18 in-depth interviews performed with young people who have experienced predictive genetic testing for either Familial Adenomatous Polyposis or Huntington Disease. These young people ranged in age from 14 to 25 years.

The international survey uncovered 49 cases where predictive genetic tests had been provided to young people for non-medical reasons. When such tests are provided, the impacts are rarely followed-up as part of a formal research protocol. Clinicians’ reasons for providing and refusing tests are highly varied and are driven more by the nuances of individual cases than by any one ethical principle or set of guidelines.
When young people talk about the predictive genetic tests they have experienced, they refer to the entire experience of being at risk of a genetic condition, not simply the time after receipt of their test result. Young people speak about a far more extensive range of harms and benefits associated with the testing process than have been previously researched.

I argue that some young people growing up at risk of a genetic condition suffer several harms prior to their request for predictive genetic testing, because of their risk status. I argue that when we understand this, it becomes clear that for these mature young people who seek such testing, the provision of a test may not only serve to alleviate some of these harms, but may in fact create benefits for them, irrespective of their test result. In these cases, the provision of a predictive genetic test is appropriate, logical and ethical.
**Declaration**

This is to certify that:

(i) the thesis comprises only my original work towards the PhD except where indicated in the Preface,
(ii) due acknowledgment has been made in the text to all other material used,
(iii) the thesis is less than 100,000 words in length, exclusive of tables, maps, bibliographies and appendices.

Signed: .............................................
Preface

Some sections of this thesis have been published, or will be published shortly. Sections of Chapters One, Two, Three and Eight appear in:

- **Duncan RE.** Predictive genetic testing in young people: When is it appropriate? *J Paediatr Child Health* 2004;40,593-595.


- **Duncan RE**, Delatycki MB, Collins SJ, Boyd A, Masters CL & Savulescu J. Ethical considerations in pre-symptomatic testing for vCJD. *JME (in press).*

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For you, Mum.
# Table of contents

- **Abstract** iii
- **Declaration** v
- **Preface** vii
- **Acknowledgments** ix
- **List of tables, figures and boxes** xiii
- **Chapter 1: To test or not to test** 1
  - 1.1 Introduction 2
  - 1.2 Guidelines concerning predictive genetic testing in young people 7
  - 1.3 “Not to test”: Justifications for opposing testing in young people 12
  - 1.4 “To test”: Justifications for supporting testing in young people 29
  - 1.5 Surveys of opinion about predictive genetic testing in young people 39
  - 1.6 The essence of the debate 44
  - 1.7 Summary 46
- **Chapter 2: Empirical research** 47
  - 2.1 Introduction 48
  - 2.2 Evidence about testing young people for non-medical reasons 49
  - 2.3 Evidence about testing young people for medical reasons 51
  - 2.4 Evidence about testing adults for Huntington Disease 56
  - 2.5 Conclusion 63
- **Chapter 3: Young people** 65
  - 3.1 Introduction 66
  - 3.2 Psychological features of being young 67
  - 3.3 Social features of being young 79
  - 3.4 Predictive genetic testing in young people specifically 86
  - 3.5 Conclusion 90
- **Chapter 4: Research methodology** 93
  - 4.1 Introduction 94
  - 4.2 Fundamental gaps in our knowledge 94
  - 4.3 Two phases of empirical research 101
  - 4.4 Conclusion 122
Chapter 5: Current practice
5.1 Introduction 124
5.2 Survey respondents 125
5.3 Occurrence of testing 127
5.4 Effects of testing 137
5.5 Follow-up 141
5.6 Refusals to provide testing 142
5.7 Views on existing guidelines 147
5.8 Limitations of the survey 151
5.9 Conclusion 153

Chapter 6: Listening to young people
6.1 Introduction 158
6.2 Young people's experiences of predictive genetic testing 158
6.3 Conclusion 200

Chapter 7: Holding your breath
7.1 Introduction 202
7.2 What are we listening for? 203
7.3 Harms and benefits as described by young people 204
7.4 The risks of simplicity 221
7.5 Something more complex 222
7.6 Conclusion: Holding your breath 248

Chapter 8: Conclusions
8.1 Introduction 252
8.2 Summary 252
8.3 Key findings 254
8.4 Implications for future research 257
8.5 Implications for existing guidelines and current practice 263
8.6 Implications for the ethical debate 265

References 269

Appendix A: Interview theme list 284
Appendix B: CD Script 287
Appendix C: Letter of invitation 289
Appendix D: Plain language statement 290
Appendix E: Letter sent to parents of potential participants 296
List of tables, figures and boxes

- Chapter One
  
  Table 1.1: Arguments in current guidelines 16  
  Box 1.1: Arguments opposing testing in immature young people 22  
  Box 1.2: Arguments opposing testing in mature young people 24  
  Box 1.3: Arguments opposing testing in mature and immature young people 28  
  Box 1.4: Arguments supporting testing in immature young people 33  
  Box 1.5: Arguments supporting testing in mature young people 38

- Chapter Four
  
  Table 4.1: Codes used to analyse qualitative aspects of the survey 112  
  Figure 4.1: Three target populations for interviews with young people 114

- Chapter Five
  
  Table 5.1: Respondents’ job title 126  
  Table 5.2: Respondents’ country of employment 127  
  Table 5.3: Cases in which tests were performed on immature young people 130  
  Table 5.4: Clinicians’ reasons for performing tests on immature young people 132  
  Table 5.5: Cases in which tests were provided to mature young people 134  
  Table 5.6: Clinicians’ reasons for providing tests to mature young people 137  
  Table 5.7: Harmful and beneficial outcomes of testing in young people 141  
  Table 5.8: Clinicians’ reasons for refusing to provide tests to young people 146  
  Table 5.9: Clinicians’ views of current guidelines 147  
  Table 5.10: Clinicians’ justifications for their views of current guidelines 151

- Chapter Seven
  
  Table 7.1: Harms associated with testing in young people 210  
  Table 7.2: Benefits associated with testing in young people 218
CHAPTER ONE: TO TEST OR NOT TO TEST
1.1 Introduction

“I was sitting in a car with my cousin ... he said to me do you know that your dad has this disease and ... you might get it too... I didn’t know anything about it, I hadn’t been told by my dad or mum ...I really didn’t know what was going on... in retrospect looking back my dad did show signs of it but I just thought he was drunk”

Zach, 26 yrs

“I sort of knew, because ... grandma had it and she was always sort of jittery and sort of weird and stuff... mum sort of, she went a bit troppo, like, she was saying she was going to commit suicide and all this sort of stuff...[she] used to psych out a bit, and then she, after a while, she used to get like movements and stuff”

Troy, 26 yrs

“My father, you know, he doesn’t believe in counselling and all of that, he’s a bit sort of old fashioned... he said oh your mother’s got Huntington’s Disease ... you know, she’s very sick, she’s going to die in 10 to 15 years time and ah there’s a 50-50 chance that you’ll get it too”

Oliver, 24 yrs

Genetic knowledge has now reached a level of precision at which we are able to predict elements of an individual’s medical future. Genetic predictions, or predictive genetic tests, can be made from the day a person is conceived and in the complete absence of symptoms. For individuals who live in families with a history of a genetic condition this translates into the real possibility of finding out, with frightening accuracy, whether they too will go on to develop the condition they are at risk of manifesting. The three individuals I quote above all chose to undergo such testing. Most choose not to.¹
Although theoretically predictive genetic tests are possible from the time of conception, professionals disagree about the age at which such tests should be performed. Adults (those who have reached the age of majority) are able to undergo predictive genetic testing if they wish to do so. Young people (under the age of majority) do not share such ease of access. Why don’t they? Should they? How young is too young for a predictive genetic test?

In this first chapter, I provide a background to current debates concerning predictive genetic testing in young people. I am concerned here with clarifying who is arguing about testing in young people and what they are arguing about. Beginning with a summary of existing guidelines, I describe the kinds of arguments used to oppose testing in young people. I then turn my attention to the range of objections emerging in response to these guidelines. That is, the arguments used to support the testing of young people. Finally, I articulate the core issues in the current debate and specify one particular aspect of this core that I focus my thesis upon. The aspect I focus upon is the issue of potential harm to young people.

In order to perform the literature review that forms the basis of my discussion in this first chapter, I utilised a three-tiered approach. Firstly, I searched the electronic resource ‘Medline’ for published guidelines concerning predictive genetic testing in young people. I also searched Human Genetics Societies web-sites in order to access guidelines that had not been published in peer-reviewed journals. I then searched the reference lists of these guidelines. Secondly, I performed a detailed search using Medline again, to access general literature on the topic. The MeSH headings of “Genetic Screening” “Child”, “Adolescence” and “Genetic Counseling” were too broad to elicit the papers I was interested in. Therefore the MeSH heading “Huntington Disease” was used, in addition to the following key-words and phrases: (1) Predictive Genetic Testing, (2) Child, (3) Adolescent/Adolescence, (4) Minors, 1 and 2, 1 and 3, 1 and 4, 5 or 6 or 7. All searches were limited to English language articles. Finally, I searched the reference lists of these articles.
1.1.1 Initial clarifications

Some initial clarification is necessary in order to elucidate the meaning of specific terms I use throughout this thesis. I also use this space to briefly describe the two genetic conditions I refer to most frequently throughout this thesis: Huntington Disease (HD) and Familial Adenomatous Polyposis (FAP).

1.1.1.1 Gene-positive and gene-negative test results

I use the term ‘gene-positive’ to refer to the presence of a genetic mutation associated with a condition. Individuals receiving a gene-positive test result have inherited the genetic mutation that runs in their family and therefore have an increased risk of developing the condition later in life. Accordingly, I use the term ‘gene-negative’ to refer to the absence of a genetic mutation associated with a condition. Individuals receiving a gene-negative test result have not inherited the genetic mutation that runs in their family and therefore are at a decreased risk of developing the condition later in life.

1.1.1.2 Medical reasons and non-medical reasons for tests

Throughout this thesis I use the terms ‘medical benefit’ and ‘non-medical benefit’ (or ‘medical reasons’ and ‘non-medical reasons’) to distinguish between different motivations for predictive genetic testing. When using the term ‘medical benefit’ I refer to clinical benefits only. That is, a predictive genetic test creates medical benefit when knowledge of genetic status is able to assist in either preventing or treating the condition, altering clinical outcome. For example, with Familial Adenomatous Polyposis, if a child is gene-positive for a familial mutation, appropriate surveillance and treatment can be instituted to reduce the risk of disease morbidity. When using the term ‘non-medical benefit’ I refer to the range of psychosocial benefits that predictive genetic testing may create, such as decreased uncertainty or a greater ability to plan for the future. That is, benefits of testing that do not alter disease progression or outcome, but have the potential to impact positively on an individual’s life nonetheless.

1.1.1.3 Huntington Disease

Huntington Disease (HD) is a neurodegenerative condition characterised by involuntary movements, cognitive impairment and changes in behaviour and
personality. The movements associated with HD may become highly disabling.\(^2\) On average, HD results in death 15 years after symptom onset.\(^3\) The mean age of onset for HD is 37 years.\(^2\) There is currently no effective treatment or prophylaxis available.

Individuals with a parent who has the mutation associated with HD have a 50% chance of inheriting the condition themselves. A gene-positive result generally provides 100% certainty that HD will manifest later in life and a gene-negative result provides 100% certainty that HD will not manifest later in life. Predictive genetic testing for HD is generally undertaken within a formalised protocol where individuals receive counselling and have the opportunity to explore what a gene-positive and gene-negative result may mean for them.\(^4\)

As there is no effective treatment or prophylaxis available for HD, all predictive genetic tests performed to gain knowledge of HD genetic status are performed for non-medical reasons. In other words, learning of one’s HD gene status does not, at this time, create any opportunity for preventing or treating the condition. Such knowledge is sought for alternative reasons, which I discuss in detail later.

1.1.1.4 Familial Adenomatous Polyposis (FAP)

Individuals with Familial Adenomatous Polyposis (FAP) develop hundreds of small polyps in their colon and occasionally their small intestine during their first 20 years of life.\(^5\) The polyps themselves are usually asymptomatic, but eventually progress to colon cancer. Clinical management for FAP involves annual endoscopic screening, starting at approximately the age of 10 years. It is also recommended that young people at risk of FAP receive genetic counselling and consider predictive genetic testing if the familial mutation is known.\(^6\) Complete removal of the colon, when polyps appear, gives gene-positive individuals the chance of a normal life expectancy.\(^6,7\)

Individuals with a parent who has the mutation associated with FAP have a 50% chance of inheriting the condition themselves. Young people who are gene-positive for FAP are certain to develop cancer later in life if they remain untreated.\(^5,8,9\) Conversely, young people who are gene-negative for FAP have no greater chance of developing colon cancer than any other individual in the general population and thus have no need for regular screening or for surgery.
As individuals who are gene-positive for FAP are able to undergo screening and surgery in order to prevent the development of colon cancer, such testing is offered for medical reasons. In other words, predictive genetic testing in young people for FAP brings about a direct medical benefit.

### 1.1.1.5 Mature and immature young people

When talking about young people, it is necessary to make a distinction between immature and mature individuals. When referring to immature young people I refer to young people who do not possess cognitive capacities that allow them to appreciate the implications of predictive genetic testing. When referring to mature young people, I refer to young people who are able to engage in discussions about genetics and predictive genetic tests. Of particular importance here is the ability to perceive long-term implications of these tests. Because both cognitive and psychological capacities vary between individuals, I refrain here from providing specific age ranges to associate with the terms ‘immature’ and ‘mature’.

### 1.1.1.6 Interview codes

Throughout this thesis I quote from interviews with young people who have undergone predictive genetic tests. As I describe in detail later, I interviewed these young people as part of my empirical research concerning the impact of predictive genetic testing in young people. At the end of each quote I use a code to identify the young person who provided the quote and some basic information about him or her. The codes refer to the following pieces of information:

- **Pseudonym : Gender : Age at interview : Condition tested for : Age when tested : Result**

For example, Belinda is a young woman who was tested for the HD gene mutation at the age of 21 years. She received a gene-positive test result and when I interviewed her she was 25 years of age. Therefore her code reads:

Alternatively, Doug is a young man who was tested for the FAP gene mutation at the age of 14 years. He received a gene-negative test result and when I interviewed him he was 18 years of age. Therefore his code reads:

- Doug:M:18:FAP:14:-ve

Following these initial clarifications, I now return to the topic of predictive genetic testing in young people and begin my review of current guidelines.

1.2 Guidelines concerning predictive genetic testing in young people

In 1990 the International Huntington Association and the World Federation of Neurology published a policy statement concerning the provision of predictive genetic testing for HD. Although not solely concerned with the topic of testing young people, this statement was the first to outline specific policy concerning young people and predictive genetic tests. The statement recommended that the HD predictive test only be available to “those having reached the age of majority” (p 34).10 This statement was then revised in 1994 but the recommendation remained unchanged.11 A working party of the Clinical Genetics Society (CGS) (a subcommittee of the British Society of Human Genetics or BSHG) was then commissioned to write a report specifically on genetic testing in children.

The research conducted by the working party culminated in the publication of a 13-page report.12 This report was the first of its kind. It was solely concerned with the topic of predictive genetic testing in young people, as opposed to adults, and it widened the focus from HD to a range of additional conditions that could be tested for genetically. The report outlined definitive recommendations regarding predictive genetic testing in young people and clinical practice. The first two conclusions of the report were:
1 “The predictive genetic testing of children is clearly appropriate where onset of
the condition regularly occurs in childhood or if there are useful medical
interventions that can be offered (for example, diet, medication, surveillance
for complications)” (p 785).

2 “In contrast, the working party believes that predictive testing for an adult
onset disorder should generally not be undertaken if the child is healthy and
there are no medical interventions established as useful that can be offered in
the event of a positive test result. We would generally advise against such
testing, unless there are clear cut and unusual arguments in favour. This does
not entail our recommending that families should avoid discussing the issues
with younger children, but rather that formal genetic testing should generally
wait until the “children” request such tests for themselves, as autonomous
adults. This respect for autonomy and confidentiality would entail the deferral
of testing until the person is either adult, or is able to appreciate not only the
genetic facts of the matter but also the emotional and social consequences of
the various possible test results. In circumstances when this type of testing is
being contemplated, there should be full discussions both within the family and
between parents and genetic health professionals; the more serious the disorder,
the stronger the arguments in favour of testing would need to be” (p 785).

The working party thus set up a clear division between predictive genetic tests that are
provided for medical reasons and those that are provided for non-medical reasons.
They acknowledged their support for tests that are capable of providing young people
with medical benefit, or tests that are provided for conditions that often manifest in
childhood. However, in cases where testing does not provide medical benefits, they
recommended caution. In these cases, they suggested that testing only be performed at
the request of the person at risk, once he or she had grown into an autonomous adult.

In 1995, following the BSHG report, the American Society of Human Genetics
(ASHG) Board of Directors in combination with the American College of Medical
Genetics (ACMG) Board of Directors published a similar report.13 This report was
drafted by a subcommittee of the ASHG’s Social Issues Committees. Their report took
a less directive approach than the BSHG report. Rather than stating ultimate
conclusions, the ASHG report simply outlined “points to consider”, with one general
conclusion at the end of the report:
“Providers who receive requests for genetic testing in children must weigh the interests of children and those of their parents and families. The provider and the family both should consider the medical, psychosocial, and reproductive issues that bear on providing the best care for children. This will require the provider to engage individual families in comprehensive discussions of these issues and to provide them with specific information and recommendations about genetic testing. Because such testing has potential for both great benefit and great harm, and because the availability of tests continues to expand, providers of genetic services will play increasingly important roles in counseling families about the suitability of genetic testing for their children” (p 1239).

At first glance, the ASHG therefore appeared to refrain from drawing conclusions as definitive as those found in the BSHG report. However, closer inspection reveals that several of the “points to consider” found in the ASHG report are in fact very similar to the “conclusions and recommendations” to be found in the BSHG report. Consider the following points:

1. “Timely medical benefit to the child should be the primary justification for genetic testing in children and adolescents. Under this condition, genetic testing is similar to other medical diagnostic evaluations. Medical benefits include preventive measures and therapies, as well as diagnostic information about symptomatic children. If the medical benefits are uncertain or will be deferred to a later time, this justification for testing is less compelling” (p 1233).

2. “Substantial psychosocial benefits to the competent adolescent also may be a justification for genetic testing. The benefits and harms of many genetic tests are psychosocial rather than physical. Relevant issues include anxiety, self-image, uncertainty, and the impact on decisions relating to reproduction, education, career, insurance, and lifestyle” (p 1233).

3. “If the medical or psychosocial benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult-onset diseases, genetic testing generally should be deferred. Exceptions to this principle might occur when the adolescent meets conditions of competence, voluntariness, and adequate understanding of information. Further consultation with other genetic service providers, pediatricians, psychologists, and ethics committees may be appropriate to evaluate these conditions” (p 1233).
The ASHG therefore mirrored the recommendations made by the BSHG, that medical benefit should form the primary justification for predictive genetic testing in young people. The ASHG also expanded these recommendations to incorporate the possibility of psychosocial benefits of testing, as well as the possibility of competent adolescents. However, the psychosocial benefits referred to by the ASHG remained narrow and an explicit, unsubstantiated assumption was made. The ASHG made an assumption that the psychosocial benefits related to adult-onset conditions would not occur until adulthood (see point 3). Taken alongside the recommendations made by the BSHG, a theme therefore began to emerge. Predictive genetic tests that do not provide medical benefit to young people should generally be deferred.

The European Society of Human Genetics (ESHG) did not produce a policy specifically concerning genetic testing in young people, but already had a policy on the “Provision of Genetic Services in Europe”. This policy focused on “current practices and issues”. The fifteenth recommendation is related to genetic testing in children and adolescents:

“Genetic diagnosis in children and adolescents requires careful consideration of what is in their best interest. It is indicated if it is necessary for the differential diagnosis of manifest symptoms or for establishing the case of a disease. A predictive genetic test is indicated during childhood if the onset of a disorder can be expected at this age and if medical measures can be taken to prevent the disease or its complications or to treat the disease. Other predictive tests and tests for carrier status should be delayed until the person is old enough to make an informed decision.”

The theme therefore continued, with predictive genetic tests being recommended for young people if the test is able to provide a medical benefit. In cases where medical benefit is not an outcome, testing should be deferred until the young person is of an age where he or she can make a competent decision about the test.

The Human Genetics Society of Australasia (HGSA) also made public a policy regarding the genetic testing of children and adolescents, titled “Predictive Testing in Children and Adolescents”. This policy existed in addition to the HGSA policy titled
“DNA Presymptomatic and Predictive Testing for Genetic Disorders”. This more general policy stated:

“Children should, in general, only have presymptomatic/predictive testing when the resulting information will be used to help with their health management in the immediate future, and not simply because parents wish to know. However, the age at which presymptomatic/predictive testing can be offered to a child should be given flexible consideration by the testing team (see HGSA policy on testing of children and adolescents).”

The HGSA policy specifically about children and adolescents adds to this:

“Genetic health professionals within Australia and overseas have followed international guidelines recommending that testing of children under 18 years of age be considered only when the result is likely to be of direct benefit to the child through medical surveillance, use of prevention strategies, or other medical interventions. However, some parents and writers have challenged the view that testing under the age of 18 years should be restricted, suggesting that there may be other benefits for the individual and the family that have not previously been considered… If a child under the age of 18 years requests predictive genetic testing for an adult-onset disorder, the health professional must seek to establish the competence of the child to make an appropriate decision regarding the test.”

The HGSA policy then incorporated a discussion of relevant concerns to assist in assessing the competence of an individual, following this with a discussion of relevant ethical principles.

The HGSA thus again confirmed the emergent theme. Human genetics societies around the world recommend against predictive genetic testing in young people unless the test is capable of creating a medical benefit. When this is not the case, it is recommended that testing be deferred until the young person is autonomous and able to make a competent decision about whether to undergo such testing. Some guidelines purport that this does not occur until the age of 18 years, while others are less specific.

The guidelines I have summarised in this section remain the most detailed and most influential recommendations to date. I turn now from what current guidelines recommend to why these recommendations are made. That is, in the next section I
describe the kinds of arguments used to oppose predictive genetic testing in young people.

1.3 “Not to test”: Justifications for opposing testing in young people

In this section I set out the reasons for current resistance towards the provision of predictive genetic tests to young people. I begin by clarifying the arguments provided in existing guidelines and then describe the arguments articulated in the wider literature.

1.3.1 Justifications in current guidelines for opposing testing in young people

Guidelines published by the BSHG, ASHG, ESHG and HGSA all provide some (if minimal) justification for the conclusions reached within them. The BSHG report includes the following statement:

“Given the dearth of evidence, it is the ethical consequences (loss of adult autonomy and confidentiality, and the possibility of causing harm to the developing child), together with the limited empirical evidence that is available, that has led us to advocate this cautious policy, erring towards a presumption of non-maleficence (“primum non nocere – the first goal is to cause no harm”)” (p 791).

And then follows this with:

“In our judgement, the possible emotional harm to the child, abrogation of their autonomy and the breaching of their confidentiality, will generally outweigh the possible benefits that we can see” (p 791).

The BSHG report then contains a detailed discussion to justify the conclusions outlined within it. The authors explain how “knowledge that a child will develop such a disorder may, at least in some family contexts, cause worse problems than continued uncertainty” (p 791). They also note that “there are now good legal reasons for professionals to make decisions in this area primarily on the basis of the long term best interests of the child; failure to do so could lead to professionals later being sued” (p
The authors highlight research concerning adults who undergo predictive genetic testing for HD, explaining that “only a minority of at risk adults in fact choose to undergo such testing (about 10-15%)” (p 791). They also refer to the “precedent of the decision made by those performing predictive genetic testing for Huntington Disease” where “a consensus was reached by those involved in the predictive testing of those at risk of Huntington Disease, who decided that children should not be tested” (p 791). However, the authors also acknowledge that this decision was arrived at on the basis of the possible effects of such testing and not from the experience of harmful effects.

The ASHG report contains a slightly more elusive justification for the conclusions outlined within it:

“This report is grounded in several social concepts: first, the primary goal of genetic testing should be to promote the well-being of the child. Second, the recognition that children are part of a network of family relationships supports an approach to potential conflicts that is not adversarial but, rather, emphasizes a deliberative process that seeks to promote the child’s well-being within this context. Third, as children grow through successive stages of cognitive and moral development, parents and professionals should be attentive to the child’s increasing interest and ability to participate in decisions about his or her own welfare” (p 1233).

Rather than entering a detailed discussion of justifications for their stance, the authors of the ASHG report provide direction relating to how a decision about testing in children can best be made. The authors state that “decisions about genetic testing in children should be based on an assessment of the possible benefits and harms that may be associated with the tests” (p 1234). They go on to describe that “the putative benefits and harms include medical, psychosocial, and reproductive issues that have implications for the child, the immediate family, and more distant relatives” (p 1234), then describing these in detail. One could perhaps therefore infer that the authors of the ASHG report have used similar methods of deliberation in order to reach the conclusions they describe, although such methodology is not explicitly stated.

The ESHG recommendations do not include an obvious justification for the conclusions that are outlined. The policy does state that:
“Genetic diagnosis in children and adolescents requires careful consideration of what is in their best interest”

The HGSA policy does not contain an explicit justification for the conclusions outlined within it either. However, it does contain a description of three “underlying ethical principles”. Firstly, that of autonomy:

“The principle of autonomy relates to a person’s ability to make or exercise a self-determining choice. Predictive testing in childhood removes the possibility of the child making an autonomous decision as an adult. It is for this reason that predictive testing programs usually limit testing to individuals over the age of 18 years unless there is a clear and immediate benefit to the child. Some adolescents may have sufficient maturity to make their own decision about predictive testing. There is a need for flexible consideration of the age at which children and adolescents will be ready for information which will help them in decision-making about their own predictive testing.”

Secondly, that of non-maleficence:

“Predictive testing of children may result in harm for the child. Potential harms that can arise from predictive testing include: disclosure of the test result to others resulting in loss of privacy, discrimination and loss of options directly arising from predictive testing results, stigmatisation in the family and community with reduced opportunities for education, marriage and reproduction, and alteration of parenting as the result of knowing the child’s genetic status.”

And, thirdly, that of beneficence:

“The interests of the child, the parents and the family need to be considered when a request is made for predictive testing. The benefits and harms should be categorised into medical, psychological and reproductive issues. Ultimately the best interest of the child must prevail and it is important that the health professional be an advocate for the child during pre-test counselling of the family and child.”

The HGSA acknowledges that there is a lack of evidence-based research concerning this issue. It recommends that “there be further studies to document the psychological and social effects of predictive genetic testing of children and the consequences of not providing such testing.” Much like the ASHG report, one must infer that the authors of
the HGSA policy statement have reached their conclusions following an examination that incorporates the ethical principles they describe, although once again such methodology is not explicitly stated.

Viewed as a whole, existing guidelines concerning predictive genetic testing in young people rely heavily on the ethical principles of autonomy, beneficence and non-maleficence to justify the conclusions outlined within them. The only justification that is used in all sets of guidelines summarised above is the concept of young people’s “best interests” or “well-being”. Critically absent from current guidelines is empirical evidence specifically about the impacts of predictive genetic testing in young people. Thus, in order to justify the conclusions reached, existing guidelines rely on a desire to maximise young people’s best interests, combined with a view that withholding predictive genetic tests for non-medical reasons from them is the best way to do this. This is a cautionary stance.

Table 1.1 summarises the arguments found in current guidelines that are used to oppose predictive genetic testing in young people.
Table 1.1 Arguments in current guidelines

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<th>Arguments based on the ethical principle of autonomy</th>
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<th>Arguments based on maximising best interests of the young person</th>
<th>BSHG Guidelines</th>
<th>ASHG Guidelines</th>
<th>HGSA Guidelines</th>
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As guidelines were published internationally, an extensive body of literature relating to predictive genetic testing in young people also emerged. To date, the vast majority of this literature purports a conservative stance; arguing against such testing and thus lending support for the guidelines. I now summarise the arguments found in this body of literature.

1.3.2 Justifications in the wider literature for opposing testing in young people

The same year that definitive policy about predictive genetic testing in young people was published for the first time, a seminal paper was published by Bloch and Hayden. At the time, Bloch and Hayden were involved in a Canadian national program of predictive testing for HD. A founding principle in the establishment of this program was the decision “to offer testing only to persons above the age of 18 years” (p 1). Bloch and Hayden therefore wrote their paper in an attempt to detail a justification for this position. They also noted that at the time of writing a number of parents had already requested predictive testing for their minor children. This paper was thus also the first to provide an indication of current practice.

Bloch and Hayden stated that “testing for minors should not be offered at the request of a third party, and prenatal testing which would result in the birth of a child at increased risk for HD should, where possible, be avoided” (p 4). They cited the ethical principle of autonomy as a major influence in the development of their program, arguing that a person has a right to decide what is best for him or herself. This paper, together with the guidelines that had been published that same year by the International Huntington Association and the World Federation of Neurology, set a definitive line. The line they set was unequivocally at 18 years. Testing before this age, at least for HD, was unacceptable.

Such a precise line proved to be rare in the literature that followed. The subsequent literature, although also opposing predictive genetic testing in young people, was far less specific about the age at which testing may be considered. Difficulty arose in distinguishing between mature young people who may be requesting testing
themselves and immature young people who would remain ignorant of the testing process entirely.

Harper and Clarke were the first to highlight a distinction between mature and immature young people. They stated that “to test children at an age when they are not considering or are not able to consider their own future reproductive plans would seem unnecessary and unethical.” (p 1206). Thus a vital shift occurred conceptually. The argument evolved from a categorical position that testing of any minor was wrong, to the assertion that it was wrong because the young person was not involved in the decision to be tested. The literature opposing predictive genetic testing in young people that followed this paper by Harper and Clarke often echoed such a conception. That is, the arguments often focused on the inappropriateness of testing immature young people. I now summarise these papers that argue against the testing of immature young people specifically.

1.3.2.1 The inappropriateness of testing immature young people

The arguments used to oppose predictive genetic testing in immature young people can be divided into two categories. Firstly, those that are based on the consequences of parents knowing their children’s genetic status and, secondly, those that are based on the consequences of immature children learning of their own genetic status.

Many arguments opposing testing in immature young people focus on the ethical principle of autonomy. Arguments based in theories of autonomy hold that it is unethical for young people to undergo testing in ignorance as they lose the opportunity to decide if they want to have testing themselves. Thus testing of young people, it is argued, is only acceptable when the young person is directly involved in the decision. These arguments fall into the former category outlined above, that is, arguments based on the consequences of parents knowing their children’s genetic status.

Hoffman and Wulfsberg argue against the testing of very young children. They advise delaying the test until the child is of an age where the implications of the test can be properly understood. Clarke and Flinter make a similar argument, using statistics on adults’ uptake of predictive testing as substantiation. They argue that “testing in childhood removes the individual’s future right to make their own decision
about testing as an autonomous adult. The majority of eligible adults have decided not to take such tests” (p 166). Davis writes of a child’s so-called “right to an open future” (p 554), stating that “a better account … protects the child’s right to an open future by preserving into adulthood his own choice to decide whether his life is better lived with that knowledge or without” (p 589). Holland states that “not everyone wants to know their genetic makeup” (p 337). Bloch and Hayden state that “predictive testing for Huntington Disease strongly highlights the need for autonomy and the need for each individual to decide about his or her willingness – or unwillingness – to obtain genetic information predictive of the future outcome” (p 4). Other authors have articulated similar arguments against testing in young people, justified through theories of autonomy.

Confidentiality has also been used as a justification for opposing the testing of immature young people. This also falls into the former category of arguments, those that relate to parents learning of their children’s genetic status. Fryer notes that when a young person undergoes predictive genetic testing, he or she is not granted the same confidentiality that an adult would receive due to the disclosure of results to the child’s parents. Clarke and Flinter agree, stating that “testing in childhood removes the confidentiality that would be expected and provided for any adult undergoing the same test, both for the fact of having the test performed and for the test result” (p 166).

Additional arguments relating to parental knowledge of their children’s genetic status concern the ways in which parents may alter their beliefs. Wertz and colleagues write of the altered expectations parents may have of their child following predictive genetic testing:

“Parents … may not expect a child who tests positive for a genetic disorder with shortened life expectancy to train for a profession. Such children grow up in a world of limited horizons and may be psychologically harmed even if treatment is subsequently found for the disorder. In families with a chronically ill child, there is less socialisation to future roles for all the children, including those who are healthy. Parents are less likely to say “When you grow up…” or “When you have children of your own…” to any of the children, because they cannot say these words to the ill child” (p 881).
Lessick and Faux write of the “vulnerable child” syndrome, arguing that this is a possible outcome for young people who find they are predisposed to a condition. Lessick and Faux state that “the identification of a genetic diagnosis or predisposition may create a “vulnerable” child who is over-protected or unnecessarily restricted in childhood activities by parents” (p 40). Wertz and colleagues also note that such over-protectiveness can begin before a definitive diagnosis, and can last long after the risk has passed. Ross and Moon raise the possibility of “parents modifying their expectations, even subconsciously” (p 877). Fanos writes of the vulnerable child syndrome, stating that parents can react to “the threatened loss of a child by establishing emotional distance from the child” (p 24). Holland notes that “parents may spend less resources on the afflicted child because they may not want to “waste” time, money, counseling, love etc., on a child who will not be able to utilize the resources” (p 341).

Additionally, several of the arguments used to oppose predictive genetic testing in immature young people relate to consequences of young people learning of their own genetic status.

Meiser suggests that young people may blame themselves for a bad test result. She states that “testing may also result in feelings of guilt in children, who often blame themselves for illness, especially at younger ages” (p 128). This is also suggested by Fanos, who writes that “children often perceive their illness as punishment for misbehavior or for harbouring bad thoughts” (p 24).

The possibility that immature young people will misunderstand the genetic information that is imparted to them constitutes an additional justification for opposing such testing. Fanos asks whether parents will “be able to understand and transmit accurate information to their offspring concerning their genetic risks” (p 25). Wertz and colleagues similarly note that “sometimes misunderstandings by parents or child lead to serious misconceptions about the future” (p 876). They also suggest that “in families where one sibling tests negative for a genetic disorder, this may result in perceptions of increased risk in other siblings who are not tested” (p 876).
There is also legal support for the arguments that oppose predictive genetic testing in immature young people. Clayton writes specifically on the legal aspects of predictive genetic testing in young people.\textsuperscript{30} She acknowledges that “the law has little to say about how physicians resolve these dilemmas” but that “parents do not in fact have a constitutional right to demand that physicians perform genetic tests for adult-onset diseases … on their children” (p 633). Clayton writes specifically about the case of testing immature young people, as requested by their parents, and states that “physicians need not fear liability for refusing to perform genetic tests, unless results would significantly alter medical management in a way that averts physical harm to the child” (p 633). Clayton thus provides legal support for arguments opposing the testing of immature young people.

Box 1.1 summarises the arguments used to oppose predictive genetic testing in immature young people.
Box 1.1 Arguments opposing testing in immature young people

- Testing immature young people fails to respect their future autonomy as it removes their right to make a decision about testing as an autonomous adult.
- Many adults choose not to undergo testing and the decision should therefore be left up to the individual, not their parents.
- When a young person is tested, confidentiality is breached because parents are informed of the test result.
- Parents may alter their expectations of the child who receives a gene-positive test result.
- A child receiving a gene-positive test result may be raised in a world of limited options.
- Testing may create a ‘vulnerable child syndrome’, where the child is over-protected.
- Less resources may be spent on a child who receives a gene-positive test result.
- Parents do not have the right to gain knowledge of their children’s gene status.
- Children may blame themselves for a gene-positive test result, viewing it as punishment.
- Children may feel guilty about a gene-positive test result.
- Children and parents may misunderstand the genetic information that is conveyed to them.
- If one child receives a gene-positive test result, siblings may believe they are at decreased risk.
- If one child receives a gene-negative test result, siblings may believe they are at increased risk.
Next, in my summary of justifications for opposing predictive genetic testing in young people, I outline the objections that apply to testing in mature young people.

1.3.2.2 The inappropriateness of testing mature young people

Some of the opposition to predictive genetic testing in young people focuses specifically on the developmental processes required of mature young people and the ways in which these may be affected by such testing. Fanos writes in detail about the process of separation that needs to occur between parents and young people and the ways that this may be affected by testing:

“In adolescence the loosening of emotional dependencies upon the family must be achieved…. The realisation that there is a bond with the parent because of a serious disorder may delay this separation process, either on the part of the adolescent or on the part of the parent. Similarly, guilt over not being at risk when a parent is a carrier or affected may interfere with the normal task of separation” (p 26).

She also writes of the process that adolescents must go through of realising that their parents are not all-powerful, and how this may be affected by testing:

“The perception of parents as alternately powerful or weak is a normal adolescent process. The adolescent needs to be exposed … to a relinquishment of idealisations. The presence of carrier state or illness in a parent, or the realisation that a parent could not save the affected child may affect this process. This sudden and dramatic loss of perceived parental power destroys the normal psychological transition” (p 27).

Fanos writes of the ways in which establishing a personal identity may be made difficult by testing. She states that “adolescence is a time of establishing one’s personal identity” and that “self-stigmatisation by learning that one is a carrier is a danger, as is stigmatisation by peers” (p 26).

Box 1.2 summarises the arguments used to oppose predictive genetic testing in mature young people.
Box 1.2 Arguments opposing testing in mature young people

- Testing may interfere with the natural separation process that needs to occur between parents and their children.
- A gene-positive child may be forced into the realisation that their parent is not ‘all-powerful’ and couldn’t save them from the genetic condition. This could be psychologically damaging if it occurs too early in the young person’s life.
- The process that occurs during adolescence of establishing a personal identity may be made particularly difficult by a gene-positive test result.

Finally, in this section on arguments opposing predictive genetic testing in young people, I outline the justifications used to oppose testing that apply to both immature and mature young people.

1.3.2.3 The inappropriateness of testing both immature and mature young people

Several of the arguments used to oppose predictive genetic testing in young people are not specific to either immature or mature young people. These types of arguments may be used as a justification for opposing testing in either of these groups.

Wertz and colleagues suggest that “a child or adolescent who tests positive may become a scapegoat who is abused because he or she reminds a parent of the parent’s own unacceptable traits” (p 876).\(^{25}\) Meiser similarly notes that “harm to the parent-child relationship may result from parents becoming overprotective or, alternatively, discriminating against or scape-goating a child with a positive result, or experiencing continued anxiety over a child despite a negative result” (p 128).\(^{29}\)

Fryer writes of the more general family dynamic of treating a child as a victim. He believes testing may in fact strengthen such schemas:
Persons are sometimes singled out for no rational reason as being destined to develop the disorder. Such family ‘preselection’ has been recognised as having potentially adverse effects on the emotional development of the unfortunately labelled child. One could envisage this being an even greater problem if a test confirmed the family dynamic that had selected one child as the ‘victim’” (p 98).22

Fanos discusses possible changes in self-concept that could occur, stating that “being identified as a carrier of a gene mutation may reinforce latent feelings of unworthiness…. these feelings may interfere with the normal identification process with peers and disturb the child’s self concept” (p 24).28 Holland similarly notes the feelings of inadequacy that may result. She suggests that one may be “afraid of relationships … believing that one is “unmarriageable” because of ones genetic disorder” and notes that these are “barriers to a full and happy life” (p 339).21 Wertz and colleagues talk of the identification process, suggesting that testing may interfere with the constant drive children and adolescents have to integrate with their peers.25 Meiser notes that if the identification process is disrupted, it may lead to “loss of self esteem and harm to the child’s self-concept” (p 876).20 Bloch and Hayden and Lessick and Faux also refer to the potential negative effects to self-esteem that testing may have.26 16 Clarke and Flinter write of losses to self-esteem caused by altered parental expectations. They note that “altered expectations of the child’s intellectual abilities, future health and future relationships could affect the deepest levels of self-esteem and have devastating social, emotional, psychological and educational consequences” (p 166).19

Increased anxiety is yet another possible effect of testing in young people.26 28 Holland describes in detail the scenario of the ‘unpatient’:

“A possible repercussion of finding out that one has tested positive for a genetic disorder has been labelled … as the “unpatient syndrome”. Unpatients appear mentally and physically well on the outside; however, they know that they are at an increased risk of suffering from serious disorders. Unpatients may develop severe anxiety and believe that they are under a death sentence. They may visit doctors frequently, seeking monitoring and reassurance. In extreme cases, they may contemplate suicide. They may be ostracized by friends and family and be discriminated against in insurance and employment” (p 338).21
Other concerns about psychosocial harms of testing relate to feelings of guilt. Lessick and Faux refer to the concept of survivor guilt in the young person who is tested. They state that “children or adolescents whose test results exclude them from risk for a condition may experience “survivor guilt” based on knowledge that one or more of their siblings will develop a serious genetic condition” (p 40). Meiser also refers to this possibility, suggesting that a gene-negative test result “may separate siblings who were formerly united by a bond of risk” (p 128). Holland suggests that parents may also suffer from guilt, which could indirectly affect their child. She states that “genetic disorders can make parents feel guilty, which can lead to “overindulgence and protectiveness” (p 340). Ross and Moon also note that “disruption of normal family relationships because of guilt on the part of parents or on behalf of the unaffected siblings” may occur (p 877).

Yet another concern about psychosocial harms of testing is the stigmatisation and discrimination that may occur as a result of testing. Holland states that “testing classifies individuals according to their genetic make-ups” (p 344) and that classifications can lead to discrimination… including insurance discrimination and job discrimination. Hanson and Thomson realise that this may affect more than just the individual who is tested. They state “the individuals being tested, their families, and even the wider communities to which they belong may feel stigmatized by the results” (p 287). It has been noted that insurance companies generally refuse to issue life insurance to adults who have received a gene-positive test result but do offer insurance, albeit at increased rates, to adults who are at risk but have not yet been tested. Thus, if young people are tested under the age of 18 years and receive a gene-positive test result, there is a possibility that they will not be able to obtain life insurance once they are adults.

There are also other, more indirect outcomes of testing that may have negative psychosocial effects. Hanson and Thomson state that “among the adverse and unintended consequences of genetic testing in children is an increase in financial burden” (p 288). They note that “those families whose children are screened may incur additional expense or anxiety by virtue of a false-positive or an indeterminate test result” (p 288). Ross and Moon similarly note the possibility of incorrect results. They state “genetic testing … is accompanied by … medical risks created by false-
negative and false-positive results” (p 876). Finding cases of mistaken paternity is yet another possible negative outcome. Hanson and Thomson note that “testing of one or a few family members may reveal highly sensitive information about other family members (eg. misidentified paternity within a family)” (p 287).

Box 1.3 summarises the arguments used to oppose testing in both immature and mature young people.
Box 1.3 Arguments opposing testing in immature and mature young people

- A child who receives a gene-positive test result may become a scapegoat who is abused because he or she reminds the parent of the parent’s own unacceptable traits
- Harm to the parent-child relationship may occur, especially if parents become over-protective or experience anxiety
- Young people may feel guilty if they receive a gene-negative result, and their parent or other siblings are gene-positive
- The child may be treated as a victim in the family
- The child may develop an altered self-concept, being identified as a ‘carrier of a gene mutation’
- A child who receives a gene-positive result may feel unworthy and unmarriageable
- Testing may cause a loss of self-esteem
- Parents may alter their expectations of the child, affecting the child’s psychological state
- The child may become stigmatised or discriminated against
- Testing may remove the possibility of a child obtaining certain types of insurance later in life
- The child may develop increased anxiety
- The ‘unpatient’ syndrome may develop in a child who is tested, where the child begins ‘sick’ behaviours such as visiting doctors frequently, even though he or she is not yet sick
- Testing may affect the bond that siblings shared when they were all at risk
- Parents may feel guilty about their child’s gene-positive test result
- Testing may place added financial burden on the family
- Testing may uncover findings such as mistaken paternity
A body of literature arguing against predictive genetic testing in young people has therefore evolved. It began as a categorical statement that predictive genetic testing for HD in minors was inappropriate. However, as discussion broadened to incorporate predictive genetic testing for conditions other than HD, the positions moved. Authors remained adamant that testing of immature young people was inappropriate, but only because immature young people could not be involved in the decision-making process. Thus, while arguing for a prohibitive stance regarding the genetic testing of immature young people, these authors were often indirectly supporting the testing of mature young people in some circumstances. There were, at the same time, authors who specifically focussed on objections to testing mature young people. Critically absent from arguments opposing predictive genetic testing in young people is empirical evidence to substantiate the arguments made about the potential for harm.

Of course, as the body of literature opposing predictive genetic testing in young people evolved, there was simultaneously a range of literature emerging in support of such testing. The authors of this second body of literature argued not only for testing of mature young people, but also for testing of immature young people. In the next section I describe their arguments.

1.4 “To test”: Justifications for supporting testing in young people

In this section I review the arguments used to support predictive genetic testing in young people. I begin with arguments used to support testing in mature young people and then turn my attention to arguments used to support testing in immature young people.

1.4.1 The appropriateness of testing mature young people

Many of the arguments used to support predictive genetic testing in mature young people focus on the existence of competent young people, who are capable of making decisions about testing themselves.
Wertz, Fanos and Reilly write in detail about the “emancipated minor” (p 877) who is treated as an adult by law with respect to medical decisions. Citing leading developmental theories and various methods of assessing competence in their paper, they conclude “some minors may benefit from testing carried out to make plans for the future, provided that they themselves initiate the request” (p 882). Clarke and Flinter, while justifying their opposition for testing in immature young people, reiterate such a position. They state that while granting adolescents control over a decision to undertake predictive genetic testing may be beneficial for them, there are no reasons to pre-empt the decision by testing in childhood.

Geller also acknowledges the concept of a mature young person, but argues that competence alone is not enough of a justification for testing minors. Geller indicates that several factors must be taken into account in addition to competence, namely “the implications for ethical reasoning, the economic and social impacts and how remote in time the onset of the condition is” (p 1065). Dickenson writes solely about consideration of competent young people under the age of 18 years. She asserts that “if the young person’s values and identity seem reasonably coherent and secure, then her consent should be honoured” (p 1065). Dickenson notes that this is not only consistent with existing case law, but also with existing guidelines. Binedell and colleagues also focus solely on cases of adolescents who request testing. They argue for an assessment approach to adolescents requesting predictive testing for HD and provide detailed advice on what an assessment should involve.

Elger and Harding discuss competent adolescents, focusing specifically on the case of predictive genetic testing in adolescents for breast cancer predisposition. They write about the need to distinguish between the testing of very young children and the testing of adolescents. They argue that requests by adolescents can even be considered in cases where the adolescent in question is not as competent as an adult may be. This, they argue, is justified by the concept of a “sliding scale” of competence, where competence is task-specific and related directly to the magnitude of harm associated with the related decision. Thus, an adolescent may be competent to make a decision about predictive genetic testing for breast cancer susceptibility, while remaining incompetent to make other decisions that adults routinely make.
Several authors also describe specific benefits that are created by allowing mature young people to make decisions about predictive genetic testing.

Elger and Harding state that “respecting adolescents’ autonomy in decisions concerning genetic testing increases their sense of themselves as active and responsible participants in questions related to their own psychological and physical health rather than as powerless victims of adults and genes” (p 118). Savulescu similarly states that “participation of a child in decisions about testing can promote the development of autonomy” (p 380). Clayton notes that young people “do not emerge form a cocoon at age 18 with full blown decision-making capacity; rather their abilities to make good choices must be developed over time, a process that requires practice” (p 244). Robertson and Savulescu also suggest that “testing could be viewed by some as a necessary step towards the attainment of autonomy” (p 40). Sharpe suggests that “the sincerity of the geneticist in seeking to protect the child’s right to autonomy is not in question. The issue is whether the decision not to test effectively abrogates what it seeks to protect, the child’s personal rights and dignity subordinated to, if not replaced by, objectives, values, and rationality of the geneticist” (p 251).

Several authors refer to benefits that are associated with being able to make realistic plans about the future. Elger and Harding note that:

“For a rational person wishing to pursue rational life plans, knowing whether she is carrying a BRCA1 mutation can be a major benefit because in our society important decisions about career and life orientation have to be made between the ages of 14 to 17 years. A minor may also wish to decide about becoming pregnant. Knowing that she is a BRCA1 mutation carrier, she might decide against a long university education to have children early and pursue a university career after having had bilateral mastectomy and oophorectomy” (p 117).

Savulescu similarly notes that “information about one’s predisposition to disease can be beneficial in a non-medical sense to allow more informed reproductive decision-making, career choice, financial planing and end-of-life decision-making” (p 380). He also mentions that “self-knowledge can promote more autonomous decision making about one’s life” (p 380). Robertson and Savulescu speak of prudential concerns. They state that “knowledge of one’s lifespan and disease susceptibility is
important in prudential terms. If one were going to live to 75, it might be prudent to invest time in long term projects, defer child bearing until one’s career was established, put money aside for retirement, and so on. If this kind of information is of value to adults, why is it not also of value to children?” (p 42).\textsuperscript{39} Michie similarly notes that “there may also be benefits, such as giving more opportunity to prepare psychologically and practically for the future (p 178).”\textsuperscript{41}

In response to concerns about confidentiality when young people are tested, Elger and Harding argue that we should grant adolescents confidentiality. They state that “in the case of adolescents … good arguments exist to grant them confidentiality regarding the results of a BRCA1 gene test… In other medical areas related to sexual behaviour (contraception, sexually transmitted disease), adolescents have already been given the right to confidentiality” (p 116).\textsuperscript{37}

There are also arguments that not testing mature young people could in fact cause harm. Sharpe asks “what of the harm which may result from not administering the procedure? What of the depression and anguish a child could experience due to uncertainty as to risk status?” (p 250).\textsuperscript{40} Binedell and colleagues similarly note that “less attention has been paid to the psychological harm that may be caused by withholding genetic testing” (p 913).\textsuperscript{36}

Box 1.4 summarises the arguments used to support testing in mature young people.
### Box 1.4 Arguments supporting testing in mature young people

- Some mature young people are capable of making a competent decision about testing and therefore these individuals should have access to testing
- Granting adolescents control over a decision about testing may benefit them
- Decisions about testing should be based on competence level, not an arbitrary age
- Testing helps to increase the young person’s sense of themselves as an active participant in their life, rather than a powerless victim of genes
- Testing helps to promote the development of autonomy
- Young people need the opportunity to practice and refine their decision-making skills. This must happen gradually, not all at once as soon as they reach the age of 18 years
- Testing is a necessary step towards to attainment of autonomy
- Not testing young people abrogates the autonomy it seeks to protect, replacing it with the objectives and values of the geneticist
- Information about genetic status is useful to a person wanting to pursue rational life plans such as financial planning, end-of life decision making, and reproductive decisions
- In our society decisions about career are made between the ages of 14 and 17, and genetic information can assist in making these decisions
- Testing young people provides more opportunity for them to psychologically prepare for the future
- Not providing testing could cause psychological harm

I move now to the arguments used to justify predictive genetic tests in immature young people.
1.4.2 Benefits of testing immature young people

Arguments used to support predictive genetic testing in immature young people fall into three broad categories: arguments about parental rights, arguments about benefit to immature young people and arguments about autonomy.

Several authors have argued in favour of parental rights in relation to testing immature young people. These arguments are based on the assertion that parents should have the right to test their children and that, in fact, they are the ones best placed to make such a decision.

Clayton argues that parents are the best decision-makers for their children. She notes that “other adults who deal with the children ought to be sensitive to the desires of the parents, out of deference to the fact that the parents are the people who bear primary responsibility for the child and to the wide variations that exist in views of the good life” (p 236).

Robertson and Savulescu also argue that parents are best placed to make decisions regarding their children’s best interests. Robertson and Savulescu state that “parents know best their child’s social relations, abilities, economic situation, and so on. … it should be the parent’s judgement that carries the greater weight in deciding about the relative impact of such benefits and harms for their child in undergoing testing” (p 46). Savulescu suggests that “testing can show respect for parental autonomy and avoid professional paternalism” (p 380).

Ross and Moon similarly argue:

“Various arguments have been made to support parental control over medical decision making for minor children. First, parents are presumed to have the child’s best interest at heart because they naturally care deeply for their children and because they are in a position to know the child best. Second, for parents to fulfil the responsibilities of child rearing, they need significant leeway in how they rear their children. This includes control over decisions about medical care, provided that the parents and their decisions are neither abusive nor neglectful. In part, this parental discretion is supported by the fact that many of the financial
and emotional consequences of these decisions will be borne most heavily by the child and his or her parents. Third, society has an interest in supporting the family as the primary child-rearing institution. To do so requires that families be afforded a wide degree of privacy and freedom from governmental intrusion” (p 874).²⁷

Meiser writes of parents arguments for testing, stating that “some parents argue that it is their right to have the test done earlier to resolve uncertainty, reduce anxiety and allow them to plan for the future” (p 128).²⁹ Sharpe agrees with the assertion that parents should be making such decisions. He writes

“…may not one argue with equal force that the child’s parents alone can best weigh the social, familial, economic and emotion factors? Has the geneticist effectively imposed upon others her/his own values and beliefs about what is “best” for the child and the family? Why are these deemed more valid, and given a higher priority, than those of the parents who are the child’s legal representatives?” (p 251)³⁰

Robertson and Savulescu also note, in response to concerns about confidentiality when immature young people undergo testing, that “disclosure of such information to parents is hardly a significant breach of confidentiality in itself. … Parents are privy to all sorts of sensitive and personal information about their children” (p 44).³⁹

Other arguments used to justify predictive genetic testing in immature young people focus on the benefits that the provision of such testing will create.

McLean argues that early knowledge of a condition may mean parents can help to prepare their children and help them to adjust.⁴² Ross and Moon argue that “the value of testing children is to minimize uncertainty, to allow a person to incorporate positive or negative status as part of his or her self-concept, and to allow for better lifetime planning” (p 874).²⁷

Savulescu agrees, noting that “early testing may result in better psychosocial adjustment than later testing, when lifestyle and life plans have been firmly established” (p 380).²⁴ Fryer suggests that “having a diagnosis may enable parents to adjust to the circumstances and plan the disclosure of the news to their children…. 
There may be advantages to the child in the parents being able to break the news over a period of time, according to the child’s developmental age” (p 98). Lessick and Faux also agree, arguing that “a significant psychological benefit of genetic testing is the resolution of uncertainty and the opportunity for parents to adjust to the circumstances and plan the disclosure of news to their children over time” (p 39).

Binedell and colleagues note that “outside the field of genetic screening and testing, empirical evidence suggests that being informed of a serious illness at an early age may facilitate adjustment and coping” (p 913). Fryer and Lessick and Faux suggest that testing may facilitate openness in the family, resolving parental anxiety and creating a healthier environment. Savulescu also refers to the possible reduction of anxiety. He states that “testing can resolve uncertainty and consequent anxiety in parents and children” (p380).

Arguments outlining the range of benefits associated with testing immature young people are also accompanied by assertions about the lack of evidence of harm. Robertson and Savulescu highlight that “good evidence to suggest predictive genetic testing in children is harmful is conspicuously missing” (p 42). Michie agrees, noting simply that “we lack the evidence” (p 178).

There are also authors who focus specifically on the issue of autonomy when arguing in favour of predictive genetic testing in immature young people. These authors argue that testing immature young people may enhance their autonomy as opposed to threaten it. They also believe that testing immature young people does no more to alter their future options than not testing them.

Michie responds to the assertion that predictive genetic testing in young people removes young people’s rights to make that same decision as autonomous adults. She argues that it is true young people who are tested lose the opportunity to make the decision themselves as adults, but that young people who are not tested have lost their right to be tested as a child and to grow up with that knowledge.

Robertson and Savulescu similarly note that “the formulation that childhood testing results in reduced options is incorrect” (p 39). They argue that “the child who is not
tested is denied an option of growing up and adapting to the knowledge of their genetic status during their formative years. Thus the choice is not between two courses of action, one which simply has more choice for the later adult, but between two mutually exclusive futures” (p 39).\textsuperscript{39} Robertson and Savulescu state:

\begin{quote}
“More choice is not necessarily better. Is my autonomy enhanced by being offered the option of donating a kidney to my brother? Maybe not. I might prefer that I had never been given this choice. I might be worse off either if I do or do not donate the kidney, than I would have been if I had never been given this choice. It may be rational to prefer not to have such a choice” (p 38).\textsuperscript{39}
\end{quote}

Box 1.5 summarises the arguments in favour of testing immature young people.
Box 1.5 Arguments supporting testing in immature young people

- Parents bear primary responsibility for their children and should therefore be afforded the right to make a decision about whether to have their children tested
- Parents are best placed to know what is in their child’s best interests
- Many of the financial and emotional consequences of testing are borne most heavily by the child and his or her parents, so they should make the decision
- Society has an interest in supporting the family as the primary child-rearing institution. This requires that families be afforded freedom from intrusion
- Testing avoids professional paternalism
- Parents make many decisions on behalf of their children. This is no different.
- Testing reduces uncertainty and anxiety for the parents
- Disclosing results to parents does not constitute a breach of confidentiality (and thus a reason not to provide testing) as parents are privy to all kinds of sensitive information about their children
- Substantial psychosocial benefits are possible
- Early knowledge of gene status may help parents to prepare their children
- Testing will help in planning for the future
- Testing early allows the gene status to be incorporated into the child’s self-concept
- It is better for parents to be able to break the news to their children over time
- Empirical evidence outside the field of genetic testing demonstrates that early disclosure to ill children may facilitate coping
- Testing facilitates openness in the family
- Having bad news is better than living in a family that has become dysfunctional from uncertainty
- There is a lack of evidence demonstrating that testing young people is harmful
- Young people who are not tested lose their right to be tested as a child, and the opportunity of growing up with this knowledge and adapting to it
Arguments in favour of predictive genetic testing in young people have therefore also evolved. Initially, the justifications for testing mature young people expanded, as reasons for opposing the testing of immature young people emerged. However, several authors then began writing specifically about immature young people, making strong arguments in favour of testing this group as well. Again, as was the case with arguments used to oppose such testing in young people, critically absent from arguments supporting predictive genetic testing in young people is empirical evidence to substantiate the arguments that are made about the potential for benefit.

I have now provided a summary of the opinions that individual academics, clinicians and researchers purport about predictive genetic testing in young people. In the next section I review the opinions of larger groups of people, revealed through surveys. The vast majority of these surveys convey the opinions of people who are not academics, clinicians or researchers, but rather people who live in families with a history of a genetic condition. Many are parents.

1.5 Surveys of opinion about predictive genetic testing in young people

The surveys that have been conducted about opinions regarding predictive genetic testing in young people demonstrate a wide range of views. In order to convey these views, I divide the surveys into three categories: (1) surveys of people who have a personal connection to predictive genetic testing, (2) surveys of young people, and (3) surveys of professionals.

1.5.1 Surveys of people with a personal connection to predictive genetic testing

Several surveys have been conducted in order to gauge the opinions of people who have some personal connection to predictive genetic testing. Some of these people have been tested themselves, others have a relative who has been tested and others are living in families with a history of a genetic condition.
Surveys reporting the views of people with a personal connection to predictive genetic testing have found diverse opinions. In some cases, the existence of a medical benefit alters people’s opinions about the acceptability of such testing in young people.

Levy and Richard found that a majority of the parents they surveyed were in favour of testing their own children for von Hippel Lindau, a condition that is characterised by a predisposition to develop a variety of benign tumours and malignant neoplasms. As part of the French von Hippel Lindau Study Group they surveyed 14 people with von Hippel Lindau. Most of the respondents (11 individuals) were willing to have their children tested as soon as possible. Some had already asked for testing. Also, three of these respondents who had developed the condition themselves would not have wanted to know their genetic status earlier, indicating a discrepancy between what people desire for their children and what they desire for themselves.

Benkendorf and colleagues also found great support for testing in young people. They surveyed the unaffected first-degree female relatives of women with breast or ovarian cancer and found that 88% of respondents thought parents should be able to consent to such testing in their minor children.

Umans-Eckenhausen and colleagues demonstrated support for testing young people again. Their participants were parents who were part of a couple where one partner in the couple had a mutation associated with familial hypercholesterolaemia. All couples had children under 16 years. Of the 70 individuals surveyed, 87.1% wanted their children tested. However, familial hypercholesterolaemia is a treatable condition, and therefore this is only indicative of support for tests with a medical benefit.

Patenaude and colleagues found that support for testing in young people increased if medical benefit was an outcome as well. They surveyed 47 mothers with a child who had been diagnosed with cancer. If testing were available 42% would test their children, if the test did not provide any medical benefit. If the test did provide a medical benefit, 91% would test their children.

Several surveys of people with a personal connection to predictive genetic testing also demonstrate opposition to such testing. A working party of the Clinical Genetics
Society in the UK surveyed family support groups in the UK and received 78 responses, some from groups as a whole, others from individuals within the groups. Of the 78 responses received, only 19 were in favour of policy advocating predictive testing in childhood, while 55 were against such policy.\textsuperscript{12}

Hamman and colleagues also surveyed a group who was largely against such testing in young people. They surveyed adults who had undergone predictive genetic testing for breast cancer.\textsuperscript{47} Of the 218 respondents, only 26.1\% thought that individuals under the age of 18 years should be able to undergo such testing for breast cancer, as opposed to 61\% who did not believe they should. Respondents who had children themselves were even more opposed to testing in young people, with 82.7\% against such practice.

Meissen and Berchek reported additional opposition to predictive genetic testing in young people. They surveyed individuals at risk of HD and found that only 35\% would want minors to be tested.\textsuperscript{48}

1.5.2 Surveys of young people

Some surveys have been targeted specifically at young people in order to assess their personal views about predictive genetic testing. When it comes to young people, it seems most have an interest in learning of their genetic status.

Harel and colleagues surveyed high-school students in grades 10-12 about their views on being tested for breast cancer predisposition and hypercholesterolaemia.\textsuperscript{49} Of 361 respondents, 67\% of girls stated a desire to be tested for breast cancer predisposition. Girls who had a relative with breast cancer were significantly more likely to want testing themselves (78\% vs 61\%). The majority of girls (54\%) also stated a desire to be tested for hypercholesterolaemia. This was much stronger than boys desire to be tested for hypercholesterolaemia (39\%). A family history of hypercholesterolaemia also meant that students had a stronger desire to undergo such testing (70\% vs 34\%).

Bernhardt and colleagues surveyed parents and children who lived in families at increased risk for either breast cancer or heart disease.\textsuperscript{50} Children were asked if they would want to be informed of their test result if they had received a predictive genetic
test for breast cancer as part of a research study. Only six of the 37 children (aged 10-17 years) stated they would not want to be informed of the result. The children were also asked the same question in relation to predictive testing for heart disease. In this case, only two of the children would not want to know their test result.

### 1.5.3 Surveys of professionals

Professionals surveyed show wide discrepancies in their views concerning predictive genetic testing in young people, depending on the condition in question, the exact professional body being surveyed and the age of the person being tested. Some of the groups surveyed are professionals currently working in the area of predictive genetic testing while others are in training, but may work in that area in the future.

The working party of the Clinical Genetics Society in the UK surveyed a range of professionals involved in predictive genetic testing including geneticists, co-workers, paediatricians and haematologists. These people were asked about the right of parents to request predictive tests in their children. The majority of the 512 respondents thought parents should be able to request testing in their children. However, large differences in opinion were evident between professional groups. For example, 76% of haematologists believe parents have a right to request predictive testing in their children, but only 57% of geneticists believe this. Only 2 of 49 geneticists would test a 5-year-old for HD, whereas 100 of 260 paediatricians would provide such a test. 12

Rosen and colleagues surveyed paediatric residents by giving participants a vignette and asking them to respond. 51 Of the 64 respondents, 39% would order a predictive test for Huntington Disease in a 10-year-old following a parental request. When asked about testing a 17-year-old for Huntington Disease, 52% stated that they would order a predictive test at the request of parents. If the same test were requested by the 10-year old minor, as opposed to the parents, 44% would provide the test. If the 17-year-old requested the test, as opposed to the parents, 89% would provide the test.

Some surveys of professionals report stronger opposition to testing in young people.
Wertz and Reilly surveyed genetic laboratories who provide predictive testing services. They found that only 26% of laboratory respondents thought parents should have the right to test their children.

Geller and colleagues surveyed women at high-risk of developing breast cancer, nurse practitioners and physicians. They found that only approximately one third of each group would agree to testing a 13-year-old at risk of inheriting a mutation associated with breast cancer.

Taken as a whole, the opinions of people with a personal connection to predictive genetic testing, young people and professionals lend themselves to little generalisation. However, they do provide a glimpse into the array of opinions that exist and provide an indication of the variables that may alter these opinions such as age of the person tested, the existence of medical benefit and the condition being tested for. Broader opinion about predictive genetic testing in young people therefore appears to mirror the diversity of opinion found within the academic community.

When contemplating the findings of surveys such as those reported above, it is also critical to remember that what people say they will (or will not) do is often very different from what they actually do (or do not do) in reality. Take the example of predictive genetic testing for HD. Before such testing became available, surveys were conducted to gain an indication of how many of the individuals at risk of HD would be interested in pursuing predictive genetic testing if such testing became available. Surveys reported that a vast majority of individuals at risk would seek such testing if it were available. However, in the years since such testing has become available, only a minority of individuals who are at risk for HD have in fact sought testing.

In the next section I bring together existing guidelines, arguments and surveys of opinion about predictive genetic testing in young people. I do this with the aim of articulating the core issues in the current debate.
1.6 The essence of the debate

If existing guidelines concerning predictive genetic testing in young people are combined with the arguments that have been published in the academic arena and the surveys of opinion that have been performed, two core issues are seen to recur. These issues are (1) the future autonomy of young people and (2) potential harm to young people.

Just one phrase is common to all the guidelines concerning predictive genetic testing in young people that are summarised above. This phrase is “child’s best interests”. There is no doubt that all protagonists in the current debate have a similar goal, to maximise the well-being (or best interests) of young people. The question is how do we do this? Will providing a test maximise a young person’s best interests or is it withholding a test that will achieve such a goal?

In answering this question, two key areas of discordance recur. Firstly, dissent arises in relation to the concept of young people’s autonomy, or future autonomy. Opponents of such testing in young people argue that testing is unethical because it threatens the (future) autonomy of young people. More importantly, they believe that maximising young people’s best interests entails protecting this future autonomy. However, proponents of such testing have alternative conceptions of young people’s autonomy and what it means to maximise their best interests. Some believe that parental rights override concerns about young people’s autonomy, while others argue that the provision of testing is in fact the best way to enhance the autonomy of young people. Thus the first recurring core theme in the current debate about predictive genetic testing in young people is that of young people’s future autonomy. The specific point of contention sits within theoretical definitions of autonomy, the importance of future autonomy and the ways in which autonomy is best protected and enhanced.

The second key area of discordance to recur in the debate about predictive genetic testing in young people relates to the issue of potential harm to young people and, accordingly, potential benefit. Dissension exists about what will in fact happen if young people undergo such testing. Once again, both opponents and proponents of
testing seek to promote the best interests of young people yet disagree about the ways in which this can best be done. Opponents argue that tests will cause great potential for harm when provided to young people. Proponents argue that testing will create benefits for young people and that harm will be caused by not providing such tests. Thus the second recurring core theme in the current debate is that of potential harm to young people. The specific point of contention here is what will in fact happen if a young person undergoes predictive genetic testing when there is no medical reason for the test.

In this thesis I focus upon the second core theme in current debates about predictive genetic testing in young people. I am concerned with the dispute about potential harm to young people and the conflict of opinion that exists about what will in fact occur if young people undergo predictive genetic tests for non-medical reasons.

In the introductory section of this chapter I introduced the terms ‘mature’ and ‘immature’ young people, clarifying the meaning I ascribe to each of these. In this thesis, although I refer to arguments and research concerning both of these groups, my primary focus lies with mature young people. That is, those for whom an informed decision about predictive genetic testing is possible. This choice is precipitated by a desire to address the aspect of the current debate that is most opaque.

Predictive genetic testing in adults for non-medical reasons constitutes routine practice. Such testing is widely accepted and is non-controversial. Current guidelines demonstrate strong support for such testing and this is reflected in current practice. Current guidelines are also unanimous in the stance they purport about predictive genetic testing in immature young people, for non-medical reasons. Such testing is spurned in existing guidelines. However, current guidelines remain ambiguous in reference to mature young people. Too old to be classified as immature but not yet formally recognised as adults, this group creates indecision. Opposition to such testing in mature young people exists as the default position in existing guidelines, yet this opposition is far from absolute. In fact, opposition to predictive genetic testing in mature young people is tentative at best. It is therefore mature young people I choose to focus upon in this thesis. I do this with the aim of removing the ambiguity that
surrounds predictive genetic testing in mature young people and replacing it with empirical certainty.

1.7 Summary

In this first chapter I have set the scene around current debates concerning predictive genetic testing in young people. Beginning with a summary of the existing guidelines, I described the kinds of arguments used to oppose testing in young people. I then turned my attention to the range of arguments used to support testing in young people, articulating in both cases that the arguments used were not substantiated by empirical evidence. Next I brought these arguments together as a whole, combining them with surveys of opinion, and described the two core themes that recur in current debates about predictive genetic testing in young people: autonomy and potential harm. Finally, I clarified the aspect of the current debate that I choose to focus upon from herein. In this thesis I am concerned with the issue of potential harm to young people as a consequence of predictive genetic testing for non-medical reasons. I direct this concern towards the ambiguities surrounding such testing in mature young people.
CHAPTER TWO: EMPIRICAL EVIDENCE
2.1 Introduction

“It was a really hard 2 years waiting to turn 18, but the day I did turn 18 I got my blood taken...I think if your parent has it and you’ve seen the consequences of it, I think you have every single right, no matter what age, to get tested”

Ella:F:20:HD:18:-ve

“It’s hard to argue when people have grown up you know, some take 30 years and others take 13 ... there’s a wide range of ages at which people are mature enough to talk about serious issues”

Oliver:M:24:HD:19:+ve

“When you’re that age you’re almost treated like an adult anyway. You’ve got to choose subjects at school that are going to affect the rest of your life, you’ve got to choose courses you’re going to do at uni that are going to affect the rest of your life, so if you’re old enough to do that sort of thing, you’re old enough... having the test is part of your life”


A clash in perception is taking place. Some perceive predictive genetic testing in young people to be too potentially harmful to allow. Others perceive such testing to be an opportunity for the promotion of benefit, even an opportunity for the prevention of harm. Both perceptions arise from significant expertise and neither group of perceivers is willing to concede. Extensive discourse has failed to resolve the clash. Here, I look to empirical research about the impact of predictive genetic testing in young people in the hope of finding evidence to resolve the current debate.

I begin with a review of the empirical evidence that exists in relation to predictive genetic testing in young people for non-medical reasons. I review the one study that exists within this category. Given the lack of available evidence, I then turn my
attention to two similar, but not identical, research fields and review the evidence available within these. The first is the field of predictive genetic testing in young people for FAP, a test provided for medical reasons. The second is the field of predictive genetic testing in adults for HD, a test that is provided for non-medical reasons.

2.2 Evidence about testing young people for non-medical reasons

There are two papers that have been published reporting the effects of predictive genetic testing in a young person for non-medical reasons. However, both papers refer to the same case study, that of a 5-year-old girl who underwent predictive genetic testing for Maturity Onset Diabetes of the Young (MODY). This therefore remains the only case study reported in which a young person has undergone the type of predictive genetic test that is specifically recommended against in current guidelines.

Individuals who are gene-positive for MODY have a 63% chance of developing diabetes, usually before the age of 25 years. Although current guidelines permit the testing of young people for conditions that often present in childhood, there remains controversy about the testing of young people for MODY.

The two papers I refer to describe a family with a history of MODY, consisting of two parents and two daughters. Following the diagnosis of diabetes in their 11-year-old daughter, the parents decided that they wanted their 5-year-old daughter to undergo a predictive genetic test for MODY. The main motivation for the test, as stated by both parents, was to reduce uncertainty. The parents also felt it would be easier to tell their daughter over time, if she was to receive a gene-positive test result, and that testing would give the family a chance to accept what was going to happen. Given the controversial nature of the predictive genetic test much discussion ensued between the family and various professionals. This resulted in the decision to provide the test.

As this was the first known case of predictive genetic testing for diabetes, researchers decided to perform in-depth interviews with the parents, both children and a range of
professionals who were involved in the case. These professionals included the consultant geneticist, the genetics nurse specialist, the paediatrician, the diabetologist and the paediatric diabetes specialist nurse. The interviews were performed prior to testing and 6 weeks after receipt of the test result.

Following receipt of the gene-positive test result, the father reported feeling “a bit guilty” as he felt that it was his fault his daughter had received a gene-positive test result (p 253). He also noted that this was short lived, lasting no more than a day. The mother described that although she felt initially disappointed by her daughter’s gene-positive test result, the “nagging feeling” at the back of her mind had ceased (p 254). Several of the professionals who were interviewed stated that the parents looked more relaxed and confident after the test. The mother confirmed this observation, stating that she was certain they had made the right decision in deciding to have their daughter tested. The father similarly noted that “now we know the results, the suspense has gone” (p 254). The authors stated that there appeared no need to worry about the child being labelled as ‘sick’ or treated differently as it seemed both parents were aware of the importance of treating their daughter normally. The parents noted that they “don’t want to take her childhood away from her just because this is going to happen in about 3, 4 or 5 year time” (p 255). They wanted their daughter to “have a normal childhood and not be burdened with it all too much” (p 255). Neither of the two papers published concerning this case study explicitly state that the 5-year-old girl was informed of her gene-positive test result. However, it is noted in the methodology sections that the 5-year-old girl was one of the family members to be interviewed by researchers during the research process. One could therefore infer that she received some explanation about the process she was undergoing.

Given that only one study exists concerning the effects of predictive genetic testing in young people for non-medical reasons, I now turn my search for empirical evidence to two similar, though not identical, fields of research. The first field encompasses research into the effects of predictive genetic testing in young people where testing does provide a medical benefit, that is, predictive genetic testing for FAP. Although the motivation for testing in these cases is different from those where medical benefit is not a possibility, it is reasonable to assume that the impact of these tests may be similar for the young people involved, whether medical benefit is an outcome or not.
In other words, the fact that a predictive genetic test provides a medical benefit does not remove the potential for harm (or benefit), it simply overrides this (and thus testing in these circumstances is permitted). The second field encompasses research into the effects of testing adults for non-medical reasons, specifically HD. Although the participant population in this field is once again different from those I am primarily concerned with (young people) it has the value of maintaining a focus on tests that do not provide medical benefit. Thus although it is likely that young people will react differently from the way in which adults react following a predictive genetic test, this field of research can provide some insight into the types of effects that occur when predictive genetic tests are provided to individuals for non-medical reasons.

2.3 Evidence about testing young people for medical reasons

There are four papers published that have reported the effects of predictive genetic testing in young people, where tests were performed because of the medical benefit associated with such a test. All of the tests described in these papers were performed to identify individuals who are gene-positive for FAP.6,62-64

The first of the three empirical studies was a case study.62 This case study involved a couple who had their two-year-old and four-year-old daughters tested for FAP. One daughter received a gene-positive test result while the other received a gene-negative test result. The couple was interviewed prior to testing, two weeks after testing and again 15 months after testing. Michie and colleagues report that, for this couple, the experience of testing their children for FAP was a valuable one. The couple’s anticipated reactions were good predictors of their actual reactions and it is reported that there was much evidence of psychological preparation. No obvious harms occurred for the couple and the knowledge of their children’s genetic status appeared to be beneficial for them. It is important to note that the couple decided to wait to inform their children of the test results over a gradual period as they matured. This means that at the time this research was performed the children involved were unaware of their predictive genetic test results.
The second empirical study assessed the psychological effects of predictive genetic testing in young people who were tested for FAP.\textsuperscript{63} This study assessed 41 children between the ages of six and 17 years, as well as their parents. Children and their parents were assessed prior to testing and three months after receiving the test results. It was found that mean scores for depression, anxiety and behavioural problems in the young people remained within the normal range after testing. Parents’ depression scores also remained within the normal ranges. However, some sub-clinical yet significant increases in depression were noted in young people who received a gene-positive test result and also had an affected mother. Also, sub-clinical yet significant increases in anxiety were noted in young people who had an affected mother, regardless of whether they had received a gene-positive or gene-negative test result. Unaffected parents who lived in families with both a gene-positive and gene-negative child demonstrated sub-clinical yet significant increases in depression. Finally, all young people showed a significant decrease in behavioural problems following testing.

The third empirical study assessed the anxiety and distress levels of 60 young people who were tested for FAP and compared these with the same measures in 148 adults who were also tested for FAP.\textsuperscript{64} It was found that young people who received a gene-positive test result displayed anxiety and distress levels within the normal range, while 43% of adults receiving a gene-positive test result displayed clinically significant levels of anxiety. Over the year following the receipt of test results, young people who received a gene-positive test result displayed no change in their levels of anxiety, depression or self-esteem. Young people who received a gene-negative test result displayed a decrease in anxiety and an increase in self-esteem.

The fourth empirical study assessed symptoms of depression, anxiety and behavioural problems in 48 young people who were tested for FAP.\textsuperscript{6} The parents of these young people were also assessed for measures of depression. Assessments were performed prior to testing and 3, 12 and 23-55 months after testing. This study reported no clinically significant changes in the mean psychological test scores of either the young people who were tested or their parents. There were subclinical increases in depression in the group of young people who received a gene-positive test result and also had a gene-positive sibling. Also, several individual young people who received
a gene-negative test result but had a gene-positive sibling also demonstrated clinical elevations in anxiety. Behavioural problems decreased for all groups.

Thus the (minimal) body of empirical research reporting effects of predictive genetic testing in young people for medical reasons reports a largely beneficial group of experiences. The only instance of clinically significant negative results across an entire group occurred in adults who were tested for FAP and this related to levels of anxiety. One cause for concern in young people is the finding that some young people who have a gene-positive sibling and underwent testing themselves were found to have subclinical increases in depression and clinical increases in anxiety.\(^6\)

Beneficial effects of testing in young people for medical reasons included decreases in behavioural problems and decreases in anxiety.\(^{6,63,64}\)

### 2.3.1 Benefits and harms previously researched in young people

Of the six empirical papers that have been published concerning the impact of predictive genetic testing in young people, only three report the outcomes of testing in young people specifically.\(^{6,60-64}\) The other studies are case-studies where the young people tested were too young to be properly informed of the test result at the time, meaning that the primary descriptions of test outcomes refer to descriptions provided by the parents of these young people.

There are two fundamental problems with the range of benefits and harms that have been measured within these three empirical studies. Firstly, the range of potential benefits and harms that have been measured is narrow. The potential outcomes of predictive genetic testing in young people extend far beyond depression, anxiety and behavioural problems. Secondly, the majority of outcomes that have been researched have been harms of testing, as opposed to benefits. In fact, the only two benefits that have been empirically tested, optimism and self-esteem, were measured in order to determine if these traits had a confounding effect on the harms associated with testing.\(^{64}\) Therefore, not a single empirical study has been conducted with the specific aim of measuring benefits of predictive genetic testing in young people. If we are ever to reach a balanced understanding of what predictive genetic testing is like for young people who experience it, we must seek to find out about the good and the bad,
the positive and the negative, the beneficial and the harmful. If we seek only to
document the harms associated with testing, harms are all that we will find.

There are at least six possible explanations for the failure of empirical researchers to
adequately address the possibility of beneficial outcomes in addition to harmful ones.

Firstly, researchers may believe that finding evidence of harmful effects (or a lack of
evidence of harmful effects) will be more powerful than finding evidence of
beneficial effects (or a lack of evidence of beneficial effects). Thus, researchers may
choose to research harms initially upon the assumption that if harm is demonstrated,
predictive genetic testing in young people will no longer be contemplated (unless it
provides a medical benefit) and current debates will cease. Similarly, researchers may
assume that demonstrating the existence of benefit would not produce the same level
of impact. In fact, perhaps even researchers hoping to lend support for the testing of
young people feel that an absence of harmful effects will be more powerful in lending
support than the existence of beneficial effects. In other words, a perception may exist
that no amount of demonstrable benefits could ever override the existence of
demonstrable harms.

The second, more concerning, possible explanation is that researchers have
preconceived ideas about the types of impacts that predictive genetic testing will
have. Perhaps there exists an assumption that such testing will cause harm and that
the potential for benefit is minimal. Thus, driven by this assumption, it is only the
harms that are sought through empirical means.

The third possible explanation is that researchers are simply choosing to test the
hypotheses that are most prominent in current debates. That is, perhaps researchers
feel that the primary concerns about predictive genetic testing revolve around the
potential for depression, anxiety and behavioural problems in young people who are
tested and they therefore choose to research these outcomes in order to address the
core of the debate. Thus their choice of outcome measures may have less to do with a
perceived division between harms and benefits and more to do with addressing the
most prominent reasons for the current prohibitive stance, regardless of the quality of
these.
The fourth possible explanation is that researchers are choosing to use the most standardised and widely accepted instruments for assessing psychological traits. Instruments for measuring depression and anxiety are common in psychological studies and are easy to administer to larger numbers of people. Thus, perhaps instruments measuring anxiety, depression and behavioural problems constitute the most effective means for an initial fast, efficient glimpse into the impact of predictive genetic testing in young people. The use of widely accepted, standardised assessment instruments also maximises potential for comparisons between study populations.

The fifth explanation revolves around past patterns of research in relation to predictive genetic testing in adults. Perhaps researchers have chosen to measure outcomes such as anxiety and depression because these are the types of outcomes that have been most used in research concerning the testing of adults for conditions such as HD. In other words, researchers may simply be following a widely accepted and utilised trend in empirical measurements related to predictive genetic testing.

The sixth explanation is that researchers believe the outcomes they have chosen are able to demonstrate both harm and benefit. For example, when anxiety is measured there is the possibility of demonstrating an increase in anxiety or a decrease in anxiety. It may therefore be argued that an increase in anxiety provides evidence of harm and a decrease in anxiety provides evidence of benefit. A similar argument could be made in relation to measures of depression or behavioural problems. However, in order for this to be possible, measures would have to be taken at baseline, before testing was provided and then again after testing. Only two of the studies I refer to above took measures at baseline.  

Whatever the explanation for the narrow range of outcomes that have been measured empirically thus far in relation to predictive genetic testing in young people, this range must expand. It is time for a broad variety of potential harms and benefits of predictive genetic testing in young people to be sought, so that a balanced understanding of the ways in which testing impacts upon young people may be composed.
In the next section I turn my search for empirical evidence to the field of predictive genetic testing in adults for HD.

2.4 Evidence about testing adults for Huntington Disease

Predictive genetic testing for Huntington Disease has been available since 1986 by linkage analysis, and since 1993 by direct gene testing. No other predictive genetic test has been offered for such a length of time. The body of literature documenting the impacts of such testing in adults is thus vast, spanning almost 15 years. It is therefore this field of empirical research that I choose to summarise in order to gain an insight into the impacts of predictive genetic testing in adults for non-medical reasons. I begin with a broad overview of the research findings within this field, followed by a more detailed account.

2.4.1 An overview of research concerning predictive genetic testing for HD

Research into the effects of predictive genetic testing in adults for HD began to emerge in the early 1990’s. Initial studies reported the effects of testing in small groups of people (usually under 100). Often, these reports focused on case studies, describing the impact for these individuals in detail. However, in 1999 a seminal paper was published by Almqvist and colleagues, reporting the findings of the first large-scale study. This study researched the adverse effects of such testing in 4,527 individuals.

More studies of smaller sample size again followed this paper, many providing qualitative data about the experiences of individuals undergoing testing. As well as describing the experiences of these individuals personally, research began to broaden, incorporating the impact of testing for partners of the individual tested and also the family as a whole. Attempts also began to identify ways to predict the type of reaction an individual may have to testing.
Findings have now been published concerning long-term consequences of predictive genetic testing in adults for HD. Timman and colleagues recently published research reporting 7-10 years of follow-up in 142 individuals.\(^{67}\)

This research into the impact of predictive genetic testing in adults for HD has yielded some surprising, yet pleasing, results. When such testing was initially offered fears existed about the potential for catastrophic events such as suicide, attempted suicide and psychiatric hospitalisation. Of great concern was the possibility that individuals may commit suicide upon receipt of a gene-positive test result. However, these fears have not been confirmed with empirical evidence.\(^{68,69}\)

It has been shown that in the short-term period following predictive genetic testing in adults for HD, those receiving a gene-positive test result are likely to experience a range of unfavourable outcomes.\(^{65,70,71}\) However, over time these dissipate, leaving little difference in the psychological functioning of those receiving gene-positive test results and those receiving gene-negative test results.\(^{65,70-74}\) In fact, individuals receiving both gene-positive and gene-negative test results have demonstrated decreases in psychological distress.\(^{65}\)

### 2.4.1.1 The need for caution

Although the overall picture of predictive genetic testing in adults for HD may seem generally positive, caution is required. All the research conducted thus far has involved participants who chose to undergo testing themselves and it has been noted that this may reflect a participant population that is particularly psychologically resilient.\(^{75}\) It may be that if less psychologically robust individuals were to undergo predictive genetic testing for HD, more adverse effects would occur. Recently it was reported that many of the individuals who drop out of research studies are those with high levels of psychological distress, as evaluated prior to drop out.\(^{67}\) This strengthens notions of past research over-representing individuals whom are particularly psychologically robust.

Also of concern is the recent, most long-term, research project to be published. This study reports that pessimism may begin to increase as the onset for HD approaches.\(^{67}\) As this study represents the most long-term study to date, it is possible that as more
long-term follow-up is conducted, more adverse effects may be identified in the future. However, it is also possible that the pessimism described may in fact be related to early symptoms of HD. Conclusions therefore cannot be drawn until similar long-term research is performed with a matched group of individuals who are at risk for HD but have not undergone testing, and an untested group who are symptomatic.

2.4.2 A detailed account of research concerning predictive genetic testing for HD

Given that literature about the impact of predictive genetic testing in adults for HD is so vast, I divide it into distinct categories. I begin by describing the impact that testing has for individuals. I then turn my attention to the impact that testing has on the broader family. Finally, I summarise attempts to identify predictors of individual’s reactions to testing.

2.4.2.1 The impact of testing for individuals

Much of the research published concerning the impact of predictive genetic testing for HD conveys findings that are somewhat counter-intuitive. Individuals who receive a gene-positive test result often cope better than expected, while individuals who receive a gene-negative test result often experience difficulties that were not foreseen.

Bloch and colleagues describe 4 case studies concerning people who have received a gene-positive test result. They report that although depression and anxiety tend to increase in the 2 months post-testing, individuals appear to have less depression and a “heightened perception of the here and now” a year after their test. Huggins and colleagues (as part of the same broader research group) report findings specifically related to people who have received a gene-negative test result. They report that, contrary to expectations, approximately 10% of the 105 individuals they assess have psychological difficulties coping with their test result. Wiggins and colleagues (again as part of the same research group) surveyed 135 individuals prior to testing, 6 months after testing and 12 months after testing. They found that those receiving a gene-negative test result demonstrated lower levels of distress and those receiving a gene-positive test result demonstrated no change. At the 12 month follow-up, both those receiving a gene-negative and those receiving a gene-positive test result had lower scores of depression and higher scores of well-being than those who
did not receive a result (either because they chose not to undergo testing or because their result was uninformative).\textsuperscript{77} Lawson and colleagues (again as part of this same research group) reported follow-up for a year after 135 individuals underwent predictive genetic testing for HD.\textsuperscript{78} They reported that 20 individuals (14.8\%) had experienced an adverse event. An adverse event was considered to have occurred if clinical or quantitative criteria were met. Clinical criteria used to define an adverse event included: a suicide attempt or the formulation of a suicide plan, psychiatric hospitalisation, depression lasting longer than two months, a marked increase in substance use, or the breakdown of important relationships. To meet quantitative criteria for an adverse event, subjects had to have recorded a measurable change in the scores obtained on both the General Severity Index of the Symptom Checklist-90 and the Beck Depression Inventory. Lawson and colleagues also reported that that no significant difference existed between the rate of adverse events seen in those who had received a gene-positive test result and those who had received a gene-negative test result. Of great interest was the finding that the timing of adverse events differed between these two groups. For individuals receiving a gene-positive test result, adverse events tended to occur in the first 10 days, but for those receiving a gene-negative test result all adverse events occurred six months or later after receiving results.\textsuperscript{78}

The findings that individuals receiving gene-positive test results generally cope well was initially greeted with some scepticism. It was surmised that perhaps these individuals were in denial. Decruyenaere and colleagues report that, of the 53 individuals they assessed before testing and one year after testing, individuals receiving a gene-positive test result demonstrated no change in measures such as depression and anxiety. They also report that those who received a gene-negative test result experienced a significant decrease in these measures.\textsuperscript{79} However, Decruyenaere and colleagues found that testing had an impact on the reproductive decisions of those tested. Therefore, if individuals were altering reproductive plans in response to a gene-positive test result, it was not possible that they were in denial about their result. Thus the ‘no-change’ finding on psychological measures following gene-positive test results began to hold more credence.
Tibben and colleagues found that, of the 18 individuals they retrospectively assessed, most of those receiving a gene-positive test result were functioning well. They noted too that most of the individuals receiving a gene-negative test result had failed to experience the relief they had predicted would occur. Many experienced numbness after testing. Tibben and colleagues, in another study of 63 individuals assessed for 6 months after testing, reported again that those receiving a gene-positive test result did not experience the sense of control over their future that they had expected. An absence of relief and numbness were also noted again in relation to those receiving a gene-negative test result. Nance and colleagues similarly report that, in the first 19 people ever to undergo predictive genetic testing for HD at their service in Minnesota, most individuals receiving a gene-negative result were relieved, although two experienced guilt and concern about other family members.

Research concerning the impact of predictive genetic testing for HD in individuals has also attempted to demonstrate the diversity of effects.

Codori and Brandt surveyed 68 people who had been tested for HD and provide qualitative data of their experiences. They report both good and bad outcomes for gene-positive and gene-negative individuals. For gene-positive individuals, beneficial effects included a greater closeness in the family and financial security, while harmful effects included worry and guilt. For gene-negative individuals, beneficial effects included knowing that their children would not develop HD, while harmful aspects included disappointment that the gene-negative result had not been able to eliminate other, unrelated issues in their lives. Most did not regret being tested.

Wahlin and colleagues also aimed to highlight the differences that occur between individuals in their reactions to test results. They chose 4 case studies: 2 individuals who received a gene-positive test result and two individuals who received a gene-negative test result, to illustrate the diversity of individual reactions. The reactions to gene-positive test results included a need to take long sick leave and undergo regular psychotherapy sessions, and a healthy re-orientation in life and a decision to undertake a new course in study. Reactions to gene-negative test results included great difficulty and a need for much professional support, and a move from unemployment into a successful business.
The largest and arguably most influential research to date in this field is that conducted by Almqvist and colleagues.\textsuperscript{66}

Almqvist and colleagues reported a study that involved collaboration of 100 predictive testing centres in 21 different countries.\textsuperscript{66} They reported that the rate of catastrophic events (defined as suicide, a suicide attempt or psychiatric hospitalisation) in 4,527 individuals who underwent testing for HD was 0.97% (or 44 people). This consisted of 5 suicides, 21 suicide attempts and 18 psychiatric hospitalisations. Of great significance was the fact that all individuals who committed suicide had symptoms of HD. Also, 52.4% of those attempting suicide were symptomatic, as were 44.4% of those requiring psychiatric hospitalisation. Also of importance was the finding that, of the individuals who were not symptomatic but experienced a catastrophic event, 84.6% had received a gene-positive test result. In fact, the rate of catastrophic events in those receiving a gene-positive test result was 2%, which was significantly higher than the rate amongst those receiving a gene-negative test result (0.3%).\textsuperscript{66}

When the Almqvist study was published, an editorial accompanied it by Bird.\textsuperscript{2} Bird stated that although the suicide rate reported by Almqvist and colleagues was approximately 10 times the rate in the USA, it coincided with previous suicide rates for individuals symptomatic for HD, which varied from 7-10 times the USA average. This is critical because it implies that the suicides Almqvist and colleagues refer to may have occurred in the future, despite the provision of predictive genetic testing for HD. Of course, the fact that predictive genetic testing may have brought forward these suicides remains of great significance.

\textbf{2.4.2.2 The impact of testing upon the broader family}

Two key papers have been published that focus specifically on the impact of testing upon the family as a whole. Both of these papers are authored by Sobel and Cowen and refer to interviews with the same 18 families, in which 55 individuals were interviewed.\textsuperscript{85, 86} Together, these papers indicate that predictive genetic testing is a “family matter” as opposed to an individual one. It was reported that 81% of families experienced changes in family membership, and approximately half of families
experienced changes in patterns of communication and care giving concerns about the future. Data collected from a post interview questionnaire indicates that families need to address “unfinished business” concerning the decision to be tested, reorganise patterns of communication and revise family stories to provide a meaning for HD.

Several studies have also been conducted to assess the impact of testing on partners of the individual who is tested.

For some partners, the predictive genetic testing process is difficult. Tibben and colleagues report that partners of the 18 individuals they assessed retrospectively often felt neglected by friends and family. Tibben and colleagues also note that partners are at risk of being isolated during the testing process. Quaid and Wesson examined the effects of testing on 19 couples. They found that, prior to testing, spouses were significantly more depressed than the at-risk individuals. Williams and colleagues noted that testing can be difficult for spouses. They interviewed 18 adults about their experience of the testing process and found that spouses recognised that disclosure of test results also revealed something about their own future.

However, other studies report that predictive genetic testing for HD has little negative impact on couples. Richards interviewed 14 couples and noted that most couples reported that the predictive genetic test had little or no adverse effect on their relationship. In a second study by Richards and colleagues, 23 couples who were undergoing testing were compared with 20 couples who were not undergoing testing, but where one person in the couple was at risk of HD. They reported that no significant differences were found in marital adjustment between couples undergoing testing and couples not undergoing testing. Decruyenaere and colleagues reported on the impact that testing had on relationships 5 years after testing. They note that marital status was unchanged after 5 years for 70% of people who were tested. However, they also report that of the 12 individuals whose marital status was changed, one gene-positive individual and two gene-negative individuals attributed the breakdown of their relationship to the test result.
2.4.2.3 Identifying predictors of an individual’s reaction to testing

Several studies have aimed to identify predictors of how an individual will react after disclosure of their predictive genetic test result. These report a wide array of predictors and few conclusions can be made at this time.

Avoidance of HD-related situations and feeling unsatisfied with the available support prior to testing has been associated with avoidance behaviour, becoming depressed and becoming suicidal after testing.\(^\text{92}\) Ego strength, depression level and coping strategies have also been suggested as useful traits for predicting post-test reactions.\(^\text{79}\) Additional factors associated with being well-adjusted after testing for HD include receiving a gene-positive test result, being married, having no children and being closer to the estimated age of onset.\(^\text{93}\)

Individuals who are depressed prior to testing for HD and other cancer syndromes have been shown to be more distressed after testing.\(^\text{94}\) However, those who were anxious prior to testing have been shown to have less intrusive thoughts after testing. Additional factors associated with having more intrusive thoughts after testing included being female, having children and having intrusive thoughts prior to the test.\(^\text{94}\) Religion has also been associated with less intrusive thoughts after testing.\(^\text{94}\)

Test result, age and gender have been shown to be bad predictors of reactions after testing.\(^\text{79, 94}\)

2.5 Conclusion

In this chapter I have reviewed the empirical research concerning the impacts of predictive genetic testing in young people. I began by summarising research concerning the impacts of testing young people for non-medical reasons. However, given the extreme lack of such research, I then examined two related, but not identical fields. Firstly, that of predictive genetic testing in young people for FAP and, secondly, that of predictive genetic testing in adults for HD.
Taken as a whole, the body of research that describes the impact of predictive genetic testing for FAP in young people and for HD in adults tells a positive story. The outcomes of such tests are not as grave as initially anticipated and, for the majority of individuals who request testing and are provided with pre-test and post-test counselling, the process appears to be a beneficial one for them.

Extrapolating the findings from these two similar fields to predictive genetic testing in young people for non-medical reasons could therefore lead us to believe that the likely effects of testing in young people may also be largely beneficial. We may also extrapolate these findings to assume that predicting a young person’s reaction to a test result may prove difficult and that such tests will not only impact upon the young person who is tested, but also upon the broader family. However, without empirical evidence specifically about the effects of testing young people for non-medical reasons, it is impossible to accurately anticipate how such testing will impact upon this group.

There is a pressing need for empirical evidence concerning the effects of predictive genetic testing in young people for non-medical reasons. This research must be long-term, as research into the impacts of testing adults for HD has recently indicated that the consequences of testing may change as the time of onset approaches. It is also vital that assessments are made both prior to testing and after testing, so that the impacts of such testing may be compared to base-line measures. Outcomes measures must include a range of both harms and benefits.

Until empirical research is performed concerning the impacts of predictive genetic testing in young people for non-medical reasons, the clash of opinion that is currently playing out in the academic arena will remain at the impasse it has reached. At this time, empirical evidence is not able to substantiate either support or opposition for such testing in young people.
3.1 Introduction

“If you’re too young you don’t really have much of an understanding”

Amy:F:19:FAP:14:-ve

“You’re a kid, you find out something, you blab it, it doesn’t matter how bad, and then people start teasing you cause... kids ...are little brats”

Kylie:F:20:FAP:14:+ve

“Being 14 may be a bit tough just being, you know, in the middle of puberty or even the start of puberty, it is something that can be difficult to understand at that age”


A line has been drawn between young people and adults in current discourse surrounding predictive genetic testing. Adults can access such tests with ease, yet the possibility of granting young people similar ease of access incites passionate debate. In this chapter I consider the reasons for the distinction that has been made between young people and adults, in order to determine whether it is indeed a necessary distinction. Firstly I focus upon the psychological differences between young people and adults. That is, the ways in which the minds, desires and behaviours of young people differ from those of adults. This first part is concerned with qualities that come from within young people that cause them to be different from adults. Secondly, I concentrate on the social differences between young people and adults. Here I pay attention to the ways in which society influences young people and causes them to live different types of lives from those of adults. This second part is concerned with qualities that originate outside young people that cause them to be different from adults. I conclude that the features of young people that differentiate them from adults may cause them to react differently from adults in response to a predictive genetic test.
However, I argue that it is not clear if these differences will necessarily create a greater potential for harm or a greater potential for benefit.

In order to gather the literature required for describing the psychological and social aspects that make young people different from adults, my aim was not to be exhaustive. Instead, the literature provided is taken primarily from text books concerning the development of young people and from the articles concerning predictive genetic testing that were reviewed in chapter one. My aim here was to provide a glimpse into the types of differences we know may cause young people to react differently from adults in response to a predictive genetic test. My aim was not to provide a comprehensive review of psychological and social features of being young, but to touch on some relevant issues.

3.2 Psychological features of being young

In this section I explore the ways in which the mental processes of young people are different from those of adults. Here I am concerned with the intrinsic qualities of young people, causing them to be different from adults. Although there are many aspects of mental life, my focus upon predictive genetic testing narrows the field of possibility. That is, my attention is drawn to the psychological qualities of young people that may cause them to react differently from adults in response to a predictive genetic test. I begin here with a brief discussion of the physical basis of psychological development and then turn to three aspects of the psychology of young people: competence, self-identity and sexual maturity. For reasons I will show, these facets of being young are highly relevant to predictive genetic testing. I aim here to give an overview of what is accepted within the field of psychology. My aim is not to delve into the more disputed or contentious areas.

3.2.1 Physical development of the brain

In order to understand why young people’s minds may function differently from the minds of adults, it helps to have some understanding of the biological nature of brain development. However, my primary focus in this section on youth psychology is not the physical development of the brain, but rather the psychological manifestations of
this physical development. My summary of brain development is therefore simple and brief. I aim to do no more than provide a small scaffold on which to place the concepts that proceed.

At the time of birth, the human brain is closer to its adult size than any other organ in the body. However, at birth humans have achieved only 23% of their ‘adult’ capacity, leaving the real burden of brain development for after birth. At birth, the most completely developed structures in the brain are the midbrain and the medulla. These parts are responsible for regulating basic tasks, including sleeping, attention and movement of the head and neck.

We are born with a great excess of neurones in our brains. As development progresses, these neurones form connections with each other, known as synapses, and as these connections are developed and strengthened through use over time, neurones that are not used and strengthened die off. This in turn leaves room for new synapses to develop and strengthen as required. This process is termed ‘synaptic pruning’ and is thought to begin around the age of 2 years, continuing through adolescence.

As development occurs and pathways are refined, glial cells are also produced in addition to the synapses. Glial cells produce myelin and this is involved in protecting the neural fibres, increasing the speed of transmission of electrical impulses through a process known as myelinisation. Thus, as pathways that are not used are lost, those that are used are strengthened and protected. From birth to maturity then, the brain goes through a complex process of creating, pruning and strengthening neural pathways.

While neurones are developing synapses and these connections are being strengthened, the cerebral cortex also continues to develop around the brain. The cerebral cortex accounts for approximately 85 per cent of total brain weight and consists of convoluted grey matter that wraps around the midbrain. Importantly, the cortex is the least developed part of the brain at birth. It is the last part of the brain to stop growing and is therefore thought to be the most sensitive to environmental influences. Specific sections of the cerebral cortex have specific functions.
Our brains, our experiences and our behaviours are all linked. Although much of brain development is innate, without the right environment, correct development will not occur. If newborns are brought up in environments of gross deprivation, their development is delayed. In other words, the brain must be exercised in order for neurological development to occur. For example, children brought up in complete darkness never develop the precise visual pathways seen in children who are not deprived of light. The developing brain also has a degree of plasticity where, if one part of the brain is damaged in some way, another part can take over the function that has been harmed. Plasticity is dependent upon age. That is, the older we get, the less capable our brains become of compensating for damage.

In the past it was assumed that brain development was largely completed during early childhood. It is now known that this is not the case. Several key structures of the brain do not mature until we are in our twenties. Research into brain development has found that generalised growth spurts of the brain appear to occur at particular times of our development. These also appear to coincide with performance on intelligence tests and transformations in cognitive competence.

The brain is divided into two hemispheres and within each of these hemispheres there exist four lobes. During adolescence the parietal lobe, frontal lobe and temporal lobe of the brain continue to develop. The parietal lobe is associated with problem solving, including spatial problems, while the frontal lobe is associated with higher order processes including planning and impulse control. The temporal lobe is associated with language development. Thus, during adolescence, three of the four lobes of the brain are still maturing. The last part of the brain to develop is the frontal lobe. The connection between the two hemispheres, known as the corpus callosum, also thickens during adolescence.

Thus adolescence is not simply a time where experience is collected and skills are practiced in preparation for entry into the adult world. Physical brain development continues to take place at this time of life, impacting upon thoughts, behaviours and impulses.
In the next section I describe some of the ways in which this physical development is thought to manifest psychologically in young people. I begin with a discussion of competence and then turn my attention to self-identity and sexual maturity.

3.2.2 Competence

Discussions of young people’s competence have become an unmistakable focus in discourse regarding predictive genetic testing. It is clear that young people’s minds are different from those of adults. It is also apparent that the transition from absolute incompetence to a level of competence equal to that found in adults is a gradual one, with cognitive capacity becoming more sophisticated with age. The uncertainty lies in exactly where along this continuum of cognitive sophistication young people are capable enough to perform the complex mental tasks usually reserved for adults. Of particular relevance to predictive genetic testing are capacities relating to decision-making, (as a decision must initially be made about whether to undertake testing) and capacities relating to coping (as the genetic information imparted must then be managed). In this section I explore these two facets of competence.

3.2.2.1 Decision-making

It has been proposed that good decision-makers require five skills. These are the skills to (1) identify alternative courses of action, (2) identify criteria for considering these alternatives, (3) assess alternatives, (4) summarise information about alternatives and (5) evaluate the outcome of the decision-making process. When then, are adolescents able to master these skills to a level necessary for making a decision about predictive genetic testing?

Jean Piaget is arguably the most influential cognitive developmental theorist to date. He was the first to devise empirical means to study the differences between children and adults and the first to present a systematic theoretical account of the process that constitutes development of the mind. Piaget believed that children are qualitatively different from adults, not simply miniature versions of them, or adults in training. His research set out to demonstrate this. Piaget proposed that there were four stages of cognitive development, of which the final stage was that of ‘formal operations’. He
believed that this stage occurred at the age of eleven years and that it involved the capacity to comprehend highly abstract notions and to predict future possibilities.\textsuperscript{36,95}

Since Piaget, several researchers have also devoted much energy to understanding the development of competence, specifically in relation to decision-making. Grisso and Vierling performed a systematic examination of existing studies in developmental psychology.\textsuperscript{100,101} They concluded that young people who have reached the age of 15 years are usually no less competent than adults.\textsuperscript{100,101} They also suggested that young people between the ages of 11 and 14 years may be able to provide informed consent for specific purposes.\textsuperscript{100,101} Weithorn and Campbell similarly demonstrated that 14 year olds show levels of competence to consent to medical treatment that are comparable to the competence demonstrated by adults.\textsuperscript{36,102} Nicholson also argues that by the age of 14 years the only difference between adolescent’s competencies and adult’s competencies relates to the experience and information they possess.\textsuperscript{36,103} Buchanan and Brock, in their discussion of minor’s decision-making capacity, yet again argue that developmental evidence supports the assertion that by the age of 14 or 15 years, young people have developed the capacities necessary for competence in health-care decision making.\textsuperscript{104} This stance has been echoed several times in recent literature.\textsuperscript{105-108} It has also been endorsed by the Society for Adolescent Medicine.\textsuperscript{109,110}

It has been suggested that the level of competence required for making specific decisions is in fact dynamic.\textsuperscript{36,104} That is, the faculties required for making a decision about one aspect of life may be different from those required for other, different aspects. For example, young people who have grown up in families with a history of a genetic condition may be capable of making a decision about predictive genetic testing, yet simultaneously unable to make a decision about who should rule Afghanistan.

\textbf{3.2.2.2 Coping}

Lazarus and Folkman define coping as “constantly changing cognitive, emotional and behavioural efforts to manage external or internal demands that are appraised as taxing or exceeding the resources of the person” (p 141).\textsuperscript{111}
Young people’s capacity for coping with traumatic events is of particular relevance to discussions about predictive genetic testing in young people. The ability to manage either a gene-positive or gene-negative test result is vital to considerations in the debate about young people’s access to testing. If young people do not have either the capacity for coping, or a necessary repertoire of coping skills, they may be at greater risk of harm when undergoing such testing. This would then place them in a position of greater vulnerability when compared to adults.

In the field of psychology, when a young person’s ability to cope with stressful life events is considered, a discussion of protective factors and risk factors is common. Bee, in her discussion of youth resilience, notes that:

“Children who are securely attached to someone (whether it be a parent or someone else), who have good cognitive skills, and who have sufficient social skills to make connections with peers are better able to weather the stresses they encounter” (p 420).

Bee then goes on to explain how more vulnerable children are far more likely to show a significant psychopathology in the face of stress. She states that it is helpful to conceive of children as having certain in-built vulnerabilities, as well as a range of protective factors, and to understand that these moderate their reactions to stressful life events. She also notes that the number of concurrent stresses that are occurring at the time moderates the impact of a stressful event. It may be that adolescence is a time in which young people are surrounded by many supports, given the proximity of family life at that time. However, it may also be that young people have specific vulnerabilities during adolescence, that dissipate with age.

Although the field of research relating specifically to adolescent coping is relatively small, some important findings have been reported. Some of these relate to age-related differences in coping. Williams and McGillicuddy-De Lisi report that older adolescents use a greater variety of coping strategies than younger adolescents. Frydenberg and Lewis report that younger adolescents are more likely to deal with stress by distracting themselves whereas older adolescents tend to use tension-reducing techniques. Most researchers have concluded that young children use more concrete,
single strategies to cope, while older children are more flexible, using a variety of coping strategies.\textsuperscript{114}

Some literature differentiates between two distinct types of coping. For example, a distinction is often made between ‘problem-focused’ coping and ‘emotion-focused’ coping.\textsuperscript{115} The former is associated with altering the source of stress, while the latter is associated with managing the emotional distress.\textsuperscript{111,115} Another distinction commonly used is that of ‘avoidant coping’ and ‘approach coping’.\textsuperscript{115} Avoidant coping is associated with eluding the stressful situation while approach coping is associated with engaging in direct efforts to alter the situation.\textsuperscript{116} Generally, it is accepted that approach strategies relate to more positive adjustment, while avoidant strategies lead to poorer adjustment.\textsuperscript{115,116}

It has been reported that young people’s reactions to stressful situations, and thus their ability to cope with these, are also dependent on the type of situation they encounter. For example, Griffith and colleagues report that, when faced with family stressors, adolescents are more likely to use approach coping. However, when confronted with peer or school stressors, they are more likely to utilise avoidant strategies.\textsuperscript{116}

Gender appears to be yet another moderating factor in young people’s coping strategies. The most consistent findings demonstrate that female adolescents tend to seek social support while male adolescents use ventilation or ignore the problem.\textsuperscript{112,113} It has also been noted that female adolescents are often more affected by stressful events than males.\textsuperscript{116}

Empirical research has demonstrated that when young people are informed of a serious issue at an early age, this may facilitate coping.\textsuperscript{36} For example, disclosure of adoption to young people at early ages has been associated with better ability to cope than disclosures occurring later in life.\textsuperscript{25} Other empirical research relating to young people and traumatic events has demonstrated that cancer alters the way that adolescent survivors view themselves, but that these alterations can have both positive and negative consequences.\textsuperscript{117} It has also been reported that children who know their HIV status have higher self-esteem than children who are unaware of their status.\textsuperscript{118}
Thus young people may be more vulnerable to predictive genetic testing if they have not yet established appropriate coping strategies, or a necessary repertoire of coping strategies. However, it is also possible that young people may cope better in response to a predictive genetic test than older individuals, as has been demonstrated through past research concerning HIV diagnoses in young people, or disclosure of adoption to young people.

Schmidt, Petersen and Bullinger conclude that currently there is “no consensus about the basic issue of whether coping in children is more diverse or less diverse than in adulthood” (p 65). 114

3.2.3 Self identity

Personal identity continues to develop and change throughout our lives. However, the development of a stable and unique self-identity is a task that is particularly related to adolescence. 28,36,108,119 Adolescence is a time when attachments with parents (or other significant carers) become less important and the emergence of an independent self becomes crucial. Importantly, once self-concepts (or self schemas) are established, they are known to be extremely difficult to alter. 28

Erik Erikson proposed eight specific stages of development that individuals must pass through as they mature. Each stage, he argued, is associated with mastering a particular task. 120 Erikson believed that the fifth stage in this developmental process, which occurs during adolescence, relates specifically to self-identity. He claimed that if a clear self-definition does not emerge during adolescence, ‘role confusion’ may occur later in life, where adolescents are unsure of who they are to both themselves and others. 120 Erikson believed that the achievement of this task centred around peers and role models. 120

Predictive genetic testing, if performed at a time when self-identity is being formed and strengthened, may influence the way in which this process occurs. For example, if young people receive a gene-positive test result during adolescence and then associate such a result with not being able to form an intimate relationship or being married, they may incorporate this conception into their self-concept. These impressions may in
turn affect a young person’s self esteem; a particularly vulnerable trait during adolescence. However, it is also possible that if young people are able to incorporate their gene status into their self-concept while this self-concept is still malleable, the process may be easier for them to achieve.

Self-identity could also potentially be harmed by a refusal to provide a predictive genetic test. Following a refusal, young people may develop a perception that they are not worthy of access to such services, or that they are not mature or intelligent enough to be provided with such information. A refusal may then also affect young people’s self-esteem. Thus in some cases, provision of a predictive genetic test may boost self-esteem.

In the following sections I describe two characteristics of being young that are associated with this development of self-identity: the establishment of independence and engagement in risk behaviours.

3.2.3.1 Establishing independence

An important task of adolescence is the relinquishing of emotional bonds shared with parents (or other caregivers) so that secure attachments can be formed with others. This transition from dependence to independence can be a particularly vulnerable time for young people.

Predictive genetic testing in adolescence may affect the separation process that needs to occur between young people and their parents. For example, if young people receive a gene-positive test result, they may seek to reinforce the attachments that were beginning to appropriately weaken with their parents. Additionally, parents may reinforce this process. Reinforcement is particularly likely with the parent who shares the gene-positive status. Alternatively, when young people seek predictive genetic testing, based on their own motivations, this may help them in establishing a separate and independent identity.

The establishment of independence in adolescence can also involve behaviours that are perceived by others as closed, antisocial, argumentative and rude. There is a significant body of literature aimed at clinicians that is devoted to the topic of working with
adolescents for this specific reason. Adolescents can be difficult. In relation to predictive genetic testing, this type of defensive behaviour may lead to difficulties in the counselling setting. Pre-test counselling is traditionally aimed at gaining a sense of the person who requests testing and preparing them for the experiences ahead. For young people, this stage of the predictive testing process holds particular importance. Clinicians meeting with young people will be attempting to gain some insight into the young person’s coping strategies, comprehension of genetic information, ability to perceive themselves in the future and network of support available. If young people present to clinicians in a closed, argumentative manner, albeit appropriate for their age, this clinical process could become particularly awkward and complicated. If adolescents remain unresponsive in the counselling environment, it may be tremendously difficult for clinicians to gain insight into their emotional state both before and after testing.

3.2.3.2 Risk behaviours
During the late 19th and early 20th centuries, Sigmund Freud proposed a complex theory in which he categorised the human mind into three facets: the id, the ego and the superego. Freud postulated that the id is present at birth and represents our most basic, biological needs. It seeks satisfaction immediately, and takes up the largest part of the mind. The ego was thought to represent the conscious, rational part of the mind, helping the id to find gratification in a socially acceptable manner and managing the id. The superego was then proposed as the seat of the mind in which conscience sits, representing society’s values and acting as the moral voice. For example, when a child plays with a friend and desires the toy their friend plays with, the id may exert a pressure to hit the friend and seize the toy. At the same time, the superego may exert a pressure to wait and ask for a turn with the toy, while the ego is left with the difficult task of balancing these competing pressures. Although Freud was not particularly concerned with adolescence (his focus was upon the earlier years of childhood) his daughter, Anna Freud, contributed to this field specifically. Anna Freud believed that the id “flared up” during adolescence, causing strong and passionate desires for pleasure. She believed that aggressive impulses are heightened at this time and that hunger becomes voracious, often giving rise to naughty behaviours. Although much of the Freuds’ theories have now been discarded or superseded, this particular conception of adolescence is a useful one for considerations about risk behaviours.
Adolescence is a time of experimenting and testing boundaries. In many cases, this experimentation is driven by a “voracious hunger” for love, acceptance, freedom, pleasure, inspiration and independence (p 65). Testing boundaries always entails some risk. However, some types of risk behaviours can be particularly worrying during adolescence because of the potential for harm, both in the short term and long term. It may even be that a desire to undergo predictive genetic testing forms part of young people’s engagement in risk behaviours.

Drug abuse is an example of a risk behaviour that young people engage in. It also has potential for harm. Drug abuse, as opposed to drug use, is categorised as the use of drugs to the point of causing risk or harm to oneself or others. Experimental drug use is common in adolescence. However, when this use is prolonged and intensified, it poses potential for specific harm not only in the immediate sense, but in relation to long-term behaviours and patterns. Studies frequently show that the most common drugs used during adolescence are alcohol, tobacco and marijuana. Drug abuse has the potential to alter a range of factors within young people’s lives. Examples include cognitive capacity, emotional state, attachment to others and self-esteem. These effects may then in turn create greater vulnerability to the potential harms and benefits of predictive genetic testing.

Unsafe sexual practices are another example of a risk behaviour that young people may engage in. Such behaviour may occur as a result of decreased cognitive capacity brought about by drug use, ignorance or ambivalence. The consequences of engaging in unsafe sex include pregnancy, contraction of a sexually transmitted infection and, at the extreme end of potential, death as a consequence of infection with HIV.

3.2.4 Sexual maturity

One of the more obvious aspects of adolescence is puberty and the associated sexual maturity that is obtained as a consequence of this process. Adolescence is the time when young people first become sexually mature and appropriately, begin to desire intimate relationships with others.
Future reproductive choices are often considered seriously for the first time during adolescence. For a young person who is at risk of a genetic condition, such considerations may hold particular significance. It may be that considerations about future reproduction provide a strong impetus for young people’s desire to know their genetic status. Thus knowledge about future reproductive options may constitute a practical benefit of predictive genetic testing.

Given that sexual maturity is reached during the period of adolescence, it is also possible that young people may present for testing because they are pregnant. Predictive genetic tests are currently available to pregnant women in order to help them make better informed decisions about their pregnancies. Predictive genetic testing would thus be capable of providing similar benefits to pregnant adolescents.

3.2.4 Summary of the psychological features of being young

Psychologically, young people differ from adults in several fundamental ways. During adolescence, young people’s brains have not yet reached a final stage of development. Cognitive capacities are lower during early adolescence, although these are thought to reach a level similar to that of adults by the age of approximately 14 years. Young people may cope in a variety of ways in response to a stressful situation. Their choice of coping mechanism may be influenced by their age, gender and the type of situation they encounter. During adolescence, the establishment of a stable self-identity is vital. This process involves separation from parents and the development of stronger attachments with peers. Development of self-identity may also entail the testing of boundaries and engagement in risk behaviours. Sexual maturity is also reached during adolescence, meaning that young people may begin to consider their reproductive options.

These psychological factors combine to make young people different from adults and therefore in need of special and separate attention in the debate about predictive genetic testing. The range of psychological differences between young people and adults may both increase young people’s vulnerability to the potential harms of testing.
and provide specific opportunities for benefit that may not be possible if testing is performed in adulthood.

In the next section I move from these psychological differences between young people and adults to the social differences that exist between these groups. Although in this chapter I separate the psychological features of young people from the social features of being young, this separation is artificial. In reality, the social and the psychological are interrelated. I have separated them only for the sake of organising my discussion in a clear way.

3.3 Social features of being young

In this section I explore the ways in which society influences young people and in some ways persuades them to live different types of lives from those of adults. Here I am concerned with the factors extrinsic to young people that make them different from adults. Once again, I pay particular attention to the sub-set of social qualities that may potentially cause young people to react differently from adults in response to a predictive genetic test. I begin with a discussion of the social interactions that are of importance to young people and then I move to the social tasks required of young people.

3.3.1 Social interactions

Albert Bandura has been particularly concerned with social learning theory in adolescence. That is, how adolescents learn from those around them. Bandura refers to a process known as ‘modelling’, where young people mimic behaviours surrounding them. He refers primarily to parents, siblings and the broader family but, during adolescence, he notes that peers become an important source to model from. In its most extreme conception, modelling behaviour can be understood in terms of behavioural theory, such as the theories put forward by Skinner and Watson. Behavioural conceptions of development are based on a simple, biological understanding such that when behaviour is rewarded through positive means it is reinforced, and when behaviour is punished it is then avoided in the future. For
example, if a young person who skips school is praised by peers (or rewarded), this behaviour will then be reinforced and most likely repeated.\textsuperscript{120}

Bandura went on to extend social learning theory by relating it to cognition. He argued that young people and adults are cognitively responsible for choosing the environment in which they inhabit and are therefore active in influencing who their role models and peers will be.\textsuperscript{120}

In this section relating to young people’s social interactions I am interested in the relationships that young people share with other people. I focus upon those relationships that hold particular importance for young people precisely because they are young. I begin with a description of relationships shared with parents and then focus on siblings and peers.

3.3.1.1 Parents

Most young people, although gradually moving from a state of dependence to one of independence, remain under the care of adults during adolescence. Most often these adults are the young person’s parents, although not always. In fact, young people living in families with a history of a genetic condition are potentially more likely to be cared for by people other than their parents as one parent is often sick. However, for simplicity, I refer to all carers as parents.

It has been suggested that the family is probably the single most important influence in an adolescent’s life (p 239).\textsuperscript{99} The fact that most young people live with their parents effectively amplifies the relationship that they share. This characteristic is unique to young people, with adults experiencing a less intense relationship with their parents, once they have left the parental home.\textsuperscript{99} Rice and Dolgin note that:

\begin{quote}
“Because many family members typically share the same physical space, they come into frequent contact with each other (whether they want to or not)… Interactions with family members are frequently intense and impassioned” (p 231).\textsuperscript{99}
\end{quote}

Interactions that occur between young people and their parents have the potential to influence not only a decision about whether to undergo predictive genetic testing but
also the way in which such a test impacts upon the young person involved. Parents are perhaps the most obvious people to provide support when their children undergo predictive genetic testing. It is also possible that this need for parental involvement and support may not be a conscious or explicit desire of young people, but it is nonetheless crucial. If parents are dealing with their own personal issues, such as guilt about their children being at risk of a genetic condition or difficulty coping with their own or their partner’s illness, this may impact upon their ability to provide support. As Rice and Dolgin state, “the behaviour of one family member often affects one or more of the others” (p 231). Alternatively, if parents have the resources to provide support during the process of predictive genetic testing, the proximity within which family relationships exist may serve to enhance the support that is provided.

3.3.1.2 Siblings

Just as the relationship between young people and their parents is amplified by the fact that they live together, so too are the relationships between siblings intensified during adolescence. Of added importance for sibling relationships are two factors. Firstly, full biological siblings generally share the same level of risk that is associated with the genetic condition in their family. Secondly, they share the increased volatility and vulnerability that is associated with being young. In many ways then, there is even more that can be amplified within sibling relationships. Rice and Dolgin note that:

“The relationships between brothers and sisters are vitally important because they may have a lasting influence on development and on the individual’s ultimate adult personality and roles” (p 246-7). Alternatively, siblings living in families with a history of a genetic condition may find themselves sharing a special bond based around their potential for inheriting the condition themselves. These may then be threatened if a discrepancy in test results occurs between siblings (if one sibling were to receive a gene-positive result while another received a gene-negative result). Alternatively, the shared bond of risk that is experienced between siblings may serve to enhance their ability to support each other through the predictive genetic testing process.
3.3.1.3 Peers

“Adolescence is a time of profound changes in relationships with peers. As teenagers break away from their families, they spend increasingly more time with their friends” (p 277).  

Peer relations become a focus during adolescence. This focus is associated with the process of moving from dependence to independence and thus forming new attachments with individuals other than one’s parents. Becoming part of a peer group and feeling accepted, liked and secure are important aspects of young people’s social interactions. Being neglected or rejected by peers in adolescence can lead to serious problems later in life (p 278).  

The desire to conform to peer values and desires is sometimes seen as an impediment to autonomy. In other words, if young people are primarily absorbed with desires for acceptance within their peer group, they are less likely to engage in behaviours or thoughts that are independent of this group mentality. Rice and Dolgin highlight this, stating that “one way the individual can become liked by a particular group is to be like other members of the group” (p 284).  

Stigmatisation is a possibility following predictive genetic testing. It is possible that insensitive peers may contribute to the potential for stigmatisation, driven by either a lack of knowledge or perhaps their own desire for acceptance within a peer group. However, strengthening of peer relationships may also be an outcome of predictive genetic testing. It has been reported that, following the diagnosis of cancer in children and adolescents, improved relationships are often a beneficial outcome of the experience.  

I turn now from the social interactions that affect young people to the social tasks that are relevant to them.
3.3.2 Social tasks

Uri Bronfrenbrenner has developed a specific model for understanding the ways in which society influences individuals, known as his Ecological Model. Bronfrenbrenner’s theories are widely accepted and used frequently.

At the heart of Bronfrenbrenner’s Ecological Model is the existence of four distinct systems that surround the individual: the microsystem, mesosystem, macrosystem and exosystem. The microsystem encompasses family, school, peer groups, doctors, religious groups and other entities that young people are in close, direct contact with. The microsystem exerts the most immediate influence on young people. The mesosystem then represents the interactions that take place between entities within the microsystem. For example, the interactions between school and home would form part of this mesosystem. The exosystem represents the environments in which young people do not participate actively, but which nonetheless impact upon them, for example, their parent’s workplace or the media. The macrosystem contains far-reaching attitudes and ideologies of the culture within which young people live. This incorporates legal, educational, economic, religious, political and social values. Bronfrenbrenner argues that adolescent social development is best understood when we consider the influences of several entities in relation to each other.

Taking Bronfrenbrenner’s Ecological Model and applying it to predictive genetic testing in young people allows us to perceive young people in a dynamic, multi-faceted sense. It also highlights the range of ways in which young people can be affected, inspired, persuaded, included, intimidated, pressured and accepted by society in a broad sense. Specifically, Bronfrenbrenner’s macrosystem highlights the ways in which young people may be pressured to perform particular social tasks as a consequence of the culture in which they live.

In this section relating to young people’s social tasks I am interested in the expectations that are placed upon young people in our culture. I begin with a specific discussion of career choice and then turn to the life transition young people are expected to perform.
3.3.2.1 Career choice

Vocational, educational and lifestyle choices become a focus in middle adolescence. The macro-system of our culture places particular importance upon the choice of a career during the final years of schooling. For many young people, this period entails great stress and angst about their choice of subjects, performance in school and likelihood of future success. As Williams and McGillicuddy-De Lisi note:

"Adolescence is recognised as a particularly stressful period of development. During this period adolescents deal with … educational demands and expectations and decisions about schooling and careers" (p 537).

Even under the best of circumstances, choosing a career is an increasingly difficult task (p 380). Young people who have no desire to learn of their genetic status may find that repressed anxieties resurface at this time, as the future becomes a focus. These young people may feel forced into considerations about their genetic future before they feel capable of this, conferring potential for emotional harms and even displacement from the school environment. However, young people who have a desire to learn of their genetic-status may find that vocational decisions become increasingly difficult without this knowledge. For these young people, predictive genetic testing may facilitate their decision making process.

3.3.2.2 Life transition

Adolescence has been described as a bridge between childhood and adulthood that must be crossed in order to assume position as an adult member of society. In our society, adolescents are not generally expected to perform adult roles. Adolescence is perceived as a period in which young people learn to become adults and practice the associated skills, not where they begin to behave as adults. In many other cultures around the world, for example non-western countries, adolescents are given responsibility earlier, not only for themselves but also for others, and thus adulthood is practiced much earlier.

The social transition from child to adult involves developing several distinct capacities. Adolescents in our society must master the expertise they will require in order to live successfully as adults. Examples of skills required include cooking,
cleaning, managing finances, caring for others, time management and forward planning. The achievement of this transition can place intense stress upon young people, causing potential for greater vulnerability.  

Sometimes extreme behaviour during adolescence can reflect a desire not to perform this transition. For the young person who receives a gene-positive test result, the move from child to adult may seem unobtainable or inappropriate. A consequence of such a test result may be the perception that a normal adulthood is not possible, forming a barrier to the transition from child to adult and the behaviours and tasks that are required as part of this process. Alternatively, for young people who are motivated about this transition from childhood to adulthood and who wish to learn of their genetic status, an inability to undergo predictive genetic testing may be viewed as an impediment to the transition.

3.3.3 Summary of social features of being young

Socially, young people differ from adults in several fundamental ways. These differences contribute more endorsement for the separation of young people from adults in debates about predictive genetic testing. Young people most often live with their parents and siblings and this creates an amplification of these relationships. Peer relationships are also of particular importance during adolescence, providing an avenue for secure, non-familial attachments and a source of behaviour to model.

In addition to these social interactions, young people are faced with specific social tasks during adolescence. These are brought about by the culture in which young people live. Two specific components of society’s expectations are the need to choose a career path and the need to acquire the skills necessary for completing the transition from childhood to adulthood successfully. These tasks can place enormous pressure on young people. The range of social differences between young people and adults means that it is likely predictive genetic tests will impact differently upon young people from the way in which they impact upon adults. However, it remains unclear if these differences will result in a greater potential for harm or greater potential for benefit when young people undergo such testing.
Once again, I note here that my separation of the social features of being young from the psychological features of young people is an artificial one. In reality, both the psychological and the social intertwine.

As stated above, in this chapter I aimed to touch on some broad, relevant issues rather than to provide a comprehensive review of the psychological and social features of being young. Some of the ideas presented in this chapter are based upon good scientific evidence, while others are more speculative. Those based upon empirical evidence include the physical development of the brain, the discussion of the development of competence in young people and the discussion of sexual maturity. The ideas that are more speculative include the discussion about risk behaviours in young people, the ideas presented about coping strategies and the discussion of the task of establishing independence.

### 3.4 Predictive genetic testing in young people specifically

In this chapter I have been concerned with the question of why young people need to be separated from adults in discussions about predictive genetic testing. I have outlined key characteristics associated with being young, both psychologically and socially. To conclude this chapter, I now return to the arguments used to oppose predictive genetic testing in young people in order to articulate which of these are in fact most powerful, given the preceding discussion.

If arguments are being made for withholding predictive genetic tests from young people but providing such tests to adults, they must be specific to young people. In other words, any argument that opposes predictive genetic testing in young people must be based on a consideration of the differences between young people and adults, thus providing a justification for withholding such testing from young people but not adults. Arguments made to oppose testing in young people must not apply, or must apply only in an attenuated form, to adults.
Some of the arguments used to oppose predictive genetic testing in young people do indeed focus on the types of issues raised within this chapter. These arguments oppose predictive genetic testing in young people because of the ways in which young people differ from adults. These are the most powerful arguments. However, many of the arguments used to oppose predictive genetic testing in young people are also relevant to adults. These arguments are therefore less powerful in the current debate concerning predictive genetic testing in young people.

In this section I consider the range of arguments used to oppose predictive genetic testing in young people and the significance they hold to young people specifically.

**3.4.1 Arguments used to oppose predictive genetic testing in young people**

In chapter one I reviewed the arguments used to oppose predictive genetic testing in young people. I gathered these arguments not only from current guidelines concerning such testing but also from the broader base of literature concerning the topic of predictive genetic testing. I now list the arguments used to oppose testing in mature young people. I choose only to provide the arguments related to mature young people, as opposed to both mature and immature young people, as mature young people represent my primary field of interest in this thesis. The arguments are as follows:

1. Testing may interfere with the natural separation process that needs to occur between parents and their children.
2. A gene-positive child may be forced into the realisation that their parent is not ‘all-powerful’ and could not save them from the genetic condition. This could be psychologically damaging if it occurs too early in the young person’s life.
3. The process that occurs during adolescence of establishing a personal identity may be made particularly difficult by a gene-positive test result.
4. A child who receives a gene-positive test result may become a scapegoat who is abused because he or she reminds the parent of the parent’s own unacceptable traits.
5. Harm to the parent-child relationship may occur, especially if parents become over-protective or experience anxiety.
6. Young people may feel guilty if they receive a gene-negative result, and their parent or other siblings are gene-positive.
7. The child may be treated as a victim in the family.
8. The child may develop an altered self-concept, being identified as a ‘carrier of a gene mutation’
9. A child who receives a gene-positive result may feel unworthy and unmarriageable
10. Testing may cause a loss of self-esteem
11. Parents may alter their expectations of the child, affecting the child’s psychological state
12. The child may become stigmatised or discriminated against
13. Testing may remove the possibility of a child obtaining certain types of insurance later in life
14. The child may develop increased anxiety
15. The ‘unpatient’ syndrome may develop in a child who is tested, where the child begins ‘sick’ behaviours such as visiting doctors frequently, even though he or she is not yet sick
16. Testing may affect the bond that siblings shared when they were all at risk
17. Parents may feel guilty about their child’s gene-positive test result
18. Testing may place added financial burden on the family

If these arguments are now considered again, in light of the ways in which young people differ from adults, it becomes evident that they hold varying levels of significance to young people specifically. Some of the arguments listed above can only be applied to young people. These arguments are the most powerful for opposing predictive genetic testing in young people. Other arguments in the list above could be applied to both adults and young people, yet are magnified when related to young people. These are less powerful.

The arguments that are most powerful for opposing predictive genetic testing in young people, because they relate only to young people specifically, include:

- Testing may interfere with the natural separation process that needs to occur between parents and their children.
- A gene-positive child may be forced into the realisation that their parent is not ‘all-powerful’ and could not save them from the genetic condition. This could be psychologically damaging if it happens too early in the young person’s life.
- The process that occurs during adolescence of establishing a personal identity may be made particularly difficult by a gene-positive test result.
• Harm to the parent-child relationship may occur, especially if parents become overprotective or experience anxiety.
• A child who receives a gene-positive test result may become a scapegoat who is abused because he or she reminds the parent of the parent’s own unacceptable traits.
• Parents may alter their expectations of the child, affecting the child’s psychological state
• Testing may place added financial burden on the family
• Testing may remove the possibility of a child obtaining certain types of insurance later in life, as life insurance cannot be obtained before young people become legal adults.

These arguments are most powerful because they are specific to young people. For example, the natural separation process that needs to occur between parents and their children is not relevant to adults undergoing predictive genetic testing. Similarly, the process of establishing a personal identity that is associated with adolescence is not a consideration for adults undergoing testing. These arguments relate only to young people and therefore hold the most weight in current debates about the provision of such testing to young people.

The arguments that are less powerful for opposing testing in young people, because they can be applied to both adults and young people but are magnified in young people specifically, include:

• The child may develop an altered self-concept, being identified as a ‘carrier of a gene mutation’
• Young people may feel guilty if they receive a gene-negative result, and their parent or other siblings are gene-positive
• The child may be treated as a victim in the family
• A child who receives a gene-positive result may feel unworthy and unmarriageable
• Testing may cause a loss of self-esteem
• The child may become stigmatised or discriminated against
• The child may develop increased anxiety
• The ‘unpatient’ syndrome may develop in a child who is tested, where the child begins ‘sick’ behaviours such as visiting doctors frequently, even though he or she is not yet sick
• Testing may affect the bond that siblings shared when they were all at risk
• Parents may feel guilty about their child’s gene-positive test result
This second group of arguments could also be applied to adults, but hold particular weight when applied to young people. For example, testing could cause a loss of self-esteem for both adults and young people who undergo testing. However, a loss of self-esteem may be particularly difficult for young people as they are in the process of developing a unique personal identity. Similarly, both young people and adults may feel guilt about a gene-negative test result if siblings are gene-positive. However, this may be particularly difficult for young people as they are likely to be living with their siblings.

Thus it is clear that not all of the arguments used to oppose predictive genetic testing in young people hold the same weight. Most powerful are arguments that are highly specific to the differences that occur between young people and adults (or young people’s lives and adult’s lives). Less powerful are those that can be applied to both young people and adults. Consideration of the types of arguments used in debates about predictive genetic testing in young people is therefore critical.

3.5 Conclusions

In this chapter I have demonstrated the ways in which young people differ from adults and thus the justification for separating them from adults in discussions about predictive genetic testing. I began with an exploration of the psychological differences between young people and adults. Here I paid particular attention to the cognitive capacities for decision-making and the coping strategies that adolescents often utilise, including the factors that moderate these. I then focussed on the development of young people’s self-identity, their struggle for independence and the likelihood that they will engage in risk behaviours. Next, I changed my focus from psychological differences between young people and adults to social differences between these groups. I contemplated the social interactions that are of particular importance in young people’s lives, including their relationships with parents, siblings and peers. I then concentrated on social tasks, examining decisions young people have to make about a future career and the life transition from childhood to adulthood that is expected of them during adolescence. Finally, I returned to the topic of predictive
genetic testing in young people to produce a list of the most powerful arguments used to oppose predictive genetic testing in young people. These arguments hold the most credence because they focus upon the ways in which young people differ from adults. It is clear that the many differences that exist between young people and adults imply a likelihood that predictive genetic testing will impact differently upon young people from the way in which it does upon adults. However, it remains unclear if these differences will confer a greater potential for harm or a greater potential for benefit if young people undergo such testing.
CHAPTER FOUR: RESEARCH METHODOLOGY
4.1 Introduction

Young people differ from adults psychologically and socially. It is likely that young people will therefore react differently from the way in which adults react in response to a predictive genetic test. However, it is not clear if these different reactions will create greater potential for harm or greater potential for benefit if young people undergo such testing. Perhaps it will create neither. Of certainty are two facts. Firstly, professionals have been unable to reach consensus about the likely outcomes of predictive genetic testing in young people. Secondly, the available empirical evidence is not substantial enough to corroborate any opinion. Thus we find ourselves in a situation of professional conflict with no means of substantiating the theoretical claims that are being purported.

In this chapter I will argue that the best hope for resolution in the debate occurring about predictive genetic testing in young people is empirical research. In fact, I will argue that this type of theoretical clash requires empirical research. I begin by articulating two fundamental gaps that exist in our knowledge about predictive genetic testing in young people. I then outline the two phases of empirical research that I conducted in order to initiate the task of filling these gaps in knowledge. I describe how I planned, conducted and analysed this research. Embedded within my description is also a defence of the merit in combining quantitative and qualitative research methodologies, so that I may clarify my reasons for doing this.

4.2 Fundamental gaps in our knowledge

In this section I describe two fundamental gaps that exist in our current knowledge of the issues concerning predictive genetic testing in young people. I begin by outlining our lack of knowledge about current practice. I then articulate the deficits in our knowledge about the subjective effects of predictive genetic testing in young people. Finally, I argue that these gaps require empirical research and that this empirical research should involve young people specifically.
4.2.1 Current practice

The first fundamental gap in our knowledge concerning predictive genetic testing in young people relates to current practice. What is happening at the moment? Are clinicians following existing guidelines? Are they aware of the existing guidelines? Do they agree with them? Is it possible that while an academic debate continues to build, predictive genetic tests are being routinely provided to young people already? There is a need to know where we are before we can argue about where we should be going.

There have been two key surveys published that sought to answer questions about the occurrence of predictive genetic testing in young people. The first survey reported results about the incidence of genetic testing in children in Britain.\textsuperscript{12} This survey found that widespread genetic testing of children was occurring, but that the majority of tests reported were performed because the genetic condition usually manifests in childhood, or because the test was necessary for good medical practice. Not a single case was reported of predictive genetic testing in a young person for strictly non-medical reasons (for example, a test for HD).

The second survey to be published reported the incidence of genetic testing in asymptomatic young people, as disclosed by laboratories in the USA.\textsuperscript{52} This survey demonstrated that many laboratories offering testing for ‘later-onset’ conditions were performing such tests on asymptomatic young people. That is, they were providing tests for non-medical reasons. Tests had been performed for 13 different conditions, including HD, Charcot-Marie-Tooth disease and Myotonic Dystrophy.

Although these two published surveys provide important information about the occurrence of predictive genetic testing in young people, they do not provide any information concerning the psychosocial impacts of testing in young people. They also do not provide any information about the clinicians involved in the provision of these tests. Were clinicians simply unaware of the existing guidelines or did they believe them to be wrong? Were the young people who were tested involved in the decision to be tested or were they too young to understand what was happening? Have results of the tests been disclosed to the young people involved? I reiterate the need to
know what is happening now, before continuing discourse about what should happen in the future.

4.2.2 First-hand accounts from young people

The second fundamental gap in our knowledge concerning predictive genetic testing in young people relates to the opinions of young people themselves. Of the five empirical studies that have been conducted in order to answer questions about the impact of such testing, not one has reported the opinions of young people. What do young people who have undergone predictive genetic testing think about the experience? What were the best and worst parts of the process for them? Do they feel harmed by the knowledge of their genetic status or are they glad to know? Do they simply feel ambivalent? We don’t know. The vast majority of potential harms that are purported to be associated with predictive genetic testing in young people are psychosocial harms, not physical harms. These are therefore at least partly subjective. In order to assess these outcomes accurately, we must consult young people who have experienced predictive genetic testing themselves. They are the only ones capable of providing us with real insights into the effects that such testing has upon young people.

Psychological instruments are vital for making standardised assessments of changes that occur in individuals’ mental states, but they are only able to find what they are designed to look for. In other words, a psychological instrument designed to measure levels of depression is only capable of finding increases, decreases or no change in depression. It is not capable of finding increases in happiness or decreases in anxiety or descriptions of the most depressing or exultant moments. Take the following example.

If my partner asks me if I love him, and I say yes, he can conclude that he is loved by me (provided he also trusts me). However, my answer about the love I feel for him tells him nothing about the love I also feel for my next-door neighbour, nothing of the fact that I plan to leave him and nothing about the confusion, sadness and anxiety I also feel, in addition to love. My answer is specific to his question, but it is not sufficient for what he is really interested in.
If we are interested in the broad implications of predictive genetic testing in young people, we must measure more than depression, anxiety and behavioural problems. This is not to say that measurements of depression, anxiety and behavioural problems are not worth documenting. These measurements provide essential contributions in the quest for evidence and, given the scarcity of evidence at this time, they inhabit a particularly valuable space. However, research at this early stage must remain explorative. We must resist making assumptions about the likely impacts of predictive genetic testing in young people and searching to document only these. Investigatory research is required. There is a need to ask young people themselves what predictive genetic testing is like for them, how they experience and perceive its effects upon them and what meaning they ascribe to it.

In the next section I clarify why these two gaps in our knowledge of the issues concerning predictive genetic testing in young people cannot be filled by an appeal to theory alone. I argue that empirical research is required.

4.2.3 Why this ethical question begs empirical research

The question about the permissibility of predictive genetic testing in young people for non-medical reasons is an ethical question. It is concerned with the autonomy of young people and asks us to weigh the harms and benefits of performing such tests.\textsuperscript{12,128} It is thus a question about three primary ethical principles: autonomy, beneficence and non-maleficence and which of these principles should override the others, if any.\textsuperscript{129} It is also a question about paternalism. That is, the right of clinicians to override the wishes of the people they treat, based on a desire to protect these people and prevent them from harming themselves. Here I argue that, although many ethical questions can be resolved through appeals to theory, this ethical question is not one of them. The fundamental gaps that exist in our knowledge about predictive genetic testing in young people require empirical research.

Some ethical questions can be examined purely via appeals to theory. Take the ethical debate currently raging about the rights of lesbians and single women to access IVF in order to become pregnant. Some people believe that only individuals who are medically infertile should have access to IVF. Others believe that those individuals
who are socially infertile (due to their sexuality or relationship status) should also have access. This ethical argument is about meaning. It is about how we understand infertility and what the primary purpose of IVF should be. This debate does not require empirical research, it requires us to re-think our previous assumptions.

This is not to say that the debate about the rights of lesbians and single women to access IVF may not be helped by empirical research. It may be highly beneficial to know of the reasons why some people oppose the use of IVF by lesbians and single women, or to learn of how many lesbians and single women actually request IVF. However, the debate does not require empirical research in order to advance. It is not stuck at an impasse concerning facts, such as what will happen if access is granted.

The ethical debate concerning predictive genetic testing in young people for non-medical reasons is indeed an argument about what *should* happen. But the argument continues because experts are unable to agree upon what *will* happen if young people undergo testing. It is an argument about what a given cause will affect. The only way to answer such a question is to measure the effects that are caused when testing is performed. Empirical research is required.

4.2.4 Involving young people in research

There has been a recent, international trend towards the involvement of young people in research. That is, a move away from treating young people as ‘objects of research’ and towards treating them as ‘informants’. Docherty and Sandelowski acknowledge this change in stating that “the focus has shifted from seeking information about children to seeking information directly from them” (p 177). This move has been attributed, in part, to the ratification of the UN Convention on the Rights of the Child. Article 12 stipulates that “the child who is capable of forming his or her own views has the right to express those views freely in all matters affecting the child”.

When young people are not involved in research that concerns them, it is often adults who are consulted to speak on their behalf. This is problematic, as Douglas notes here:
“Adults cannot assume that because they were once children that they know what life is like for a child. It is only through engaging with children that we can know and learn about their perceptions of life, events and people, coping mechanisms, understandings, concepts of relevance and importance” (p 5). 

Hart similarly argues that we cannot assume parents will be able to accurately reflect the feelings and needs of their children. Hart refers to a study about quality of life in children with asthma, reporting that measurements of quality of life by a proxy may in fact have reflected the proxy’s quality of life more than that of their child. Hart also refers to a study by Jessop and colleagues that reports a relation between the mental health of mothers and their ratings about the degree of disability in their children.

Waters and colleagues provide further evidence for distortions in reports that parents provide about their children. They asked parents and their adolescent children to report on aspects of emotional, physical, mental and social health and well-being. They found that the adolescents were much less optimistic than their parents. They warn clinicians that if “adolescents are unable to report for themselves, it is clear that relying on parent report will only partly inform the clinician of the severity of health experienced” (p 507-8).

Stalker and colleagues again detail the difficulties involved with using proxy accounts, stating that “it cannot be assumed that interviewing parents or professionals enables us to understand how life feels from a child’s perspective” (p 378). Bricher similarly reminds us of the importance of parents’ opinions, while still highlighting the need to distinguish these from those of young people:

“The failing to focus on the children’s perspectives in healthcare literature denies them the opportunity to share their thoughts and experiences. This is not to deny the importance of parent’s voices, but to recognise that the story they tell may be different.” (p 66).

Bearison also talks of the benefits associated with listening to young people’s experiences:
“The children were speaking in a voice that often was remarkably free of the social conventions, distortions, and defenses that adults use to hide similar questions and anxieties” (p 98).

And of the great deal there is to learn from providing young people with this space to talk:

“Simply listen to the children speaking in their own voices about issues and events that are important to them. There is a great deal to be learned and appropriated from their narratives. They teach us the value of listening to children on their own terms without judging them so that their internal voices will become louder in our time” (p 107).

There is thus not only a trend, but also a need to consult young people themselves about issues that concern them directly. The theoretical clash occurring in relation to predictive genetic testing in young people is one that requires the investigation of young people’s opinions. Not just because this is a topic that affects them and therefore the UN Convention on Rights of the Child suggests we should consult them, but because the stories young people are able to convey about this topic can shed new light on the current clash. Young people are in the unique position of being able to subjectively describe the effects of predictive genetic testing in young people. More importantly, they are the only ones who can.

My purpose in this section has been to argue that (at least) two fundamental gaps exist in our knowledge about predictive genetic testing in young people. I have outlined the need for detailed information about where we are now in relation to such testing and for the broad documentation of young people’s opinions about the impact of the predictive genetic tests they have experienced. I have also argued that in the ethical debate concerning predictive genetic testing in young people, advancement requires empirical research. This is due to the fact that the core point of dissent surrounds the topic of what will happen if young people undergo predictive genetic testing, as opposed to what should happen in the event that we learn such testing does (or does not) harm young people. Importantly, in this ethical debate, evidence can be produced
concerning what will happen. Young people must be involved as active participants in the documentation of this evidence.

4.3 Two phases of empirical research

Here I describe the two phases of empirical research that I conducted in order to initiate attempts to fill the gaps that exist in our knowledge about predictive genetic testing in young people. I begin with a discussion of reflexive research, which leads to a presentation of the epistemology, theoretical framework and methodologies I utilised in my research. I then go on to describe in detail the specific methods I employed. Embedded within this discussion is a defence of combining qualitative and quantitative research methodologies.

4.3.1 Reflexive research

Rice and Ezzy in their comments about researcher reflexivity in relation to qualitative research, articulate that:

“Reflexive research acknowledges that the researcher is part and parcel of the setting, context and culture they are trying to understand and analyse. That is to say, the researcher is the instrument of the research. … Qualitative research reports that avoid the pronoun ‘I’ are, in some sense, attempting to disguise the role of the researcher in the research. Rigorous qualitative research is honest about the role of the researcher in the project” (p 41).

Thus, as the instrument of the research that I describe in part two of this thesis I now provide a detailed account of the epistemology, theoretical framework and methodologies that guided my research. In initiating this task, I present some details about myself that may have impacted upon the way in which I prepared, conducted and analysed the research that I describe. These characteristics may also have influenced the way in which the participants of my research reacted to, responded to and engaged with me.
At the time I performed the research reported within this thesis I was 25-26 years of age, so I was close in age to the participants I interviewed. I am female. My previous education has involved completion of a Bachelor of Science, majoring in both psychology and genetics. I have also completed an Honours year in Bioethics. I am not aware of any genetic condition that runs in my own family and I have never undergone a predictive genetic test, so I do not share any of the experiences of a familial condition that my participants have.

4.3.1.1 Epistemology

An epistemology is “a way of understanding and explaining how we know what we know” (p 3). Take the example of a flower. Picture a time before the existence of humans and then picture situated within this world, the existence of a yellow daffodil. The epistemology of objectivism would hold that this yellow daffodil held innate meaning, even in the absence of human perception. Objectivists believe that the human perceiver discovers meaning that already exists. Alternatively, the epistemology of subjectivism holds that the yellow daffodil had no meaning in and of itself until a human noticed it and created a meaning for it. Subjectivists believe there is no ‘real’ world, only our subjective experiences. The meaning the daffodil holds in this case is dependent on the existence of a sentient being.

In describing the epistemology that governs me, as the instrument of my research, I reject both objectivism and subjectivism, drawing instead upon the epistemology of constructionism. Crotty describes the epistemology of constructionism as one in which:

“truth, or meaning, comes into existence in and out of our engagement with the realities in our world. There is no meaning without a mind. Meaning is not discovered, but constructed” (p 9).

Crotty goes on to explain that because of the way in which meaning is constructed, different people may construct different meanings, even in relation to the same event. Take the example of the flower once again. Constructionism holds that the flower does indeed exist as part of the ‘real’ world, irrespective of the presence of a human being. However, the meaning constructed in relation to the flower is dependent on the
specific human present. Thus a florist perceiving the yellow daffodil may construct a
different meaning from a person who suffers from hayfever who may, yet again,
construct a different meaning from a farmer wanting to clear the area. Thus
constructionism rejects both objectivism and subjectivism. It claims that there cannot
be only objective meaning, as meaning requires a mind, but that there also cannot be
only subjective meaning, as subjects must perceive a real world.

It is the epistemology of constructionism that guided me throughout my research. I
recognise that a real world exists, a world where predictive genetic tests can be
performed in order to provide information about an individual’s future health (or
illness). However, I deem that the meaning attached to such a test cannot be described
objectively. I am interested in the individual’s interpretation of the test, the meaning
they construct around it.

4.3.1.2 Theoretical framework
A theoretical framework consists of the assumptions that are brought to research, that
is, the “context for the process”. ( p 3)\textsuperscript{145} Rice and Ezzy, when speaking about the role
of a theoretical framework in relation to qualitative research, note that:

\begin{quote}
“Different theoretical frameworks direct attention to different aspects of a
phenomenon. For example… psychoanalytic theory points to the role of the
irrational and unconscious in shaping behaviour” (p 11).\textsuperscript{144}
\end{quote}

In describing the theoretical framework that grounded my own research, I must
differentiate between substantive theories and more general theoretical frameworks of
inquiry. The substantive theories that guided my research are those that I presented in
chapters one, two and three. These theories specifically relate to my topic, that is,
predictive genetic testing in young people. They include theories about harm and
benefit, theories about the impact of predictive genetic testing in young people and
theories about the differences between young people and adults. However, my
theoretical framework of inquiry is what I refer to within this section. This relates to
the theory that guided my research process. This framework is phenomenology.
Phenomenology can be understood in both simple and extremely complex terms. Becker provides a simple explanation:

“Phenomenologists study situations in the everyday world from the viewpoint of the experiencing person” (p 7).

Crotty provides a slightly more complex description:

“Phenomenology suggests that, if we lay aside, as best we can, the prevailing understandings of … phenomena and revisit our immediate experience of them, possibilities for new meaning emerge for us or we witness at least an authentication and enhancement of former meaning” (p 78).

Crotty describes how phenomenologists seek to return to “things themselves”, where things themselves are “phenomenon that present themselves to us immediately as conscious human beings” (p 78). Phenomenology asks us to “bracket” our existing conceptions and “let the experience of phenomena speak to us at first hand”. (p 79)

Crotty explains that phenomenology is concerned with our experience of phenomena “before we start thinking about, interpreting them or attributing any meaning to them.” (p 79)

Rice and Ezzy describe phenomenology in terms of individual “life worlds”. They state:

“phenomenology emphasises the individual’s construction of a ‘life-world’. Taken together, the whole of people’s unquestioned, subjective experience of their biological worlds can be termed their ‘life-world’” (p 15).

Rice and Ezzy then go on to describe how “phenomenologically oriented researchers study everyday events from within the life-world of the person experiencing them” (p 16).

Moustakas similarly notes that:

“The aim is to determine what an experience means for the persons who have had the experience and are able to provide a comprehensive description of it” (p 13).

Thus phenomenology guided my own research concerning predictive genetic testing in young people. Specifically, it guided my second phase of empirical research, in-depth
interviews with young people who have undergone predictive genetic tests themselves. I was, and am, interesting in learning of the meaning that young people ascribe to this experience and how the experience sits within their life-world.

4.3.1.3 Methodology
Methodology refers to the “strategy or plan of action” used to carry out research (p 3). Although I utilise both quantitative and qualitative methodologies within my research, I focus my attention here on a description of qualitative methodology primarily. I do so because empirical research in the field of predictive genetic testing has constituted primarily quantitative methodologies, such as standardised psychological assessment tools. The use of these quantitative methodologies has been particularly prevalent in the field of predictive genetic testing in young people.

“Qualitative research methods focus on meanings and interpretations” (p ix). They are distinct from quantitative research methods, which focus heavily on statistics and discrete measurements. Research in the field of health has traditionally relied heavily on quantitative data. However, qualitative research methods are now becoming more common (p 4).

As qualitative research becomes more common, its benefits also become more widely articulated. Baum states that qualitative data are more “powerful in allowing an understanding of the context issues that have become the concern of public health in recent years” (p 463). Rice and Ezzy note that qualitative research can be particularly useful in the exploratory phase of research (p 4). Holman states that “when qualitative methods are clearly established in our research repertoire, the advance of medical knowledge will be greatly accelerated” (p 5).

Take the example of research into HIV transmission, used by Rice and Ezzy. They explain that many of the health problems we are faced with are in fact “problems of meaning and interpretation” (p ix). In the case of HIV, we have a good biomedical understanding of how the virus is transmitted, yet this has not allowed us to prevent transmission. Rice and Ezzy posit that HIV remains a problem because of social factors, the ways in which people understand the implications of their actions and because of confused governments. Thus qualitative research, being concerned with
meaning and interpretation, has real value in helping to advance towards resolution a
problem that bio-medicine alone has failed to solve.

Although the field of predictive genetic testing has been largely dominated by
quantitative research methods, particularly in the field of testing in young people,
examples of qualitative research methods are becoming more common in the field of
genetics. Chapman and Smith, discussing the role of qualitative research in health
psychology, pay particular attention to the emergence of qualitative research
concerning the new genetics. They note that a number of studies have been published
using interpretive phenomenological analysis to explore issues in the new genetics.
These issues include genetic counselling, prenatal screening, decision making around
predictive genetic tests, reproductive choices following genetic testing and quality of
life for people living with a genetic condition. ¹⁵¹ Sandelowski and Corson Jones
recently described 40 interviews they conducted with women about experiences of
prenatal diagnosis. ¹⁵² Cox reported on 16 in-depth interviews with people who were
contemplating predictive genetic testing for HD, in order to articulate how their
decisions were made. ¹⁵³ Binedell, Soldan and Harper used qualitative methods to
detail the accounts of people who are living at risk of HD and were contemplating
testing.¹⁵⁴ Smith, Michie, Stephenson and Quarrell analysed transcripts of 5 in-depth
interviews with women who were offered testing for HD, in order to analyse
descriptions of risk status and the process involved in making the decision.¹⁵⁵

A qualitative-quantitative divide is often assumed. That is, researchers often perceive
that these two categories of methodology are relevant to different fields and should
always be kept separate. Crotty refers to this divide, noting that “in most qualitative
research textbooks, it is qualitative research and quantitative research that are set
against each other as polar opposites” (p 15).¹⁴⁵ However, he then goes on to explain
that “whatever research we engage in, it is possible for either qualitative methods or
quantitative methods, or both, to serve our purposes (p 15).¹⁴⁵

In choosing my own research methods, I chose to use the methodologies that best
suited the research questions I was asking. This resulted in the use of both quantitative
and qualitative methodologies, as outlined below.
The first gap in our knowledge about predictive genetic testing in young people relates to current practice. Within this first gap, there are three distinct research questions:

(a) Is predictive genetic testing in young people occurring for non-medical reasons in the countries where guidelines exist?
(b) What are the psychosocial outcomes for those who are tested?
(c) What are clinical geneticists’ reasons for providing or refusing these tests?

Question (a) was suited to quantitative analysis, as this was a question about occurrence. Research questions (b) and (c) were better suited to qualitative research, as these asked questions about psychosocial impacts of testing and clinicians’ reasons for provision or refusal of such tests. A quantitative methodology in this case would have been limiting. A qualitative methodology, allowing respondents to answer in their own terms, provided a better means for obtaining a range of possible responses and descriptions. A survey was therefore my choice of method for addressing this first gap in knowledge, concerning current practice. The survey allowed both forced-choice, quantitative components and open-ended, qualitative components.

The second gap in our knowledge about predictive genetic testing in young people relates to the opinions of young people who have undergone testing themselves. Within this second gap, there are two distinct research questions:

(d) What types of non-medical benefits and harms do young people who have undergone such testing refer to?
(e) What meaning do young people ascribe to the predictive genetic tests they have experienced?

In answering these two research questions a qualitative methodology, aimed at meaning and interpretation was appropriate. I therefore chose to conduct in-depth interviews with young people who had experienced predictive genetic testing themselves. I analysed these interviews using thematic analysis. However, this thematic analysis was divided into two stages in order to answer research questions (d) and (e) separately.
4.3.1.4 Thematic Analysis

Rice and Ezzy describe thematic analysis as a tool that allows researchers to identify and develop “concepts, categories and themes” as they perform their research (p 194). They go on to explain that such analysis should not begin with theories or laws that are applied to the data, but rather the theories should be discovered from within the data itself (p 194). That is, thematic analysis aims at discovering themes which emerge from within the data.

Given that concepts and theories in thematic analysis are meant to derive from the data itself, it has been suggested that it would be inappropriate to search the data with any pre-existing categories. Strauss and Corbin note that:

“It makes no sense to start with received theories or variables (categories) because these are likely to inhibit or impede the development of new theoretical formulations, unless of course your purpose is to open these up and to find new meanings in them (p 50).

However, within this assertion, Stauss and Corbin are explicit about the fact that, if the aim of using categories is to open them up and discover new meanings, than such categories may be useful (p 50).

As I described above, when analysing the in-depth interviews I performed with young people I used two stages of thematic analysis so that I was able to address research questions (d) and (e) separately. In the first stage of my thematic analysis, aimed at answering research question (d) I utilised two units of analysis, or categories. A unit of analysis has been described as a “tool to use in scrutinising your data” (p 71). I utilised the units of harm and benefit in this stage. I therefore interrogated the data for descriptions that fitted into these two units.

My second stage of thematic analysis, aimed at answering research question (e) involved a more holistic method. Here I refrained from the use of specific categories, allowing themes to emerge from the raw data and young people’s experiences to unfold.
In the next section I describe the specific methods I used to execute these quantitative and qualitative methodologies.

4.3.2 Phase one: An international survey of clinical geneticists

In this section I describe how I planned, executed and analysed a survey of clinical geneticists in the UK, USA, Canada, Australia and New Zealand.

4.3.2.1 Aims

The survey aimed to (1) Document examples of the occurrence of genetic testing in young people for non-medical reasons, in the countries where guidelines exist. (2) Gain information about the psychosocial outcomes of such testing, and (3) Gain information about clinical geneticists’ reasons for providing or refusing tests and their opinions of existing guidelines.

4.3.2.2 Participants

In choosing the participant population, I assumed most cases of predictive genetic testing in young people for non-medical reasons would involve a clinical geneticist, in addition to a genetic counsellor, given the controversial nature of such tests. The survey was thus targeted at clinical geneticists. All members of the following associations who had e-mail addresses were invited to participate in the web-based survey between June and September 2003: the American Society of Human Genetics (ASHG) (Medical Doctors only); the Australasian Association of Clinical Geneticists (AACG) and the Clinical Genetics Society (CGS) of the UK. I received completed surveys as anonymous e-mails. One reminder notice was sent to all potential participants via e-mail.

Ethics Approval was granted by the Royal Children’s Hospital Ethics in Human Research Committee in Melbourne, Australia (reference number EHRC23065B).

4.3.2.3 Instrument

The survey was validated in three stages. In stage one I sent a preliminary questionnaire to 10 clinical geneticists in order to gain information about inclusion criteria. Stage two involved content validation by an expert panel who commented on
and revised the survey. This panel consisted of two clinical geneticists, two ethicists, a molecular geneticist, an epidemiologist, a genetic counsellor, a genetics education researcher with expertise in questionnaire design and a lay person. Finally, I piloted the survey on Australasian participants before inviting subsequent participants to respond.

The survey consisted of 17 questions in forced-choice and short answer formats. The survey remains on-line (http://www.mcri.edu.au/pages/Rony/Questionnaire2.asp?). Quantitative aspects of the survey included, but were not limited to, information about the number of tests clinicians had performed for non-medical reasons, demographic information about respondents, the number of tests clinicians had refused and clinicians’ views of existing guidelines. Qualitative aspects of the survey included, but were not limited to, the identification of cases where a predictive genetic test had been provided to someone under the age of 18 years who was asymptomatic for a condition where onset may be in adulthood, and the description of what follow-up had demonstrated in these cases. Clinicians were also asked to give reasons for their views of current guidelines.

4.3.2.4 Analysis
Given the combination of forced-choice and short-answer questions, analysis of the survey involved both quantitative and qualitative methods of analysis. Quantitative analysis was performed using SPSS 11.5 (Statistical Program for the Social Sciences, SPSS Inc, Chicago) for descriptive statistics. As surveys were received electronically, responses were transferred directly into SPSS in order to reduce the opportunity for human error. Ten percent of the data in SPSS was cross-checked against the original e-mails received in order to verify the reliability of this method of automatic transfer. E-mails were identifiable by the time and date they were received.

Qualitative analysis of the survey was performed in two phases. Firstly, in order to ensure that the tests reported were performed for non-medical reasons, as opposed to medical ones, clinicians’ reasons for providing each test were analysed. This analysis was performed by Associate Professor Martin Delatycki; a clinical geneticist. Although the survey specifically asked for details of cases that were performed for conditions that were not treatable or preventable (ie. tests that were performed for
non-medical reasons), many respondents did in fact include such cases. Medical reasons for test provision included a suspicion of symptoms in the young person and the availability of prophylaxis in the event of a gene-positive result. This is important because it indicates that past surveys measuring similar occurrences, but lacking in such detail, may have unknowingly reported tests that were performed for medical reasons. I therefore reiterate here that although some of the conditions I report on can have their onset in childhood, all reports refer to tests that were performed for non-medical reasons.

The second phase of qualitative analysis consisted of Associate Professor Martin Delatycki and myself independently coding the four major qualitative aspects covered by the survey. These aspects were: (1) Clinicians’ reasons for providing the tests (2) Outcomes of the test, as identified through follow-up (3) Clinicians’ reasons for refusing such tests, and (4) Clinicians’ justifications for the views they hold about existing guidelines. The codes generated for each of these aspects are listed in Table 4.1.
### Table 4.1 Codes used to analyse qualitative aspects of the survey

<table>
<thead>
<tr>
<th>Aspect of Qualitative Research</th>
<th>Codes Generated</th>
</tr>
</thead>
<tbody>
<tr>
<td>(1) Clinicians’ reasons for providing the tests</td>
<td>(a) To plan for the future, (b) To resolve uncertainty, (c) Because of reproductive reasons (including that the person being tested was pregnant, their partner was pregnant, or they planned on starting a family in the near future), (d) To resolve parental anxiety (or because parents had requested the test), and (e) The test was performed prenatally.</td>
</tr>
<tr>
<td>(2) The outcomes of the test, as identified through follow-up</td>
<td>(a) positive event (b) adverse event, and (c) neutral event. An adverse event was defined as any outcome that is potentially negative for the individual involved, a positive event was defined as any outcome that is potentially beneficial for the individual involved and a neutral event was defined as any outcome that was not adverse or beneficial.</td>
</tr>
<tr>
<td>(3) Clinicians’ reasons for refusing such tests</td>
<td>(a) In order to protect the autonomy of the young person, (c) Counselling resolved the issue, (d) In order to prevent harm, (e) There was no medical benefit to the test, (f) Because of policy, and (g) Because of privacy</td>
</tr>
<tr>
<td>(4) Clinicians’ justifications for the views they hold about existing guidelines.</td>
<td>(a) It’s a case-by-case decision, (b) It’s about weighing up benefit and harm, (c) It’s not the clinician’s decision, it should be up to the family involved, (d) The guidelines are logical, and (e) We need to protect the autonomy of young people</td>
</tr>
</tbody>
</table>

### 4.3.3 Phase two: Interviews with young people

In this section I describe how I prepared for, completed and evaluated 18 in-depth interviews with young people who had undergone predictive genetic tests themselves.
4.3.3.1 Aims

The aims of these interviews were to (1) Identify a broad range of non-medical benefits and harms of predictive genetic testing in young people, as described by those who have experienced such tests, and (2) Provide an opportunity for the emergence of new ways to understand the experience of such testing for young people.

4.3.3.2 Planning

The planning phase for the interviews involved an initial focus group with young people who suffered from chronic illness. These young people were part of a support group known as Chronic Illness Peer Support, or ‘ChIPS’. The focus group I facilitated consisted of five young people between the ages of 15 and 19 years, and was performed in order to learn about the types of research methods that would appeal most to young people. I sought opinions on potential locations for interviews, possible means young people could use to describe their experiences and methods of inviting participation and capturing the interest of young people.

4.3.3.3 Participants

In choosing my participant population, I consulted with Associate Professor Martin Delatycki, a clinical geneticist at Genetic Health Services Victoria (GHSV). He was able to provide me with anonymous information about the number of potential participants who had undergone predictive testing for a range of genetic conditions at GHSV. Initially, I aimed to interview young people with two specific traits: (1) an age less than 18 years at the time of testing and (2) first-hand knowledge of a predictive genetic test performed for non-medical reasons. However, as there were so few young people who fitted both these criteria for the primary target population, I widened my participant population, aiming to access two populations with similarities to the primary population. Firstly, young people who were under 18 years of age but had undergone predictive genetic testing for medical reasons. Secondly, young people who had undergone predictive genetic tests for non-medical reasons, and though not under 18 years of age were still young enough to fit into some standard definitions of a “young person” (under the age of 25 years). Figure 4.1 represents the three distinct participant populations I targeted. The group sitting within the overlapping segment in this venn diagram represents my primary participant population of interest.
In qualitative research, it is not usual to pre-empt the number of participants that will be required. Instead, participants cease to be sought when a level of “saturation” has been reached in the analysis process. In thematic analysis, saturation occurs when the emergence of new themes becomes rare and older themes repeat themselves. The researcher makes the decision about when this stage has been reached (p 196).\footnote{144}

Ethics Approval was granted by the Royal Children’s Hospital Ethics in Human Research Committee in Melbourne, Australia (reference number EHRC23065B).

**4.3.3.4 Preparation**

I undertook several means that were designed to prepare me, personally, for the task of performing the interviews with young people. These included:

1. Participation at two workshops related to ‘working with and understanding young people’, held at the Centre for Adolescent Health, Royal Children’s Hospital, Melbourne.
2. Completion of a 60-hour telephone counselling training course, held at AIDS, Hepatitis and Sexual Health Line and accredited by the National Metropolitan Institute of TAFE in Melbourne.
(3) Participation in weekly, 3-hour counselling shifts at AIDS, Hepatitis and Sexual Health Line, upon completion of the associated training course.
(4) Participation in the counselling component of the Genetic Counselling Course offered by Genetic Health Services Victoria.
(5) Participation in a series of ‘mock’ interviews with a psychotherapist, Dr John Rogers.

In addition to these means of preparing myself for the commencement of the interviews, during this time I consulted widely with professionals in the areas of genetics, ethics, counselling, epidemiology and qualitative research in order to devise and refine my interview theme list. The interview theme list I used for the interviews I performed with young people who had undergone testing for HD is presented as Appendix A. The interview theme list I used for the interviews I performed with young people who had undergone testing for FAP was almost identical to this, but referred to FAP instead of HD.

Letters of invitation were sent out to the three target groups. These letters were sent by clinicians working at Genetic Health Services Victoria. At this stage, for privacy reasons, I had no knowledge of who the letters had been sent to. Within each package that was sent out was a letter from the clinician who had been involved with the individual’s predictive genetic test, a letter from myself, a plain language statement and a CD. The CD was produced with help from the Education and Resource Centre at the Royal Children’s Hospital and could be played on a computer or in a CD player. If played on a computer, potential participants could watch and listen to me explain what was involved with participation in the research project. If played in a CD player, potential participants could listen to me only. The information I presented on the CD was based on the information contained within the plain language statement, which was sent to potential participants at the same time. The script I used on the CD is presented as Appendix B. The letter and plain language statement I sent to young people who had undergone testing for HD are presented as Appendices C and D. The letter and plain language statement I sent to young people who had undergone testing for FAP were almost identical to these, but referred to FAP instead of HD.
For potential participants who were under the age of 18 years, consent was also sought from their parents. In these cases, parents were sent a letter and a plain language statement three days prior to their children being sent information. The letter that was sent to parents of young people who had undergone testing for FAP is presented as Appendix E. There were no participants under the age of 18 years who had undergone testing for HD. Clinicians contacted all potential participants who had not responded in a month by telephone, in order to check that they had received the package and that they had in fact chosen not to participate. This was done in the knowledge that such a phone call could be interpreted as pressure to participate and clinicians were thus careful to emphasise that this was not the impetus for the telephone call. Interested participants were encouraged to get in contact with myself either by telephone, e-mail or text-message. I then clarified the process of involvement with these individuals over the phone and answered any questions they had. A time was then made to perform each interview. Locations suggested for the interviews included participant’s homes and the Royal Children’s Hospital, Melbourne. Participants were advised that I would prefer no-one else to be present during the interview, unless they felt it absolutely necessary.

Forty-eight letters of invitation were sent out to individuals who had undergone a predictive genetic test for FAP, while twenty-one letters of invitation were sent out to individuals who had undergone testing for HD. Forty-five phone-calls were made to individuals who had received a letter of invitation but had not responded. Ten interviews were performed with individuals who had undergone predictive genetic testing for FAP, giving a participation rate of 20.8%. Eight interviews were performed with individuals who had undergone testing for HD, giving a participation rate of 38.1%.

Of the ten young people interviewed who had undergone predictive genetic testing for FAP, four were male and six were female. Five of these individuals received a gene-positive test result (two female and three male) and five received a gene-negative test result (four female and one male). At the time these individuals were tested their ages ranged from 10 years to 17 years. At the time I interviewed these individuals their ages ranged from 14 years to 25 years. One of these individuals has a child.
Of the eight young people interviewed who had undergone predictive genetic testing for HD, four were male and four were female. Two of these individuals received a gene-positive test result (one male and one female) and six received a gene-negative test result (three male and three female). At the time these individuals were tested their ages ranged from 17 years to 25 years. At the time I interviewed these individuals, their ages ranged from 20 years to 26 years. Two of these individuals have a child.

4.3.3.5 Interviews

I performed all interviews. In initiating each interview the only information I had about participants was their name and the condition for which they had been tested. I had no knowledge of their test result until this was disclosed to me during interviews. All participants were told that, should they feel the need for counselling after the interview, or in the months following it, a session could be arranged with a genetic counsellor at no charge to them. Participants received a double movie-pass at the end of the interview as a token of appreciation for their time and contribution. Participants were not aware they would receive this movie-pass when deciding to participate in the interviews.

Interviews were recorded using a digital audio recorder and files were then downloaded onto a computer. All computer files were stored in an anonymous format and were password protected. All additional identifiable information was stored in a locked filing cabinet at the Royal Children’s Hospital, Melbourne.

I transcribed all interviews for later analysis. All identifying information was altered during this process. The average length of each transcript was approximately 11,500 words, with the shortest transcript consisting of approximately 6,700 words and the longest consisting of approximately 20,500 words.

4.3.3.6 Analysis

Qualitative analysis of the in-depth interviews consisted of thematic analysis, in two distinct phases. Phase one involved the use of two units of analysis: harm and benefit. The second phase of my thematic analysis involved the discarding preconceived categories and theories, allowing themes, concepts and theories to emerge from the data itself. In this second phase my aim was to analyse the data more holistically.
Thematic analysis was organised using the qualitative research software package NVIVO (NVIVO 2.0, QSR International Pty Ltd). All transcribing and analysis was performed by myself, though 3 interview transcripts (>16%) were also co-coded by two additional researchers: Dr Lynn Gillam and Associate Professor Martin Delatycki. I decided to cease conducting interviews once a point of saturation had been reached in my analysis. That is, once the same themes continued to emerge in interviews and the emergence of new themes became rare. I conducted, transcribed and analysed 18 interviews in total.

During the time I was performing the interviews with young people, Dr John Rogers (psychotherapist and clinical geneticist) was supervising me. This supervision consisted of replaying the audio recordings of interviews and analysing, in detail, the way in which I phrased questions, responded to participants and ordered topics within the interview.

All participants received a summary of my major research findings upon completion of the interviews and subsequent analysis.

4.3.3.7 Difficulties encountered in performing interviews with young people

While planning and preparing for the interviews I conducted with young people, I met with two major challenges. The first major challenge related to the existence of ‘gatekeepers’. That is, individuals who hold the ‘pragmatic keys’ of access to participants. The concept of a gatekeeper has been noted often. Rice and Ezzy, in their discussion about performing interviews as part of qualitative research, note that:

“It may be essential to gain permission from formal or informal gatekeepers. … Extra time taken to obtain these approvals will typically result in much easier access to participants” (p 62).  

Morrill, Buller, Buller and Larkey also refer to gatekeeping. They describe the initial question that needs to be asked in order to avoid difficulties further on:

“Whose approval do we need to obtain in order to conduct our research in this organisation?” (p 58).
Fielding notes that seeking permission from gatekeepers is not always possible:

“Obtaining a permit in most circumstances is just a matter of formality. However, in some cases, it can be difficult or even impossible to gain access to the research site.” (p 159).

In my case, although the theoretical key of Ethics Committee approval had been obtained, gatekeepers subsequently withheld information that was vital for my access to potential participants. These individuals were concerned with my lack of clinical experience, specifically counselling experience, and were the impetus for my involvement in the several different preparatory methods I undertook prior to performing the interviews. It was not until these measures had been taken that I was provided with access to potential participants by these gatekeepers.

It has been noted that research with young people often attracts specific difficulties. Hood and colleagues refer to difficulties in accessing young people for research:

“In considering access to children, adults gave priority to adult duty to protect children from outsiders; this took precedence over children’s right to participate in the decision to talk with us” (p 123).

Gilbertson and Barber also note that “given the problems in gaining access to children, the absence of their voices in the literature is not surprising” (p 255). Several factors have been suggested as contributing to this common struggle in accessing young research participants. The need to protect children from harm, protect their autonomy and obtain proper informed consent have all been referred to as ethical challenges related to research with children. Assumptions about the difficult nature of research with young people has also led to obstacles to their involvement, as Coupey explains here:

“Many clinicians believe that adolescents are more difficult to interview than patients in other age groups. There is some truth to this perception. Adolescence is an in-between age, neither childhood nor adulthood, and effective interviewing strategies require particular attention to interpersonal communication skills” (p 1349).
However, it is important to remember that research with young people can also be beneficial for them. Weithorn and Scherer remind us that “the experience of having a voice may give children practice in making life decisions and may help develop perceptions of control… Children’s participation in decision making may also promote a sense of being responsible for their own lives versus being “powerless victims of the whims of adults” (p 76). Stalker and colleagues, in their discussion of the difficulties that arise in gaining access to children for research purposes, write of a requirement for balance:

“There is a balance to be struck between measures that ensure high standards are in place, and measures that may impose unnecessary restrictions on potentially worthwhile research” (p 380).  

Thus, especially when participant populations are minors, researchers may encounter a need for several keys, in addition to the one stamped ‘Ethics Committee approval’, in order to gain genuine access to the participant population they are interested in.

The second major difficulty I faced when preparing for the interviews I conducted related to incentives provided to research participants. There is ongoing controversy about incentives provided to research participants. Rice and Ezzy note that:

“In marketing research, participants are paid. In health research however, there are debates about this. … Some researchers argue that payment is necessary if the researcher needs to recruit those who are ‘hard to find’ because of their busy schedules. Other researchers also argue that payments should be made to poor people” (p 84).  

Curtis and colleagues describe the “tricky question in any research setting” of incentives or rewards, yet explain that in their research study it was agreed young people would receive a gift voucher, reflective of the participant’s age and the length of the interview (p 170).  

In my initial focus group with young people from ChIPS, we spoke about motivations for participation in research projects. During this discussion, a strong theme of material incentives became apparent. That is, when I spoke with these young people
about what would potentially motivate them to take part in a research project, they 
highlighted the need for provision of a material bonus. Suggestions they offered 
include a CD voucher or a movie pass. Thus, initially when I applied for Ethics 
Committee approval, I proposed that I would provide each participant with a double 
movie pass as a token of appreciation and that I would make this known in the letter 
of invitation in order to help motivate participation. The Ethics Committee rejected 
this proposal. A lengthy debate ensued about the need for caution in relation to 
incentives for research participants and specifically the potential for harm through 
coercion.

During this discourse with the Ethics in Human Research Committee I argued that 
such an incentive would not cause young people who did not want to participate in 
my interviews to change their minds, thus being coerced. I argued that the provision 
of a double movie pass would cause no greater potential for harm. However, I was not 
persuasive. Ethics Committee approval was finally granted, months later, on the 
condition that I provided the movie passes at the end of each interview only and that 
participants had no knowledge of this provision prior to the final moments of each 
interview.

4.4 Conclusions

In this fourth chapter I have explained why I chose to conduct the empirical research 
that I describe in the second part of this thesis. I began with a description of two major 
gaps in existing literature concerning predictive genetic testing in young people and I 
then clarified why empirical research is necessary. I argued a need for researcher 
reflexivity and, accordingly, recounted the epistemology, theoretical framework and 
methodologies that formed the scaffolding of the research I performed. Embedded 
within this account was a defence of combining both quantitative and qualitative 
methodologies. Finally, I described in detail the specific methods employed in 
conducting an international survey of clinical geneticists and 18 in-depth interviews 
with young people. This chapter completes part one of this thesis. In the next chapter, 
I begin to present the findings of these two phases of empirical research.
CHAPTER FIVE: CURRENT PRACTICE
5.1 Introduction

“The patient was obsessed with knowing if he was affected”

- Clinical Geneticist

“[The] father wanted to know if either of his daughters had a breast cancer gene so he could send the other to college”

- Clinical Geneticist

“I have a feeling that even if we (the genetics community) do not recommend testing, a family that really wants it is going to get it, no matter what we say”

- Clinical Geneticist

Chapter five initiates the second part of this thesis. In part one (chapters 1-4) I set the scene around the current debate concerning predictive genetic testing in young people. In part two of this thesis (chapters 5-7) I reveal the findings of my own empirical research.

Here, I reveal the findings of the first phase of empirical research: an international survey of clinical geneticists working in the field of predictive genetic testing. I begin by describing the individuals who responded to my survey. I then move through a description of the predictive genetic tests that have been provided to young people for non-medical reasons and the psychosocial outcomes of these. Next, I outline clinicians’ reasons for refusing such tests on other occasions and, finally, I share clinicians’ views of the existing guidelines concerning predictive genetic testing in young people. The chapter ends with a discussion of the limitations of my survey and a summary of my findings.
5.2 Survey respondents

The survey was sent to 1732 members of the American Society of Human Genetics (ASHG), 98 members of the Australasian Association of Clinical Geneticists (AACG) and 400 members of the Clinical Genetics Society in the UK (CGS). I received 347/1732 responses from members of the ASHG (20.0% response rate), 41/98 responses from members of the AACG (41.8% response rate) and 84/400 responses from members of the CGS in the UK (21.0% response rate). This gave an overall response rate of 21.2%. In sending the survey to members of the ASHG, I was not able to target clinical geneticists specifically. Instead the survey was sent to all medical doctors. Thus, many of these recipients may not have been part of my target population and may therefore have simply ignored the survey. Thus, if the survey had been sent only to members of the ASHG who were clinical geneticists, the response rate for this group may have been higher.

Of the 472 responses I received, 301 respondents indicated that they were professionally involved in predictive genetic testing and thus these 301 responses were analysed in detail. Tables 5.1 and 5.2 present the job titles and countries of employment for these 301 respondents. A small proportion of the respondents were genetic counsellors. I assume this was due to the survey being passed on.
<table>
<thead>
<tr>
<th>Job title</th>
<th>Proportion of respondents</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Geneticist</td>
<td>254 (84.4%)</td>
</tr>
<tr>
<td>Genetic Counsellor</td>
<td>12 (3.99%)</td>
</tr>
<tr>
<td>Other (eg. Neurologist, Paediatrician)</td>
<td>18 (5.98%)</td>
</tr>
<tr>
<td>Job title not given</td>
<td>17 (5.65%)</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>301</strong></td>
</tr>
</tbody>
</table>
Table 5.2 Respondents’ country of employment

<table>
<thead>
<tr>
<th>Country of employment</th>
<th>Proportion of respondents</th>
</tr>
</thead>
<tbody>
<tr>
<td>USA</td>
<td>178 (59.1%)</td>
</tr>
<tr>
<td>UK</td>
<td>62 (20.6%)</td>
</tr>
<tr>
<td>Australia</td>
<td>30 (9.97%)</td>
</tr>
<tr>
<td>Canada</td>
<td>3 (1.00%)</td>
</tr>
<tr>
<td>New Zealand</td>
<td>3 (1.00%)</td>
</tr>
<tr>
<td>Other</td>
<td>8 (2.66%)</td>
</tr>
<tr>
<td>Country not given</td>
<td>17 (5.65%)</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>301</strong></td>
</tr>
</tbody>
</table>

In the next section, I describe the predictive genetic tests that have been provided to young people for non-medical reasons by these clinicians. I also describe the psychosocial outcomes of these tests, as reported by the clinicians.

5.3 Occurrence of testing

Thirty-six clinicians (12%) have been involved in the provision of predictive genetic tests to young people for non-medical reasons. These clinicians gave details of 49 cases covering 14 different conditions. That is, the international survey of clinical geneticists uncovered 49 cases in which clinicians have provided tests to young
people, in direct conflict with existing guidelines. These cases constitute the largest and most descriptive collection of such cases to date.

In chapter one I made a distinction between immature and mature young people, yet refrained from associating a specific age range with either category. However, in order to separate the responses to my survey into these two categories, it is necessary to choose a specific age at which to make the distinction between immature and mature young people. Guidelines published by the Society for Adolescent Medicine and recent literature demonstrates that young people have decision-making capacity comparable to adults from the age of 14 years.\textsuperscript{102,105,107,108,166} When conveying the findings of my survey in this chapter, I therefore refer to young people who are 14 years of age or older as mature and those who are under the age of 14 years as immature.

Twenty-two of the 49 cases in which testing was provided to young people for non-medical reasons involved immature young people (45%). Mature young people were involved in the remaining 27 cases (55%).

\textbf{5.3.1 Immature young people}

Of the 22 cases in which predictive genetic testing was performed on immature young people for non-medical reasons, four tests were performed prenatally. These were all tests in which a gene-positive result was received, but the pregnancy was then continued. Only one test was provided to a young person between the age of 10 and 14 years, meaning that 17 of the 22 tests were performed on young people under 10 years of age. Seven immature young people were female, 13 were male and in 2 cases the clinicians did not provide information about gender.

The most common conditions tested for were HD and myotonic dystrophy, with four tests being performed for each of these conditions. Tests were also performed for a range of other genetic conditions, including Becker muscular dystrophy, spinocerebellar ataxia and Charcot-Marie Tooth disease. Twelve tests produced a
gene-positive result and nine produced a gene-negative test result. In one case the result of the test was not indicated in the survey response.

In 18 of the 22 cases where tests were provided to immature young people, the parents had requested testing of their child. In the remaining 4 cases details were not provided about who requested testing. Results have only been disclosed to two of the immature young people who were tested. In one of these cases the young person tested was 10 years old and had received a gene-negative test result for spinocerebellar ataxia. In the other case the young person tested was 9 years old and had received a gene-negative test result for fascio scapulo humeral dystrophy (FSHD).

Table 5.3 presents a summary of the 22 cases in which a predictive genetic test was provided to an immature young person for non-medical reasons.
Table 5.3 Cases in which tests were performed on immature young people

<table>
<thead>
<tr>
<th>Age of person tested</th>
<th>Prenatal test:</th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0-4 years:</td>
<td>8</td>
<td>(36%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>5-9 years:</td>
<td>9</td>
<td>(41%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>10-14 years:</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Gender of person tested</td>
<td>Female:</td>
<td>7</td>
<td>(32%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Male:</td>
<td>13</td>
<td>(59%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No Response:</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Condition tested for</td>
<td>Huntington Disease:</td>
<td>4</td>
<td>(18%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Myotonic Dystrophy:</td>
<td>4</td>
<td>(18%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Charcot-Marie Tooth:</td>
<td>3</td>
<td>(14%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Becker Dystrophy:</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Von Hippel Lindau:</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Spinocerebellar Ataxia:</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dystonia:</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Friedreich Ataxia:</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Kennedy Disease (SBMA):</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>MODY:</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>FSHD:</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Results of test</td>
<td>Increased risk:</td>
<td>12</td>
<td>(55%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Decreased risk:</td>
<td>9</td>
<td>(41%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Don’t Know:</td>
<td>1</td>
<td>(4.5%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test requested by</td>
<td>Parent(s) or guardian(s):</td>
<td>18</td>
<td>(82%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Other (details not given):</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No response:</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Was result disclosed to individual who was tested?</td>
<td>Yes:</td>
<td>2</td>
<td>(9.1%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No:</td>
<td>14</td>
<td>(64%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Don’t Know:</td>
<td>6</td>
<td>(27%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total number of cases</td>
<td></td>
<td>22</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*aSMBA  = Spinal Bulbar Muscular Atrophy
*bMODY = Maturity Onset Diabetes of the Young
*cFSHD = Fascio Scapulo Humeral Dystrophy
I now describe the reasons clinicians cited for providing these tests to immature young people, given that it was in conflict with existing guidelines concerning predictive genetic testing in young people.

5.3.1.1 Why were these tests provided to immature young people?

There were several non-medical reasons for provision of these 22 tests to immature young people. The most common reason articulated was that parents wanted to know, with 10 clinicians (45%) citing such a reason:

“Parent had suffered through years of misdiagnosis unable to succeed in sports though otherwise asymptomatic, wanted to avoid this for child”

“Parental concern”

“The father was affected – wanted to know about his daughter”

“Parents wanted information”

Three clinicians (14%) cited the opportunity for future planning as a reason:

“Preparation for the future”

“High risk planning for the future”

“They wanted to know if he had the mutation ... His mother was very disabled and the children were being placed in other homes

Table 5.4 presents a summary of clinicians’ reasons for providing predictive genetic tests to immature young people.
Table 5.4 Clinicians’ reasons for performing tests on immature young people

<table>
<thead>
<tr>
<th>Clinician’s reason for providing the test</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Because parents wanted to know</td>
<td>10 (45%)</td>
</tr>
<tr>
<td>To help in future planning</td>
<td>3 (14%)</td>
</tr>
<tr>
<td>The test was provided prenatally</td>
<td>4 (18%)</td>
</tr>
<tr>
<td>Other/No Response</td>
<td>5 (23%)</td>
</tr>
<tr>
<td><strong>Total number of tests provided to immature young people</strong></td>
<td><strong>22</strong></td>
</tr>
</tbody>
</table>

5.3.2 Mature young people

Of the 27 cases in which predictive genetic testing was provided to mature young people, the majority of cases involved individuals of 16 and 17 years of age, with eighteen tests provided to mature young people in this age range. Nine tests were provided to mature young people of 14 or 15 years of age. Sixteen mature young people were female and the remaining 11 were male.

The majority of tests provided to mature young people were performed to determine risk status for HD, with 14 tests being performed for this condition. Other conditions tested for included myotonic dystrophy, breast cancer predisposition and spinocerebellar ataxia. Sixteen of the tests produced a gene-negative result, nine produced a gene-positive result and in two cases the result of the test was not indicated in the survey response.

In four cases, the predictive genetic test was requested by the young person’s parents and in 10 cases the test was requested by the young person alone. In 13 cases both the young person and his or her parents requested the predictive genetic test together.
Results were disclosed to all mature young people who were tested except for one individual who was intellectually disabled (aged 14 years).

Table 5.5 presents a summary of the 27 cases in which predictive genetic tests were provided to mature young people.
### Table 5.5 Cases in which tests were provided to mature young people

<table>
<thead>
<tr>
<th>Age of person tested</th>
<th>14-15 years:</th>
<th>16-17 years:</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>14-15 years:</td>
<td>9</td>
<td>18</td>
<td>(33%)</td>
<td>(67%)</td>
</tr>
<tr>
<td>16-17 years:</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gender of person tested</th>
<th>Female:</th>
<th>Male:</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Female:</td>
<td>16</td>
<td>11</td>
<td>(59%)</td>
<td>(41%)</td>
</tr>
<tr>
<td>Male:</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Condition tested for</th>
<th>Huntington Disease:</th>
<th>Myotonic Dystrophy:</th>
<th>Breast Cancer (BRCA1 &amp; 2):</th>
<th>FSHD:</th>
<th>Spinocerebellar Ataxia:</th>
<th>Balanced Translocation associated with manic-depressiveness:</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>14</td>
<td>5</td>
<td>3</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td>(52%)</td>
<td>(19%)</td>
</tr>
<tr>
<td>5</td>
<td>3</td>
<td>2</td>
<td>2</td>
<td></td>
<td></td>
<td>(11%)</td>
<td>(7.4%)</td>
</tr>
<tr>
<td>3</td>
<td>2</td>
<td>2</td>
<td></td>
<td></td>
<td></td>
<td>(7.4%)</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>(7.4%)</td>
<td></td>
</tr>
<tr>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>(3.7%)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Results of test</th>
<th>Increased risk:</th>
<th>Decreased risk:</th>
<th>Don’t Know:</th>
<th>No response:</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>14</td>
<td>16</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>3</td>
<td>2</td>
<td>2</td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>2</td>
<td>2</td>
<td>2</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>1</td>
<td></td>
<td>1</td>
<td>1</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Test requested by</th>
<th>Parent(s):</th>
<th>Young person:</th>
<th>Both Parent(s) and young person:</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>4</td>
<td>10</td>
<td>13</td>
<td>(15%)</td>
<td>(37%)</td>
</tr>
<tr>
<td>10</td>
<td>10</td>
<td>13</td>
<td>(37%)</td>
<td>(48%)</td>
</tr>
<tr>
<td>13</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Was result disclosed to individual who was tested?</th>
<th>Yes:</th>
<th>No:</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>26</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>(96%)</td>
<td>(3.7%)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Total number of cases</th>
<th>27</th>
</tr>
</thead>
</table>

\[\text{FSHD} = \text{Fascio Scapulo Humeral Dystrophy}\]

I now describe the reasons articulated by clinicians for providing these tests to mature young people.
5.3.2.1 Why were these tests provided to mature young people?

There were several non-medical reasons for provision of tests to mature young people. The most common reason for testing was to resolve uncertainty for the young person, with 13 clinicians (48%) citing this as a reason:

“The patient was obsessed with knowing if he was affected”

“Resolve uncertainty”

“Stressful living with uncertainty”

“Parent recently died of disease, teen concerned”

Seven of these requests were made by the young person and six requests were made by the young person and his or her parents together.

Six clinicians (21%) cited the opportunity for future planning as a reason for test provision:

“To plan life and career appropriately”

“Future management”

“Life choices”

“Life and management decisions”

Three of these requests were made by both the young person and his or her parents, two requests were made by the parents alone and one request was made by the young person alone.

Two clinicians (7.4%) provided tests because of parental anxiety:
“Parents wanted to know status. 16 year old was not averse but ++++ pressure from mother”

“Parental anxiety”

Both of these requests were made by the parents.

Two clinicians (7.4%) also provided tests due to reproductive reasons:

“partner pregnant”

“Reproductive risk”

One of these requests was made by the young person and the other request was made by both the young person and her parents. Table 5.6 summarises clinicians’ reasons for providing tests to mature young people.
Table 5.6 Clinicians’ reasons for providing tests to mature young people

<table>
<thead>
<tr>
<th>Clinician’s reasons for providing the test</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>To resolve uncertainty</td>
<td>13 (48%)</td>
</tr>
<tr>
<td>To help in future planning</td>
<td>6 (22%)</td>
</tr>
<tr>
<td>Because of parental anxiety</td>
<td>2 (7.4%)</td>
</tr>
<tr>
<td>Because of reproductive reasons</td>
<td>2 (7.4%)</td>
</tr>
<tr>
<td>Other/No response</td>
<td>3 (11%)</td>
</tr>
<tr>
<td><strong>Total number of tests provided to mature young people</strong></td>
<td><strong>27</strong></td>
</tr>
</tbody>
</table>

In the past two sections I have presented details of the 49 cases in which predictive genetic tests were provided to young people for non-medical reasons, in conflict with existing guidelines. I have also described the specific reasons for test provision in these cases. I now move to the time after testing, in order to report some of the psychosocial outcomes of these tests.

### 5.4 Effects of testing

Accounts of the psychosocial outcomes of testing that were provided by clinicians may be limited by three factors. Firstly, clinicians were asked to provide details of outcomes in cases where they provided tests contrary to recommendations made in current guidelines. Although the survey was anonymous, it is possible that clinicians under-reported harmful outcomes. Secondly, follow-up did not occur in many cases and in those that it did, it was not systematic. This means that there may have been additional significant outcomes that were simply not known about. I come back to this
point concerning lack of follow-up later in this chapter. Lastly, there were only two known disclosures of test results to the 22 immature young people who underwent predictive genetic testing. Thus, in the majority of these cases, it is too early to draw any conclusions about the outcomes of testing.

5.4.1 Immature young people

There were no reports of harmful outcomes in immature young people who were tested although, to emphasise the point once more, only two of these young people were aware of their test results. The most interesting findings concerning the outcomes of predictive genetic testing in immature young people relate to the impact of testing on the parents of these individuals.

There were three reports of harmful outcomes for the parents of immature young people who were tested, out of 11 cases in which follow-up occurred. Two harmful outcomes followed prenatal gene-positive results for HD, where parents decided to continue the pregnancy. The third harmful outcome followed a gene-positive result for dystonia in an 8-year-old male. It was reported in all three cases that parents felt distressed by the information they had and were feeling anxious about how and when to tell their child:

“Parents distressed by having this information and not knowing how to use it”

“Anxiety about how and when to tell the child”

“Parents were confused and anxious about what to tell their children and when to say something”

There was also one report of a beneficial outcome for the parents of an immature young person who was tested for Charcot-Marie Tooth and received a gene-negative test result:

“Parent relieved, child seemed to have forgotten about the test”
5.4.2 Mature young people

Two harmful outcomes were reported in mature young people who underwent predictive genetic testing. This constitutes a rate of 7.1% across the 28 cases in which the young person was informed of the result and follow-up occurred. The first harmful outcome followed a gene-positive result for HD in a 17-year-old male:

“Initial depression and rebellion but eventual acceptance”

The second harmful outcome followed a gene-negative result for HD in a 17-year-old female:

“No psychological disturbance but worry and responsibility for affected mother and untested brothers”

There were also nine reports of beneficial outcomes in mature young people who were tested. Six of these followed gene-negative results:

“Enabled him to focus on school etc., and parents say behaviour improved and he deals with difficulties in more mature way”

“Good outcome”

“No problems, coping well with result”

“Coping well with no adverse outcome”

“A very mature [person]…who had clearly thought through the issues and made an informed decision”

“Young person was on self destruct course because of family history. Negative result really helped to stabilise life”

And three followed gene-positive results:
“So far she is doing fine and seems to have integrated this information into her thoughts about her future in a healthy way”

“Coped better than many 20 year olds”

“The person is doing fine”

Table 5.7 summarises some of the harmful and beneficial outcomes of predictive genetic testing in young people, as described by clinicians responding to my survey.
Table 5.7 Harmful and beneficial outcomes of testing in young people

<table>
<thead>
<tr>
<th>Gene-Positive Result in Immature Young People</th>
<th>Harmful Outcomes of Testing</th>
<th>Beneficial Outcomes of Testing</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>▪ Parental distress about information</td>
<td></td>
</tr>
<tr>
<td></td>
<td>▪ Parental anxiety about how and when to tell their child</td>
<td></td>
</tr>
<tr>
<td>Gene-Negative Result in Immature Young People</td>
<td>_</td>
<td>▪ Parental relief</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gene-Positive Result in Mature Young People</th>
<th>Harmful Outcomes of Testing</th>
<th>Beneficial Outcomes of Testing</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>▪ Depression</td>
<td>▪ Integration of information into thoughts about future</td>
</tr>
<tr>
<td></td>
<td>▪ Rebellion</td>
<td>▪ Evidence of mature coping strategies beyond chronological age</td>
</tr>
<tr>
<td>Gene-Negative Result in Mature Young People</td>
<td>_</td>
<td>▪ Ability to focus on school</td>
</tr>
<tr>
<td></td>
<td>▪ Worry and responsibility about other family members</td>
<td>▪ Improved behaviour</td>
</tr>
<tr>
<td></td>
<td>_</td>
<td>▪ Ability to deal with difficulties in a more mature way</td>
</tr>
<tr>
<td></td>
<td>_</td>
<td>▪ Evidence that a competent, mature decision was made</td>
</tr>
<tr>
<td></td>
<td>_</td>
<td>▪ Stabilising effect on life</td>
</tr>
</tbody>
</table>

5.5 Follow-up

Of the 22 cases reported in which an immature young person underwent predictive genetic testing, only 11 cases have been followed up (50%). Of the 27 cases reported
in which a mature young person underwent predictive genetic testing, only 18 cases have been followed up (67%). Follow up was far from systematic, occurring in varied ways. Follow-up consisted of a single phone call in some cases, yet regular contact for 6 months in other cases. Follow-up was also conducted by a range of professionals including counsellors, psychologists and clinical geneticists.

Not one of the 49 cases in which a clinician provided a predictive genetic test to a young person has been followed up systematically or as part of a formal research protocol. In some cases, clinicians report that no follow-up occurred at all. I therefore reiterate here that the statistics I quote on frequencies of harmful and beneficial outcomes cannot be used to make inferences about the incidence of these outcomes. The descriptions I provide are valuable because they are qualitative accounts of the types of harmful and beneficial outcomes that can occur. This value is heightened by the existence of detail about the context of each case, such as the age of the young person, the condition tested for, and who requested the test.

In the next section I turn my focus from tests that were provided to tests that were refused. Most interestingly, I outline precisely why clinicians chose to refuse testing in these specific cases.

5.6 Refusals to provide testing

Of the 301 respondents, 159 (52.8%) had refused to perform a predictive genetic test on a young person. These professionals, together, have refused such testing on over 800 separate occasions.

The term ‘refuse’ can be difficult to define. A refusal, as reported, may not necessarily indicate that a young person (or family) was turned away after seeking testing. Rather, it may indicate that the topic of testing was raised and that, following discussion, both the clinician and their client(s) agreed that not testing was the preferred option.
5.6.1 Reasons for clinicians’ refusals

Of the 159 clinicians who have refused to provide a predictive genetic test to a young person, 154 provided a justification for their refusal(s). In many cases, clinicians cited more than one justification.

The most common reason for refusing to provide a predictive genetic test to a young person related to the young person’s autonomy, with 75 clinicians (48.7%) stating this as a justification:

“Wanted to wait until kids were old enough to make their own choices about testing”

“Young person not demonstrating understanding of the implications of testing, therefore unable to give informed consent”

“I feel that they cannot give informed consent to lifetime of a result when they are ‘young’, as we see life differently as we become adults”

“Adolescent may change mind if they waited to consider wide perspectives (some adults change mind)”

“Too young, personality will change”

“Father wanted to have his 17 year old son’s blood drawn to test [for] a mutation found in his recently deceased wife. He wanted the pediatrician to draw blood without informing his son of the purpose. If positive, he would inform his son of the information at a later date; if negative, he would never tell him about the test or the results”

“It’s wrong to test these patients, they need to decide for themselves when they reach legal adult status”
Fifty-three clinicians (34.4%) refused testing because it did not provide a medical benefit:

“No perceived medical benefit for the individual or family for the testing”

“Like any other medical test, if there is no medical benefit, it should not be done”

“No prevention exists”

“No treatment, no benefit”

“Nothing could be done in a positive sense with the data”

“Lack of intervention”

“No change in the management based on testing”

“Did not feel testing was medically necessary or appropriate”

“No impact upon patient’s medical care”

“Testing will not benefit patient in a directly medical fashion”

Twenty-three clinicians (14.9%) refused testing because of a possibility of harm:

“Potential harm to the child...for example...father wanted to know if either of his daughters had a breast cancer gene so he could send the other to college”

“The potential for discrimination”

“Insurance implications”

“Potential for significant social, emotional and economic adverse events”
“There was more risk to the child than benefit”

“Likely to cause child harm”

“Fear of stigmatizing the young child”

“The psychological implications of a positive test”

Eight clinicians (5.19%) cited policy as a reason:

“The clinical scenario did not meet the guidelines for testing established for patients at risk of Huntington Disease”

“General departmental policy”

“Current guidelines”

“Published Huntington disease predictive testing guidelines”

Eight clinicians (5.19%) cited that counselling had resolved the issue:

“I think refused is too strong a word. The family were demanding testing, I simply went over the reasons for not testing. Once they had thought things through the family made their own decision not to test.”

“In discussion about testing the parents usually come to see it would not be helpful”

“Patient’s parents did not have sufficient counselling and it was not explained as to what these tests can do and what they cannot do”

And one clinician (0.65%) cited privacy as a reason:
Table 5.8 presents a summary of clinicians’ reasons for refusing to provide a predictive genetic test to a young person for non-medical reasons.

**Table 5.8 Clinicians’ reasons for refusing to provide tests to young people**

<table>
<thead>
<tr>
<th>Clinician’s reasons for refusing to provide tests</th>
<th>Frequency&lt;sup&gt;Ⅰ&lt;/sup&gt; (154 clinicians)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autonomy</td>
<td>75 (48.7%)</td>
</tr>
<tr>
<td>No medical benefit</td>
<td>53 (34.4%)</td>
</tr>
<tr>
<td>Possibility of Harm</td>
<td>23 (14.9%)</td>
</tr>
<tr>
<td>Counselling resolved issue</td>
<td>8 (5.19%)</td>
</tr>
<tr>
<td>Policy</td>
<td>8 (5.19%)</td>
</tr>
<tr>
<td>Privacy</td>
<td>1 (0.65%)</td>
</tr>
</tbody>
</table>

<sup>Ⅰ</sup> Clinicians often provided more than one reason for their refusals and therefore percentages do not sum to 100.

In the next section I describe clinicians’ views about the existing guidelines concerning predictive genetic testing in young people. I then describe their reasons for holding these views.
5.7 Views on existing guidelines

The majority of clinicians agree with the existing guidelines concerning predictive genetic testing in young people. Table 5.9 presents clinicians’ views of these current guidelines.

Table 5.9 Clinicians’ views of current guidelines

<table>
<thead>
<tr>
<th>Extent of agreement with guidelines</th>
<th>Proportion of clinicians who feel this way</th>
</tr>
</thead>
<tbody>
<tr>
<td>Strongly Agree</td>
<td>104 (34.6%)</td>
</tr>
<tr>
<td>Agree</td>
<td>141 (44.8%)</td>
</tr>
<tr>
<td>Don’t Know</td>
<td>10 (3.32%)</td>
</tr>
<tr>
<td>Disagree</td>
<td>15 (4.98%)</td>
</tr>
<tr>
<td>Strongly Disagree</td>
<td>5 (1.66%)</td>
</tr>
<tr>
<td>No Response</td>
<td>26 (8.64%)</td>
</tr>
<tr>
<td>Total</td>
<td>301</td>
</tr>
</tbody>
</table>

5.7.1 Clinicians justifications for their views on existing guidelines

Of the 275 clinicians who articulated their view on current guidelines, 230 clinicians also provided a justification for this view.

The most common justification cited for clinicians’ views of current guidelines, regardless of their specific view, was that each case needs to be considered
individually. Of the 230 clinicians providing a justification for their view, 99 clinicians (43.0%) cited this as a justification:

“I don’t believe in a rigid cut-off age... as I believe obtaining maturity to gain informed consent is a gradual process”

“I agree there should be guidelines to protect the right of choice of the individual in this situation but the maturity will vary”

“Each case is individual and I don’t feel that guidelines should be prescriptive”

“I agree for most families but for minority of cases it is appropriate to test and this should be done on an individual basis”

“Each family has its own needs, and no simple absolute rule can accommodate all of these”

“18 is arbitrary, but the individual will only gain ability to deal with these issues at some point in adolescence”

“Can’t always be black and white”

“I agree with the guidelines but believe they should be adaptable to individual circumstances”

Eighty-eight clinicians (38.3%) cited the need to protect autonomy as a justification for their view of current guidelines:

“Feel the decision should be up to the individual and question the ability for a minor to truly make the decision if parents involved”

“It is a child’s choice to know or not to know”
“Wish to maintain autonomy, permit ‘normal’ development, sustain hope”

“Right of this person to decide later on their own”

“I believe in the right of the individual to determine what information he or she wants to know about themselves”

“An autonomous, mature decision is required”

“Autonomy cannot be turned aside for the sake of the parents”

“I have had too many adult patients at risk for genetic conditions for which testing is available choose not to undergo testing. The same right of personal decision should be extended to their children”

Seventy-four clinicians (32.2%) cited perceptions of benefit and harm as justifications for their view:

“I think it can only add anxiety, has no benefit”

“Because it would only produce anxiety and potential family problems”

“There is generally no clear benefit to the child, in contrast, there is a possibility for harm”

“We ask parents to weigh the benefits they perceive with the potential disbenefits they may not have considered”

“The potential negative impact of a positive or negative predictive genetic testing result on a child in a family with the disease is huge”

“Medical costs are absurd already, medicine is on the brink of economic collapse or revolution, so we can’t afford to spend money unwisely”
“Information can be used for educational, lifestyle and reproductive choices independent of whether medical intervention is currently available”

“If delay/refusal makes them feel powerless it may do more harm than good”

Thirty-four clinicians (14.8%) cited the logic of current guidelines as a justification for their view:

“Because the guidelines are right”

“The guidelines are well-written, make sense and, in my opinion, promote the welfare of children, families and communities”

“The guidelines reflect most parental and medical opinions about what is best for a minor”

“The guidelines are reasonable and they inform good practice”

Finally, nine clinicians (3.91%) justified their views with a statement about the inappropriateness of clinicians making these decisions:

“It is up to the family to decide and not us. Refusing testing is paternalistic”

“In this country, the overwhelming precedent stands: parents are legally responsible for the health and welfare of their children. If the parents want testing, we provide it”

“The patient has the right to know”

“Parents make routinely thousands of decisions that affect their children’s life in the process of raising them. Most parents are genuinely concerned about doing the right things for their kids”

“Citizen’s right to knowledge”
Table 5.10 presents a summary of clinicians’ justifications for their views on current guidelines concerning predictive genetic testing in young people.

### Table 5.10 Clinicians’ justifications for their views on current guidelines

<table>
<thead>
<tr>
<th>Justification</th>
<th>Frequency f</th>
</tr>
</thead>
<tbody>
<tr>
<td>It’s a case-by-case thing</td>
<td>99 (43.0%)</td>
</tr>
<tr>
<td>Need to protect individual’s autonomy</td>
<td>88 (38.3%)</td>
</tr>
<tr>
<td>It’s all about benefit and harm</td>
<td>74 (32.2%)</td>
</tr>
<tr>
<td>The guidelines are logical</td>
<td>34 (14.8%)</td>
</tr>
<tr>
<td>It’s not the clinicians decision</td>
<td>9 (3.91%)</td>
</tr>
</tbody>
</table>

f Clinicians often provided more than one reason for their refusals and therefore percentages do not sum to 100.

5.8 Limitations of the survey

Several limitations of this survey moderate the conclusions that can be drawn from the data.

First, the low rates of response mean that inferences about current practice more generally cannot be made. Statistics quoted on frequencies of harmful and beneficial outcomes are not representative of the broader predictive genetic testing community. Second, details of the 49 cases I describe were provided by clinicians, not the
individuals who underwent testing. These descriptions are less accurate than first-hand accounts, provided directly by those who underwent testing, would be. Third, clinicians were asked to provide details of cases in which they had provided tests contrary to current recommendations. Although the survey was anonymous, it is possible that clinicians therefore under-reported harmful outcomes. Fourth, follow-up did not occur in several cases and in those that it did it was far from systematic. This means that there may have been additional outcomes, both beneficial and harmful, that clinicians were not aware of. Finally, there were only two reported disclosures of test results to the 22 immature young people who were tested. Thus, in the majority of these cases, it is too early to draw conclusions about the impact of testing for these younger individuals.

A surprising inclusion in the data I received was reports of prenatal tests where, following a gene-positive test result, the pregnancy was continued. There was no mention of a need to include prenatal tests in the instructions contained within the survey. It is therefore possible that most clinicians made an assumption when completing the survey that these types of tests should not to be included. However, descriptions that were provided of these cases highlighted the importance of such inclusion when researching the impacts of predictive genetic testing in young people, especially in relation to the impact that such testing has upon parents. Perhaps then, if I had made it explicit that I was seeking details of prenatal tests as well, I would have received more descriptions of such cases. If so, this would imply that cases of prenatal tests where, following a gene-positive test result the pregnancy was continued, were under-reported in my survey.

In spite of these limitations, this survey provides the most extensive and detailed collection to date of case descriptions in which predictive genetic tests have been provided to young people for non-medical reasons.
5.9 Conclusion

This international survey of clinical geneticists was conducted because of a need to know where we are in the field of predictive genetic testing in young people before we can negotiate where we are going.

We are in a position where, although current guidelines recommend against predictive genetic testing in young people for non-medical reasons, such testing is occurring (albeit sporadically). We now know that in at least 49 cases, a predictive genetic test has been provided to a young person for non-medical reasons, despite current recommendations to the contrary. It can no longer be assumed that such testing will cease to take place until a time when broad consensus is reached within the academic and clinical communities. Testing is happening now. Provision of these 49 tests also implies a desire for such testing. Although assumptions had been made about such desires in the past, there is now empirical evidence to support this assumption.

The survey also provides insights about the types of outcomes that occur when young people undergo predictive genetic testing for non-medical reasons. These include outcomes of testing that have not been identified in previous literature and certainly have not been analysed as part of a research protocol. Such outcomes include the impact that testing has on parents of immature young people who are tested, the impact of testing on school-related behaviour and also the impact of testing on feelings about other family members. Thus a first glimpse into the impacts of predictive genetic testing in young people has uncovered new and important effects that must be considered in clinical practice, research and discourse. These findings can be put to clinical use immediately. For example, the finding that parental anxiety occurs in relation to how and when to inform children about a gene-positive test result can be used to counsel parents requesting prenatal testing and predictive genetic testing in immature young people. We now have a need to include discussion of such risks not only when counselling parents about testing of their immature children but also when counselling parents about prenatal tests, in case the pregnancy is continued following a gene-positive test result.
The survey also raises questions about how to define harmful and beneficial outcomes of predictive genetic testing in young people. I chose to label any negative consequence as a harmful outcome and any positive consequence as a beneficial outcome. However, it could be argued that outcomes such as “initial depression but eventual acceptance” and “worry and responsibility about mother and brothers” are normal and appropriate reactions following receipt of a test result. The magnitude of these harms and benefits must be assessed in debates about the provision of such testing to young people. For example, the possibility of inciting concern about family members may not be a justification for withholding tests, just as the integration of the test result into thoughts about the future may not constitute a justification for providing tests.

Importantly, the positive outcomes of testing that were described by clinicians who were surveyed did not all relate to gene-negative test results, several related to gene-positive test results. Similarly, the two negative outcomes described did not both relate to gene-positive test results, one related to a gene-negative test result.

The survey indicates great clinical support for existing guidelines concerning predictive genetic testing in young people. One specific implication of this, as evidenced by the findings of this survey, is that predictive genetic tests in young people for non-medical reasons are refused far more frequently than they are provided. However, support for current recommendations appears to hinge on a precise understanding of the purpose of guidelines. That is, clinical support for existing guidelines exists because clinicians feel that the stance taken within current guidelines is generally applicable, but that each case needs to be evaluated individually. In other words, current guidelines are appropriate as they help to guide clinical practice, without being absolute.

The survey also indicates great variation in how clinicians evaluate each specific case. Some clinicians provide tests to immature young people because a parental desire to know exists. Others provide such tests because they believe the information will assist in future planning. Some clinicians provide tests to mature young people because of the uncertainty that testing can remove. Others provide such tests because of parental anxiety or for reproductive reasons. Some clinicians refuse tests based on the future
autonomy of the young person. Others refuse them due to a lack of medical benefit. Although clinicians often refer to the same ethical principles when explaining the choices they have made about provision of predictive genetic tests to young people, their decisions are not necessarily uniform. For example, some clinicians argue that because of future autonomy, young people should not undergo testing. They argue that young people should be able to make a decision about testing themselves, later in life. However, other clinicians argue that because of occurrent autonomy, young people should be able to undergo such testing when they wish to do so.

The most common reasons that clinicians refused to provide tests to young people were concerns about autonomy and the lack of medical benefit. The most common reasons they provide tests to mature young people are centred upon a desire to resolve uncertainty and an attempt to assist future planning. The most common reason that clinicians provide tests to immature young people is because parents request such testing.

Clinicians’ reasons for their decisions, perhaps not surprisingly, are as diverse as the cases they are confronted with. Importantly, only 15% of clinicians cited policy as a reason for refusing to provide predictive genetic tests in young people. This indicates that most clinicians are assessing cases individually and are refraining from making simple blanket decisions based on existing guidelines. The decisions that clinicians make appear to be both thoughtful and considered.

There is clearly a need for more research into the outcomes of predictive genetic testing in young people. This survey of 301 clinical geneticists has uncovered 49 cases where such testing has been provided, yet not one of these cases has been published and not one of the young people involved, or their families, have been enrolled in a formal research study. This must change.
CHAPTER SIX: LISTENING TO YOUNG PEOPLE
6.1 Introduction

“The main focus in qualitative research is the data itself, in all its richness, breadth and depth. When all is said and done, the “quality” in a qualitative research project is based upon how well you have done at collecting quality data. So, it only seems natural that when it comes time to present “the fruits of your labor”, you should make every effort to feature the data ... Present as much of the data you collected as is physically possible.”

The advice Ronald Chenail offers above about communicating qualitative data provides the foundation for my strategy in this chapter. I aim here to present young people’s explanations of what predictive genetic testing was like for them. I present their descriptions with the minimal pruning I feel necessary to make sense of their stories. I present their experiences in chronological order, as related to the predictive genetic testing process. In other words, I present the stories that were conveyed to me in the order in which they occurred. The focal point here is the subjective experiences of young people, not my interpretations of these experiences. Here, my interpretation is limited to the selection, categorisation and organisation of young people’s descriptions. The substance of my interpretation of these experiences is presented in the following chapter.

6.2 Young people’s experiences of predictive genetic testing

In presenting the descriptions of predictive genetic testing conveyed to me by young people who have experienced it, I use eight categories. These categories reflect the predictive genetic testing process and consist of: making the decision to be tested, pre-test counselling, the day of the blood test, waiting, the day of the results, immediate impact, long-term impact, and finally, the best and worst aspects.
In chapter four I gave my reasons for choosing two distinct participant populations. That is, young people who have been tested for HD and young people who have been tested for FAP. The fundamental difference between these two types of predictive genetic tests is the existence (or lack) of medical benefit as a consequence of the test. When young people undergo predictive genetic testing for FAP they do so because, if found to be gene-positive, they are able to engage in preventative measures including screening and surgery to avoid the development of colon cancer. Thus a direct medical benefit is created by the test. Alternatively, when young people undergo predictive genetic testing for HD no such medical benefit is created. At this time there is no known means for preventing or treating HD and thus tests for HD are performed for non-medical reasons. However, the existence of medical benefit as an outcome of predictive genetic testing does not negate the potential for harm. It simply overrides this; and the absence of medical benefit does not preclude the possibility of benefit. Predictive genetic testing for FAP is not offered to minors because there is no potential for harm, but rather because the potential for medical benefit is perceived to override the potential for harm. In the case of predictive genetic testing for HD, the absence of medical benefit does not increase the potential for harm, it simply leaves a greater absence of justification for overriding this potential for harm. There is no prima facie reason to assume that the potential for harm should differ greatly between predictive genetic tests provided for FAP and predictive genetic tests provided for HD. I therefore choose not to separate these two types of experiences when conveying the descriptions conveyed to me by young people below.

All identifying details of my research participants have been altered so that participants’ anonymity may be protected.

6.2.1 Making the decision to be tested

Several participants spoke of the time they first became aware of the existence of the predictive genetic test. For some, memories exist about the discovery of the gene itself:
“Shuffle two years forward to 1993 and the gene was discovered and I was 13 and my sister 16 and… yeah, I remember my dad bringing home a bottle of champagne and saying oh we found the gene… this may enable accurate predictive testing and ah… I mean, I always thought about it, right from 1991 when I was 11 years old… Huntingtons and mum getting sick and me having the possibility of it”

**Oliver:** M:24:HD:19:+ve

“I remember when they found the gene, cause we went to that big meeting… I knew that there was a test, but I don’t think I actually knew what that meant”

**Nina:** F:23:HD:23:-ve

For some, the decision to be tested was largely fuelled by their partner’s curiosity:

“It was something first of all that she wanted to know, she said, like, she was fine to continue our relationship but she just kind of needed to know what our future was gonna hold… and I wanted to know as well cause I wanted to start planning my career and like my life as well…She did bring it up a lot more than me, she was very curious about it, which understandably from her point of view she, she would have been and I do understand that”

**Zach:** M:26:HD:23:-ve

“It had a lot to do with Olivia because, as I said, you know, I thought I was going to be with her forever and marry her and have 10 kids or whatever…she influenced my decision a little bit… she was ‘the one’ and we were going to start you know, like planning, you know, future, like what I’m going to do for work when I start getting affected by it, if we have kids, if I decide to have kids or not… so yeah, they were really the main reasons why I got tested”

**Travis:** M:24:HD:20:-ve
Other participants described a strong personal desire to find out their genetic status. Some explained that this was motivated by a longing to plan for their future:

“Oh I just wanted to know, I mean, things like finances, houses, especially in Melbourne you know, where everything’s so dear and everything’s mega bucks, yeah, finances and attitudes, how I treat people I spose”

**Oliver:** M: 24; HD: 19; +ve

“So when I started to really think about what I wanted to do with my future, well I knew that I kind of had to deal with it in some way, whether it was no, all right, I’m just gonna live my life, I’m not gonna take it into consideration, I’ll just go about my normal life anyway… but I kind of felt for me that I really wanted to know so that I could plan a career… and know whether I was gonna have a family in the future”

**Zach:** M: 26; HD: 23; -ve

“I wanted to plan because, you know, what sort of work I’m going to get into, if I want to be a lawyer or a copper or whatever, a factory worker, it doesn’t matter you know, but eventually it would have started affecting my work and… I’d have to sit down with my partner or with whoever I was with and say look I’m going to have to start working part-time …because I can’t put in a full-day, a full week, a full year because I’m just stuffing up too much… so that’s something that I wanted to start planning around too”

**Travis:** M: 24; HD: 20; -ve

“It was something that I really wanted to do too, cause it was like, I just wanted to, I don’t know why, just find out so that I could set my, you know, plan my life ahead sort of thing, like it’s not something that you want to plan all your future and stuff and then you find out something like that, you don’t know how you’re going to react”

**Nina:** F: 23; HD: 23; -ve

Other participants spoke of the difficulty of living with uncertainty when describing their reasons for being tested:
“One night I just got locked up for um drunk and disorderly and I just sort of, I was just talking to me old man and I just said, you know, maybe it might pay off just to go and, you know, maybe it’s in the back of me head or something, just go and do it… and I just said I just want to do it on me own, I just don’t want anyone else there, and sort of, sort of done it… Yeah, well that’s why I went and got the test done, like, I thought, you know, maybe in the back of my mind, you know, subconsciously it sort of, maybe that was the cause of all the shit that was going on”

Troy:M:26:HD:25:-ve

“I think cause it’s easy, like if you haven’t got it you can just forget about it completely, or if you’ve got it you can start dealing with it and get over it… otherwise, yeah, you’re just waiting”

Nina:F:23:HD:23:-ve

“I spose because I didn’t want to be sitting there wondering, um, you know, sit here for 30 years and wonder whether I’m going to develop symptoms, I’d rather know that yes one day I’m going to start developing symptoms, I don’t know when that will be, but it will happen… I’m just a person that wants to know, I want to know what’s going on, I want to know what’s likely to happen, and it’s just the way I am… no matter how upset or whatever, I still thought I don’t want to sit here and just wonder, I need to know”


“I’d known everything that had happened to mum and… if there was a way to find out that I didn’t have to have those things, like ok, I hadn’t had them, but they did sound pretty bad and pretty um, worrying on the mind especially, I’ve never been very good on the mental things so, just um knowing was probably the most important thing to me, and that way I could get on with my life either way”

“The lack of being able to plan… it would have just been too frustrating, it’d have just been like well, you don’t know… we didn’t really want to risk having children and having the chance of the gene carried on, so it was like, well if we don’t know, then we can’t even think about anything like that at all, it’d just be more frustrating than knowing”


Some participants simply described themselves as having a strong desire to know:

“Yeah, I suppose for me you know, curiosity kills the cat, um, yeah, I always wanted to have it, I always wanted to have it”

Oliver: M: 24: HD: 19: +ve

“I wanted to just know then and there if I was going to grow up with the same disease as my dad or not grow up with the same disease… it wasn’t a difficult decision to make, it was just something that like yeah, I knew that I had to have it at some point in my life, the earlier the better”

Doug: M: 18: FAP: 14: -ve

“For me it was at the forefront of my mind, it was always there, and it was something that I finally said, yeah, I’m going to do this… As my mum says, once I make a decision I do it, and I just decided that I wanted to know, just for me, you know, not for anybody else around me, just for me”


“I just think it’s natural for you to want to know, if you know your mum’s got a condition you want to know if you have the condition as well”

Sally: F: 25: FAP: 17: -ve

“I’m an impulsive person… like I didn’t think of the consequences, like I just thought I had it. Obviously I didn’t have signs or anything, but, like, in my head, I just thought well, yeah, it’s a bonus if I don’t have it, cause I thought the amount of people that had it in my family, I thought that, yeah, you’d be pretty lucky to skip it”

Poppy: F: 24: HD: 17: -ve
“Just having it to know whether you’ve got the gene or you don’t, like, my gran died from it and then my dad and his two sisters all had to get their bowels removed… it’s taken what, like two generations, but it’s come down to us and you can get the test, if you have it you can control it, or you know, keep an eye on it, if you don’t have the gene then you’re clear all together kind of thing”

Amy:F:19:FAP:14:-ve

In some cases, participants felt that their ill parent had been a catalyst in their decision to be tested:

“Yeah, and our mum, like I think it does your head in when your mum’s got it”

Nina:F:23:HD:23:-ve

“My mother was dying and just about to die, and I had the predictive testing winter that year, so I spose yeah, my mother’s death sort of thing, you know, I spose my mum being extremely sick in the late stages and then passing on, I thought rightio I’ve put it off enough, let’s find out”

Oliver:M:24:HD:19:+ve

Reproductive reasons also motivated some people to seek testing:

“I met Brendan, which is Kimmie’s dad, I think I met him at 15 and then we wanted to start a family young, that’s pretty much why I got tested so young… if it had come back positive I wouldn’t have had Kimmie, definitely not, I wouldn’t have had them [kids] at all, cause I, for one I wouldn’t have wanted Kimmie, or my kid, to grow up how I grew up seeing mum, and for them not to have a mum and plus also put them at risk, yeah, so it just wouldn’t have been worth it”

Poppy:F:24:HD:17:-ve
“I just wanted to get it over and done with…from when I was 16, cause I had Charlotte when I was 16 and I know from when she was born I wanted to know…that was another thing too… I felt really guilty having her and not knowing”

Nina:F:23:HD:23:-ve

Finally, for several participants who were tested for FAP, the decision didn’t feel like theirs at all:

“Mum and dad, they decided that they wanted to just see if we had it or not…cause if we did they wanted to like do something about it early, not like wait until we were older… I don’t think it really sank in, it was more like, we’re just going to go get… an injection more sort of thing to me… I was like 11”

Liz:F:17:FAP:10:-ve

“Dad was sort of… I think back then he was going into hospital every 12 months and ah, I assume how it’s come about is that…when I got to like, I think I was 15, they said oh it’s probably time to bring Ali in now, but I never had to have the colonoscopy first because they rang up… and said there’s a new genetic testing out, both the girls can have it, that was in December of 94”


“I was 12 when I was told that I had to have the test… I didn’t want to have it, but then I sort of had to”

Harry:M:14:FAP:12:+ve

“Mum was told by the doctors I think, that we needed to get a genetic testing… that family doctor that we got was contacted and they did it that way…I can’t really remember, all I actually can remember is I think mum did sit us down and tell us we had to go for a genetic testing, it didn’t phase me”

Kylie:F:20:FAP:14:+ve
“Dad just organised it, and I just had to come in, get the um blood test”

Mark:M:21:FAP:16:+ve

“They didn’t ask us do you want to, they said you know you have to go get a blood test and they pretty much said, you know, just to see if you’ve got the gene… but they said that um, the doctors will explain it, which they did, so, yeah”

Amy:F:19:FAP:14:-ve

6.2.2 Pre-test counselling

The experience of pre-test counselling varied for young people, as did their perceptions of the value of it. For some, the hospital was not an ideal environment for counselling to take place:

“Probably not in such a big place, that was a bit, I mean for me it was big, like, everything was massive so maybe not in such a big facility… I’d probably go smaller cause it’s… yeah, cause it was like such a big, like a hospital, I mean it’s a hospital”

Kylie:F:20:FAP:14:+ve

For others, pragmatic difficulties accompanied the pre-test counselling:

“It was really hard to get in contact with the person I was dealing with, and a lot of it does come down to the money factor because I was a uni student and working a part-time job… there was the travelling down to Melbourne, having to call Melbourne, having to call mobile phones all the time, it was just a pain in the bum to me um, like it would have been easier if, if there was a 1800 number, which I know I’d always be able to contact the person on”

Zach:M:26:HD:23:-ve

For one participant, who was tested for HD at the age of 17 years, the pre-test counselling seemed minimal:
“I think I rang the association and then they put me on to the hospital and I ended up, yeah, going there, and she said oh come in for an interview and, yeah, so I went in for an interview and sat there and spoke and she said I had to do a diary and she pretty much, yeah, she did the blood test on that day… because she knew how much I wanted a family, she knew that if I was to come back positive I wouldn’t have started a family, and plus, I don’t know, because like I moved out of home at 15… I sort of had to be a bit more independent than normal 15 year olds”

Poppy:F:24:HD:17:-ve

Several participants felt that their decision to have the predictive genetic test had been made before they began pre-test counselling. For these individuals, the pre-test counselling did not form part of their decision-making process:

“It was a long time ago, um, I’m sure there would have been one or two sessions there where we just talked about whether I wanted to have it or not, um, but I mean, I was going to these meetings, so I must have wanted it you know… if I didn’t want it I wouldn’t have gone so, yeah, there was one point where I think I had about one or two meetings and I let it go for a year, and in that year I sort of thought well, can I really be bothered with this, you know, and then the next year later I called up, that was the only sort of doubt I had in having it… I couldn’t really be bothered going through it, I was sort of, you know, a bit sick of you know waiting and that, but um, yeah, I’m glad I did have it, glad I did stick with it”

Travis:M:24:HD:20:-ve

“I said look I know I have to go through all these because you know it’s a mandatory part of the testing, you have to go through the counselling, which I can understand, I have no problem with it, but I did tell her that there is nothing that will change my mind, I want it, and she continued to ask through the sessions are you sure, are you sure, are you sure and I was positive, absolutely positive”


Yet both of these individuals noted that the counselling process itself was beneficial for them:
“The counselling is good, you know, just talking about so many different things, um, sort of opens up your eyes… into what you’re going to go through”

Travis: M: 24: HD: 20: -ve

“When you have the counselling I guess it brings out anything, any problems, whether it be something to do with HD or not, cause um, most people have never had counselling of any sort, so it starts off being an HD related thing but… they talk about everything, so if there’s some sort of problem in your life… they can help you with that”


6.2.3 The day of the blood test

For many young people who were tested for FAP, the entire process entailed two visits: one where their blood was taken and they received some pre-test counselling, and one where they received their test result:

“I remember they took blood, cause I remember they had a lot of trouble getting blood from my sister, my sister’s got sunken veins, they took our blood and then spoke to us briefly… it was just, I think from what I remember, they just said to us you know, do you understand why you’re having this done and what it means, the implications if you do have it, and what it does and da da da… do you understand? And we were both like yeah, so we were sort of left at that, oh we might have spoken to her for half-an-hour at the most, kind of thing, yeah”


“They just said, you know, like, what they’re going to do if I do have it, and this and that… I think it was just, went in for the blood test, and then for the results we had to come back”

Mark: M: 21: FAP: 16: +ve
“All we remember was a bit of counselling, how do you feel, how would you feel if the result was positive, how would you feel if the result was negative, how would you feel, and they asked mum the same questions, then they took our blood and we all went home, that was it I think”

Sally:F:25:FAP:17:-ve

For some young people, the blood test had a big impact upon them, making their decision feel particularly real:

“In a way I was kind of like, whoa, this is it, yeah, it was like, this is the plunge into it, you know, you can’t go back beyond this”

Zach:M:26:HD:23:-ve

“I remember me and my cousin were sitting out when my brother was in and my cousin was just like what are you gonna do if you get it, I’m like um, I don’t know, I’ll freak out or something and he’s like yeah so will I… I was a bit quiet, like, I’m so talkative and outgoing, I’m a motor-mouth, but I was quiet”

Kylie:F:20:FAP:14:+ve

“Once you’ve sort of been pricked with this needle it sort of makes you think a bit more about it and you start to think well if dad’s got this condition and he could pass it on to any of us and it’s a 50% chance that I could have it, well the odds are pretty sort of likely in a way. So you start to think you know, I’ve got it, I might have to do something about this, and I don’t know, it just makes you start to think about all these things”

Emily:F:21:FAP:14:-ve

Yet for others, the blood test was simply a procedure, with little emotional impact:

“I’ve had quite a few blood tests, but the actual blood test itself was nothing for me… I mean the taking of the blood itself is not a result so, it was just a medical procedure I guess for me, I mean, it was the session when she called me in when they had the results that was obviously the emotional one”

“I do remember I wasn’t feeling that nervous, it was more actually waiting afterwards which was the more worrying part”


One participant felt that her age had contributed to the level of impact the blood test had, believing that she was too young to understand what was happening:

“It wasn’t strange to us I think cause we were so little, or you know, a bit younger, um, mum and dad weren’t overly really concerned about it at the time so we weren’t all that worried about it either, so it was sort of no big deal”

Emily:F:21:FAP:14:-ve

6.2.4 Waiting

The waiting time between having the blood test and receiving the test result was not difficult for some young people:

“We just forgot about it for a while and then got a letter or a phone-call or something”

Harry:M:14:FAP:12:+ve

“I didn’t really think about it after I had it done, but as we were waiting, you know, cause I’d hear my dad say to my mum oh I wonder when we’re going to get a call and I wonder when everything’s going to come back and so I sort of heard them keep talking about it… it crossed my mind and I’d think about it a little bit but honestly at 15 I was just such a socialite at that age, I was always out, I was always going to the movies, I wasn’t concerned with anything else to be honest”


“I wasn’t worried or anything, I completely forgot about it, yeah, I just completely forget about everything, so, like, if it’s nothing that I have to worry about it’s just, it’s just in the back of my head”

Doug:M:18:FAP:14:-ve
Other young people found the waiting time challenging:

“I think it was about, yeah, it was about 5 or 6 weeks and it was really tense… I didn’t eat much, I wasn’t as happy as I was, um, yeah, I was just, very snappy, very moody and everyone understood, everyone understood why, so yeah, that’s the sort of person I was, just snappy and moody”

Travis: M: 24: HD: 20: -ve

“I remember me and Emily were convinced that we’d have it and Debbie didn’t really say much about it, yeah…I think it was just kind of like, I don’t know, so that if we did it wasn’t such a shock, like, yeah, I don’t know”

Liz: F: 17: FAP: 10: -ve

“I do remember it was quite a worrying time and um, I was quite nervous waiting for the results, there wasn’t much I could do about it… just have to grin and bear it”


Some young people used distractions during this waiting period:

“I know that I got stuck into work heaps, yeah, like I was doing like 15 hour days and, crazy, but like, it was nothing, yeah, so I did heaps and heaps of work just to keep my mind off it, then I’d bring work home to do on the weekend, so I pretty much just chucked myself into work, it was awful and all that to wait and it just felt like forever”

Poppy: F: 24: HD: 17: -ve

“I was a bit worried, nervous, I tried to forget about it, thinking about other things”

Harry: M: 14: FAP: 12: +ve

For two individuals, the waiting period was extended because they were told that they had to stop using drugs before they were allowed to learn of their test results:
“I wasn’t allowed to find out while I was on drugs, that’s probably why I put it off for a few years… I rang her up and told her and she said that I wasn’t allowed to get my results until I was off drugs… I don’t think it was very fair that they just said no you’ve got to get off drugs before you can get your result, I don’t think that was fair”

Nina:F:23:HD:23:-ve

“I got onto heroin, they weren’t going to give me my result if I was still on heroin, they’d taken my blood and everything by um, I had to quit heroin to get the result… I don’t see how that makes me less of a human being and less of having the right to know”

Ella:F:20:HD:18:-ve

6.2.5 The day of the results

On the day that young people were due to receive their test results there were a range of emotions occurring. Some young people felt extremely anxious on the day their results were to be disclosed:

“We were all just really nervous, yeah, we were all just sitting around the waiting room and really kind of anxious just to find out…I think I was just more worried then that it might actually come through positive and then it would be more a reality… yeah, it was more serious than what I actually thought”

Liz:F:17:FAP:10:-ve

“We were shitting ourselves, you know, you don’t know what it’s going to be like”

Ella:F:20:HD:18:-ve

“I think from the point where I got into the waiting room until I saw [the counsellor], for the first time that day, I was a bit sort of, don’t come near me, you know… but yeah, all in all I was ok, and I don’t know why, I mean, I don’t know why I wasn’t shaking or I don’t know why I wasn’t feeling sick, but, yeah, I remember I was tense, but that it, I wasn’t shaking”

Travis:M:24:HD:20:-ve
“I was a bit nervous, cause um, oh cause you know, being young and that, like I was a bit nervous”

Amy:F:19:FAP:14:-ve

Some young people were certain that they would receive a gene-positive test result:

“[We] got a phone-call saying the results were in and I think we went and made an appointment, and all 5 of us went down there and we found out individually, we didn’t have any family sort of meeting, it was all individual, I think they chose to do it that way, I can’t remember…I walked into that room thinking yeah I’ve got this… we were just going to deal with this together and it was going to be one of those things”

Emily:F:21:FAP:14:-ve

“I was nervous but I think I worked in the morning, and then yeah, Brendan’s parents drove us up and yeah, I was really really nervous like sitting in there waiting and I was pretty much, like on the day I was pretty much waiting for her to turn around and go well yeah, you do have it”

Poppy:F:24:HD:17:-ve

For others, the day of disclosure was less of a worrying time:

“I was at school that morning”

Doug:M:18:FAP:14:-ve

“I was just you know, I wasn’t really nervous, just more, you just cross your fingers and hope you don’t have it but if you do, you know, there’s nothing you can do about it anyway”

Mark:M:21:FAP:16:+ve

“I think I just sort of tried to blank it all out, not try to think about it, and I just thought yeah, when I get the result, you know, what happens happens so, you know I didn’t really think too much about it”

Troy:M:26:HD:25:-ve
One young person who was tested for FAP, had no such anticipation on the day she received her test result, as her results were disclosed over the telephone with no warning:

“I wasn’t at home, my best friend lives across the road and I was at her house and it was a week day and my sister come over and she said oh I think you need to come home, and I said, what for, and she said oh the lady from the hospital is on the phone and I haven’t got the gene but they can’t tell me if you’ve got it, they want to talk to you. I said oh, ok, well that’s good, you know, that she didn’t have it, I didn’t think she’d have it anyway, I don’t know why, I mean, it’s stupid, but she looks so much like my mum and she’s just like my mum and I knew I would have it, I knew, because, I know it’s got nothing to do with it, but everyone’s always said from the time I was born that I was exactly like my father, I look like him, I talk like him, I’m outgoing like he is, all that kind of stuff, so I knew, I knew in my heart I swear, like I know there’s a 50-50 chance, but I knew I’d have it, and I knew my sister wouldn’t. I was a bit shocked that they told us over the phone… I think it’s different if neither of us had it, I think it’s fine to say, da da da da da, you don’t have it, but I think when one or both had it, they probably should have said, oh the results are in, we’d like you to come down and we’ll discuss it with you… I don’t think they should have just said that over the phone.”


For another young person, anticipation was the key, with a specific plan of action determined for the day of disclosure:
“We’d prepared it, so that if it was bad news, we were going to go for a, you know, a country drive or a country holiday, so we had done a lot of sort of preparing at work and that to ah, you know, be ready to dispatch straight away and just leave Melbourne, just to have a clear mind I spose. I wanted to, I’m a very strange person, but I wanted to have a lot of the initial impact in areas outside Melbourne I spose, which is sort of strange, I wanted to ah, I wanted to grieve in an area that I could leave behind and then come back to happy Melbourne if you like, I spose it’s a bit strange, but, yeah, so um, I ah had about one hour’s sleep the ah night before then and, yeah, I always sleep through anything to be honest, but that’s one of the most sleepless nights I’ve had…I knew it was going to change my life one way or the other”

Oliver:M:24:HD:19:+ve

Some individuals felt that this day was made much easier by the support that they had:

“They were pretty nerve racking, had a lot of friends and stuff SMSing like good luck and stuff like that, so a lot of friends around us knew”

Zach:M:26:HD:23:-ve

“I was sort of um very nervous and yeah, it was a bit easier for me cause my grandparents were there”

Harry:M:14:FAP:12:+ve

Some young people tried to guess their test result before actually receiving it, simply from the expression on the face of their counsellor when they were first greeted:
“The morning before I went in I was alright, I remember, I was ok, I got up and did what I normally do, um, I was talkative on the way up and that, but once, like when I got into the like, the waiting room, I was sitting down and I was just, I wasn’t shaking but I was scared, you know, and I saw [counsellor] walk past and she, she’s come up to me and she’s gone oh “hi, how you doing” and I’ve gone, oh I can’t remember what I said to her, I’ve gone yeah alright or something like that, you know when you’re not alright and you say yeah, I’m alright, you say it anyway. I think I remember saying to her “do you know what the result is?” and she’s gone “I’m not going to say anything to you yet”, but she’s gone “it’s very interesting” …but she said it with a smile on her face, so I knew from then that I was ok, but you know, what’s this interesting part sort of thing”

Travis:M:24:HD:20:-ve

“Yeah, I mean, by nature I spose, you know, I try to pick up subtleties in people or messages from people or messages from body language, I spose that is sort of my job and we sort of had a seat there and we saw [counsellor] there and instead of smiling and saying “hello Oliver and Max, how are you”, she said “Oh, Oliver” and sort of had a very ah, very ah, unhappy look on her face. Straight away we both simultaneously went oh-oh, you know, people are looking for the signals from the word go, it’s just human nature, you know”

Oliver:M:24:HD:19:+ve

Some individuals felt as though it took a particularly long time for the result to actually be disclosed, once in the meeting with their counsellor:
“Mum came with me which was good, I just remember feeling, I spose a little worried, you know, you try not to and everything but we all worry about some things sometimes, so, [counsellor] asked again if I was sure that I wanted it, wanted to know the results, and she went through what a positive result meant and what a negative result meant and what a grey area meant, continuously asking if I was ready kind of thing, which is fine, you know, but at the time I sort of got a bit frustrated, cause I just, I wanted to know, but I know she’s got to do things, she’s got to go through certain protocols and everything like that, but yeah, I just wanted to know, I just wanted it to be over and done with and wanted to know what the result was”


“They didn’t tell us straight off is what I remember, we were sitting there going what’s the result and they were still asking us how do you feel and we’re like oh god, come on”

Sally:F:25:FAP:17:-ve

One young person would have liked her results to be disclosed by her parents, instead of the counsellor:

“I reckon it probably would have been better if mum and dad could have found out first and then they could have told us, because yeah, that’s what they wanted to do and they couldn’t. I think that probably they would have been able to tell us in a better kind of way, instead of finding out from a stranger, that probably would have been better… and then we could have asked more questions and stuff, cause if you’re with a stranger you kind of feel more uncomfortable and don’t really want to be asking all these weird questions and stuff”

Liz:F:17:FAP:10:-ve
6.2.6 Immediate impact of the result

In conveying some of the immediate impacts that the receipt of test results had for young people, I divide their descriptions into those relating to gene-negative test results and those relating to gene-positive test results.

6.2.6.1 Gene-negative test results

For some young people who received a gene-negative test result, the experience was an ecstatic one:

“Yeah, and I mean, I was all buzzed up from the negative result, so I really wasn’t listening too much to what was going on, just that I remember kissing [the doctor]… You can’t explain it, just relief, relief. The first thing that enters your head when you get that negative result is not fantastic, you know, awesome I’m not going to get it but it’s like oh my god you know, thank god it’s over with, and you’ve gone this far, and to come this far… A negative result, it’s fantastic, you know, it’s, I don’t know, it’s weird, it’s, yeah, it’s, I don’t know, I don’t know how to explain it”

**Travis:** M: 24: HD: 20: -ve

“I cried… I told her, cause I knew I didn’t, and I told her my sister hasn’t got it either and yeah, I was just crying, and I thought about Charlotte, my daughter, that was the first thing that made me cry”

**Nina:** F: 23: HD: 23: -ve

“I just grabbed her and hugged her and gave her a kiss and stuff, so it was, yeah, oh it was just grouse, she was saying all this stuff and I was just, oh, I couldn’t believe it, then me dad just started crying and that… yeah, it’s just grouse, sort of, amazing that you know, yeah, I just, I sort of tried to blank it out and then all of a sudden you find out you don’t have it, it’s just grouse, it’s so good”

**Troy:** M: 26: HD: 25: -ve

“It’s pretty much like a new life that you’ve been given in a way”

**Poppy:** F: 24: HD: 17: -ve
“I went in by myself and um the lady told me that I was negative and I was just over the moon… that just means that um this part of the family tree is then cut off from any further… it’s good to just sort of be cut off from going any further, it’s just a relief”

Emily:F:21:FAP:14:-ve

“Me and Reece went in there and she came in and she sort of had a smile on her face already and yeah, she just said you don’t have the gene… so excited, I screamed and mum and max didn’t know if it was a scream of happiness or a scream of devastation, but um, yeah, it was great and I was just dying for this day all my life and it finally arrived”

Ella:F:20:HD:18:-ve

For some, the initial reaction was one of shock, with the result taking some time to sink in properly:

“I was in shock in a way, and I think I would have been to any result, I would have been in shock, it was like, oh, there was a little bit of relief”

Zach:M:26:HD:23:-ve

“Even to this day, it’s still sinking in, yeah, it hasn’t really sunken in, yeah, it’s just, yeah, I’ve just given myself up to live a life now, you know, get rid of this old thought that’s just been in my head all my life and, I mean, it’s fantastic, it is, it’s fantastic, I mean, I can’t be anything but happy about it, but yeah, it is very weird… I still haven’t, I’m in another desert you know, lost (yeah), but it’s good, I mean, it’s sort of a good thing I guess, of course, yeah”

Ella:F:20:HD:18:-ve

“Brendan’s parents are bawling, Brendan’s crying and I’m just sitting there saying oh, that’s good, like I didn’t feel anything. It wasn’t until I got home that I’m like oh ok, that it really sunk in, it didn’t sink in while I was there… like it took, yeah, sort of later on that night I thought, oh, yeah, it took about a week I reckon, to fully sink in that you know, I could have a family and it’s not gonna yeah, matter”

Poppy:F:24:HD:17:-ve
Some young people felt quite lost after the receipt of a gene-negative result:

“This may sound weird but it was almost a bit of, almost a little bit of a let down as well… it was kind of like oh, you almost, you’ve almost, you, you’ve built yourself up for something and then it just doesn’t happen, it’s a bit, yeah, it’s like oh, yeah”

Zach: M: 26: HD: 23:-ve

“I really don’t know what to do with myself, I feel really really lost and I’ve got this life to live, you know… I’m truly clueless, I really feel like a newborn child, you know, like, really clueless with what to do.. I remember going down and seeing [doctor] and he said you know, if you’ve got the gene, you know, blah blah blah, if you don’t that’s fantastic, that’s it, it’s over, and I thought really? is it over?”

Ella: F: 20: HD: 18:-ve

Some felt particularly worried about their siblings:

“Scared, because I found that Debbie had it, so yeah, cause I was more worried that she had it, like it was a big shock…um, I think I sort of felt a bit guilty because she ended up having it and I didn’t, like, it was just because she was the one who ended up having the gene and yeah, Emily and I were so convinced that we did and then we didn’t, it was a bit weird”

Liz: F: 17: FAP: 10:-ve

“At the same time, I was in a different mind, I was thinking is it gonna, cause it was 50-50 whether I got it, I was thinking, because I didn’t have it, is it more chance [brother] was going to get it and stuff like that so, yeah”

Poppy: F: 24: HD: 17:-ve

I asked participants to compare the feeling of receiving a gene-negative result to something else that had happened in their life. Some felt that there was nothing they could compare it to:
“It’s kind of like an out of the ordinary kind of situation, it’s definitely not the kind of thing you come across every day, so yeah”

Liz:F:17:FAP:10:-ve

“Totally nothing like when I got married, um, the marriage was just so much more important to me than, than the result... yeah, I can’t really think of any, like, that’s really really hard”

Zach:M:26:HD:23:-ve

“You can’t compare something like that because I mean, I don’t know, I’m only young, I’m 23, and I think doing this test was the biggest thing I’ve ever had to do thus far, so I think I haven’t really lived yet, um, I mean, I could have a baby who, who’s not well when it’s born you know, and have to go through a lot longer than what I did with a predictive test to know that the baby’s going to be ok, so I really can’t sort of... I can tell you now that the best thing that’s ever happened to me up to now is getting this result”

Travis:M:24:HD:20:-ve

“I couldn’t compare it to anything else”

Troy:M:26:HD:25:-ve

“I don’t think I could compare it to anything... I cannot compare it to like getting a new dog or anything like that cause it’s a totally different feeling cause this is like a major thing in your life, or it could become a very major thing in your life and I was, I mean, it was just the end of the line for me, I didn’t have to worry about it any more, like personally anyway, but yeah, no, nothing”

Emily:F:21:FAP:14:-ve

However, others were able to choose a specific incident in their life to compare the feeling to:
“I guess the day mum had me, the day I was born, it’s a miracle day... like [doctor] rang me up the other day and he’s like oh I wonder if you remember me... of course I remember you, you took my blood, you know, like, these are days that are going to live in my mind forever, the days I dreamt about, the days I’ve waited and waited and waited for, like, I will never, ever forget them... getting married, honestly, having a baby, yeah, it’s one of those big defining moments of my life, yeah, I’ll never forget them”

Ella:F:20:HD:18:-ve

“Well, that’s up there next to having my kid, my daughter... yeah, cause I don’t have it, so, yeah, it’s a good thing, it’s like a big present”

Nina:F:23:HD:23:-ve

“I don’t know if I could even explain it, like the relief I felt... unless you’ve given birth... yeah, once you’ve given birth and you know you don’t have to go through that for obviously a few months, that’s how big it is”

Poppy:F:24:HD:17:-ve

For several young people, passing their Victorian Certificate of Education (VCE) in their final year of highschool provided an appropriate comparison:

“I’d compare it to passing my VCE, it was good, yeah, it was good knowing that I didn’t have this disease”

Doug:M:18:FAP:14:-ve

“Oh, passing my VCE, yeah, because um, in comparison, because none of my family’s ever, like even young cousins and that, done their VCE and me to like, you know, oh I hope I can do my VCE, and I finally did it, so it’s like oh, I did it!”

Amy:F:19:FAP:14:-ve

“I don’t know, just getting results I guess, getting examination results, I wouldn’t compare it to getting my VCE results but probably just getting an exam result, a really good one, a positive one”

Sally:F:25:FAP:17:-ve
6.2.6.2 Gene-positive test results

For some young people receiving a gene-positive test result, the experience was a traumatic one:

“If I didn’t have it, I’d be much happier, and more, what’s the word, um, less worried about things, and like now cause I’m worried about all my health and that, like, a tumour or something, like that’s really what worries me”

Harry:M:14:FAP:12:+ve

“Oh you know, [I thought] I’m done in for I spose, you know, hitting the ah, hitting the wall now, you know, in a lot of trouble, mm”

Oliver:M:24:HD:19:+ve

“We went and saw the doctor and then he was telling my brother, he goes now one of you have got it and I’m like oh great… he just said outright one of us has got it, but we didn’t know which one, but I knew it was me, I just had that feeling it was going to be me… but I didn’t believe it until he actually said it and when he said it I was crying and oh I was so upset… I remember on the way home I just wouldn’t talk, I wouldn’t talk at all, I just zipped my lips and I just cried”

Kylie:F:20:FAP:14:+ve

“I was actually very calm at first, straight after um, it was actually at the um doctors request… she asked if I’d like to just walk around by myself for a couple of minutes just, you know, in the walk way, just to calm myself down. I actually got a bit angry, and then ah, once I calmed down I kind of went back and walked back in but um, I don’t think I handled it too badly, of course it was still a shock, so um, I never got emotional about it, that’s one thing I remember, um, shaky, ah, ok, very down, depressed, um, but that was probably the least of it, so I think I handled it pretty well and I guess that’s just, maybe that just came back to um, the information that my mum had given me, I’d been reassured all the way through”

Some young people spoke about the reaction of their parents, and the impact this had upon them:

“My mum and dad went in first and they were told, then I just sat out with my grandparents and they asked me to go in… I could tell that something was bad cause my mum was crying… I just thought, I’ll probably be stuck with this for the rest of my life… yeah, and I am… I just try to ignore it”

Harry: M: 14: FAP: 12: +ve

“I wasn’t, I wouldn’t say I was shocked, because like I said, I expected it, I really did, I sort of prepared myself a bit for that anyway, um, above everything else, I was hurt when I saw what it did to my parents”


For some, the gene-positive test result took a while to sink in properly:

“It took a very long time, quite a number of hours, so we drove from Melbourne to [town] and stayed there and ah, yeah, the drive was ok but then we were sort of crying a little bit and what not, and we drove from [town] to [town] in one day and then, yeah, in [town] it sort of sunk in even more I suppose”

Oliver: M: 24: HD: 19: +ve

For others, it was difficult to comprehend that something was wrong when no symptoms were evident:

“It was just a bit weird thinking that you have to go in for something if you’re fine anyway and that, in the back of your mind you’ve got something wrong with you, you know what I mean, it’s a little bit weird, but you kind of forget about it”

Mark: M: 21: FAP: 16: +ve

Some young people found themselves asking “why me” upon receipt of their gene-positive test result:
“At first, I thought, why me and not him…and it’s not fair, but then, um, I
don’t know, I just sort of accepted it”

**Kylie:F:20:FAP:14:+ve**

“I was basically thinking why me I think, more than anything, why did this
happen to me, ah, what did I do, um I don’t really know what, I can’t really
think much else about it but… the feeling I can remember, what I was
actually thinking, that’s the hard part”


Others tried not to think about their gene-positive test result too much:

“I had the kind of personality where I thought, well, I can’t worry about
what’s to be in the future, like, I’m only 15 years old, you know, that was
my attitude, I’ll just, I’ll worry about the here and now… well here and now
I’ve got it, at least I know, even if my sister doesn’t kind of thing, at least
my parents know and don’t worry about it, it wasn’t nearly as bad as like,
yeah, finding out dad, a couple of years ago, had the cancer”


One young person felt quite awkward at the time of receiving his test result and wishes
now that he didn’t know his genetic status:

“I was like, I hope I don’t have it now, you know what I mean, and it was a
bit awkward when I went in the room and they told me and then I went back
and they said you know, you’ve got it. Dad was trying to be like one of
those dads off the TV, you know, oh it’s all right son, like, you know what I
mean?…I would rather of not ever known, you know what I mean, cause
it’s always in the back of your mind, but like, if I hadn’t have known, well,
what you don’t know won’t hurt you, you know what I mean?”

**Mark:M:21:FAP:16:+ve**

However, for another young person, knowledge of her gene-positive status brought
relief:
“Relief… I knew … just as simple as that… that was the result, there was no grey area. I don’t know what I would have done if it was a grey area, cause I just wanted to know, positive or negative, and I just felt relief because I knew, and I had some sense of what was going to happen to me later in my life. I’ve always tried to stay a positive person and going through this testing and knowing that I have the HD gene has sort of increased that, and, ok, it’s over, positive, get on with my life, cause I had my answer, so, yes it was emotional, um, I can’t remember if I cried or not in there, I know I did later that day, um, I took the whole day off from work, and I know I did later in the day, so, cause I guess it’s sort of, you know, the relief, ok, it’s over, and then later the body just falls into a heap, which happens to everyone”


I also asked participants to compare the feeling of receiving a gene-positive test result to something else that had happened in their life. Some felt that there was nothing they could compare it to:

“Nothing, cause that was like um one of the worst days of my life, finding out that I had that, because it was something that I have for life and could kill me, and yeah, something I can’t get rid of… nothing else could really compare to that”

Harry:M:14:FAP:12:+ve

“I don’t think you could really compare it really cause, I don’t know, it’s not the same feeling, anytime it’s just like a weird feeling cause it’s not, you’re obviously not happy, but you’re not angry… you’re just confused kind of thing, something like that, yeah, a bit pissed off… you don’t really want to show emotion”

Mark:M:21:FAP:16:+ve

“I can’t say like it was as bad as when I found out dad had cancer or anything like that, cause that was much worse cause I understood, do you know what I mean? Probably nothing can compare to it”

Others were able to find a comparable experience:

“It was sort of the same thing knowing, like, we had to put our dog down, and it was sort of knowing that the day was coming… and the day was gonna come and then when it did come and then when he went and then when he came back, I mean, we knew he was gonna die, he had to be put down, he was so old, and then seeing him actually dead, and then we had to bury him and I was so upset… sort of like that, you know you know you know and you wait wait wait and then, for my brother, I mean, he didn’t have to worry about it, but for me I kind of did”

**Kylie:** F:20; FAP:14; +ve

“The two things that really almost killed me in my life, were the death of my mother and the death of a close friend 10 months before… yeah it was bad, but not the worst I don’t think, I mean, you know, we all don’t know what’s around the corner”

**Oliver:** M:24; HD:19; +ve

6.2.7 Long-term impact of the result

Many of the impacts described by young people relate less to the time acutely after their results were disclosed and more to the broader, long-term impacts of testing. For some young people, learning of their genetic status has appeared to help them change some of their behaviours:

“I’ve gotten off drugs since I found out…I don’t know if subconsciously it was or what, but, yeah”

**Nina:** F:23; HD:23; -ve

“Since I’ve been tested I’ve been pretty good… I haven’t been in trouble with the police or anything so…I seem to have changed a bit, just come out of me shell a bit more and… you know, a bit happier and stuff…now I want to um, start my own business, so it clears away, you know, for the next few years, now I’m not going to get symptoms in my 30s or 40s or whatever”

**Troy:** M:26; HD:25; -ve
Some individuals feel that it’s brought them closer to friends:

“In a sense it kind of brought us closer because my best friend, she has diabetes and now that I’ve got this, we can sort of always, you know, I’ve got something and she’s got something and so… yeah we can talk to each other about it and at school that really helps”

**Kylie:F:20:FAP:14:+ve**

“Oh there’s actually one girl, I never knew her kind of thing, but she went away, like didn’t play hockey for a while and um she had an operation and apparently everyone was saying oh she’s had an abortion or something but I found out later she had the same operation that dad had, yeah, so now I’ve started talking to her and we’re really good friends”

**Mark:M:21:FAP:16:+ve**

While others felt that testing distanced them from their friends:

“I didn’t get as much support as I, well maybe not wanted but more needed than anything, from my peers… but the teachers and the staff there, they gave me quite a lot of support for what I needed, so that part was good”


“Yes it did kind of distance it a bit I guess just because there was something I had that no-one else had and I just couldn’t relate to it, so in that aspect yes, it did distance it a bit”


Many young people noted that going through testing has resulted in closer relationships with members of their family:

“I think if anything it probably made us closer cause he talked about it, like more openly and stuff cause we were more aware of it, yeah”

**Liz:F:17:FAP:10:-ve**
“I’d say it’s brought us closer together… just because I think, like I’m not being rude to my sister or anything like that, but she would never have coped with it the way I did, or got through it the way I did. She would have fallen apart, and she’d be the first one to tell you that. She’s just not strong that way, um and I think because my dad’s had the surgery, my dad’s been told and stuff like that, and then for the same thing to happen to me, I think that you create a bond, you know what I mean, I was always sort of close to my dad anyway, obviously, but um, my dad was the one who was sitting there, you know, when I had all these complications after the surgery and you know, he was the one who was sleeping in a hospital chair beside my bed for me to have someone there. I think when you go through nights and days like that, over a period of time, you do just become a lot closer and, you know, I ended up probably being able to talk to my dad about it more openly and honestly, you know, because I knew whatever I’d say he’d understand, not that I didn’t think my mother or my sister would, especially my mum, but you just sort of, we can relate to each other a bit more, the things we’re saying… we were close before, but I would definitely say that we share like a special bond and we’re closer now than we were before then”


“In hindsight it’s improved the relationship between my father and I actually, cause, you know, my mum died in 2000 as you know and whether my dad likes it or not, he’s had to take on a lot of roles that she used to do, I mean, a bloke’s a bloke, but, you know, yeah, he’s ah taken on a lot more with both of us, yeah, I talk to him about HD far far far more than ever”

Oliver:M:24:HD:19:+ve

“Dad was, you know, very, very supportive and that and because dad was supportive, you know, he’s not usually sort of supportive of me when I do things so you know, I thought well you know this is a good chance for me and dad to get really close as well… sort of felt like I was abusing that privilege but, um… we got a lot closer, we got a hell of a lot closer, and it’s only because of that, that he was one of my support partners and he was there, you know, pretty much every, I mean, he took time off work to come out with me and that, yeah, lots closer together”

Travis:M:24:HD:20:-ve
“Yeah, it makes it sink in a bit more, like, deeper, I don’t think it’s actually sunk in what mum’s going through at the moment and that, it’s good seeing her and knowing that I’m not going to have to go through it too, I don’t think I could have helped her as much if I knew I had it”

Nina:F:23:HD:23:-ve

“It may have made my mum and I a bit closer because she went through that process with me, and she was my confidante, um, so I think it probably made us closer, definitely”


“My mum and I talk a lot more, with this FAP thing we can actually relate, which is good”

Kylie:F:20:FAP:14:+ve

One individual noted that she felt her relationship with her mother had eased somewhat, perhaps because her mother was not so worried any more:

“So she sort of let me go off and do what I’ve gotta do, you know, in my own way, and she knows that’s the sort of way I’ve got to do my thing, like, she can try and drill things into my head over and over again but she knows sort of, she’s free to do what she wants now and I don’t have to sort of be her carer for life any more, so yeah, if anything we’ve drifted apart but I mean, our relationship, our friendship’s still exactly the same, it’s just um, we’ve drifted apart a bit”

Ella:F:20:HD:18:-ve

Another young person felt that her mother was now jealous of her, for receiving a gene-negative test result:

“Yeah, well, me and my mum have never had a good relationship, but I don’t know, I think she’s jealous, yeah, she’s said it to me a couple of times, she’s said “oh at least you don’t have to have this shit, you’re lucky” and all that sort of stuff”

Nina:F:23:HD:23:-ve
Several young people also spoke about their relationships with their siblings specifically:

“How my brother does not want to get tested, and I respect that, I think it takes a stronger person to keep going on with life and not want to know, at first I thought, you know, why don’t you want to know, don’t you want to know!, and I slowly, through the counselling and that, I slowly learnt to respect his decision. It takes an extremely strong person to do that, to put it aside, don’t care if they’ve got it or not, and keep going, so I strongly respect that of my brother, but I think… I do want him to know, like, what if there’s treatment you know? I mean, there is a bit these days, of treatment, but you know, I just don’t know what it’d be like, watching my father go through it, and to see my brother go through it, it’d be extremely hard, I always wish if it’s gotta be one of us, please let it be me, cause I can’t watch another person go through it, it’s really hard to watch, yeah”

_Ella:F:20:HD:18:-ve_

“When I told him my result was positive he gave me a hug… (laugh)… it’s very rare for him”


“That first day that we were told, like we didn’t know how to act, like even you could just tell, cause it’s like do you, you know, would he think that we’re feeling sorry for him if we keep, oh, you know, just that first bit, but then after it, I don’t know, in a day or two, it was just back to normal life kind of thing”

_Amy:F:19:FAP:14:-ve_

Several young people noted ways in which finding out their genetic status has altered their plans for the future:
“[We] just starting talking more in depth about um having kids and just our future really, just our future as girlfriend or husband and wife, whatever it was going to be, um, but I think we just talked about the future a lot more, because I always had this, this, thing in my head that I don’t want to plan things while not knowing cause when you plan something and something doesn’t go to plan you get, you know, you get ah, disappointed, so, um, that’s what sort of changed in our relationship, we just started talking about the future a lot more”

Travis:M:24:HD:20:-ve

“I was thinking of going into the airforce but I can’t do that now… I found out that you can’t have any medical disease or anything cause it could interfere…any of the military forces, that’s scratched… I didn’t really know what I wanted to do for a while after that, but, only last month or so I’ve had ideas on a sports future… I’ve been thinking about being a sports teacher”

Harry:M:14:FAP:12:+ve

“We didn’t want to have children if they had the gene, ah, cause we didn’t want to pass it on…but, yeah, now we do”

Zach:M:26:HD:23:-ve

“It’s changed my opinion on money completely, the bottom line is, money can’t buy happiness… it can assist and help you along, but it does not buy happiness and it does not change someone’s status in a disease sense, and that’s the bottom line, so if you go and spend ah $260 on a hotel, you know, the receptionist isn’t going to come down in the morning and say things have changed, it’s not the way the cookie crumbles. I’ve always travelled beyond my financial means for the past 7 years I suppose, and get ah, just light debts, just typical silly males in their 20’s and ah, yeah, but I’ve sort of got really really funny with my money lately, you know, like oh I’m not going to waste money here, it’s not going to bring a friend by my side, it’s not going to bring, you know, it’s not going to bring, it’s just not going to change your situation, you know, I spose ah, marketing and what not can put someone under the illusion that it’ll change your situation and reverse things, but it’s not the case”

Oliver:M:24:HD:19:+ve
“Oh, we’re engaged now! I think it’s incredible, um, like, the fact that I met him and he was willing to stay with me no matter the outcome, and now the outcome’s come, you know, and it’s all good, you know…yeah, I think we’ve got a wonderful life ahead of us”

Ella:F:20:HD:18:-ve

“I can sort of um, plan for the future a bit more and sort of, I don’t know, yeah… you know I might end up with cancer or something, but this is just something that I know I’m definitely not going to get and it, it sort of helps”

Troy:M:26:HD:25:-ve

“I wanted to join the army and I knew that if I wanted to go in full time I think it was yeah, 4 years, but um, sometimes having what I’ve got affects me physically cause like I get sick sometimes if I eat the wrong food and I know cause I used to do army cadets and we used to go out bush a lot and um, I just, it was a bit hard then and I thought well maybe it would be best if I didn’t”

Kylie:F:20:FAP:14:+ve

“It reaffirmed my decision not to have children…when it came back positive I thought well, no, that reaffirms that because I don’t want to pass it on, um, and again everybody’s different, so, I mean some people will think well you can have kids, it won’t affect them until they’re 50, 60, 70 years old, they’ve got all that time to live before it affects them, which is fine, but I don’t want to pass it on, I want it to stop with me, on my side”


For one young person, simply the process of being tested seemed to impact greatly upon him, irrespective of the test result he received:
“I thought well, if I can go through this whole process of getting tested for this thing, I can pretty much do anything, except for quit smoking, so I respected myself a lot more for that, and um, I just started enjoying life a hell of a lot more, I started going out to clubs and stuff and you know, just doing what a normal person of that age would do… I didn’t go out much before that, I was a bit of a home body, just staying at home, watch videos, and that was it really, so yeah my social life really picked up”

**Travis:** M: 24: HD: 20: -ve

For another young person, having the test allowed her to begin enjoying life more. Prior to testing she felt that the uncertainty about her genetic status had stopped her from living effectively:

“...I was thinking God, 40 years from now I could be shaking and doing all this and that and I’m going to be dead sort of 20 years after that sort of thing and it stopped me from living at the time effectively, I mean, I did the day to day things, I went to school I did my homework I ate dinner, ate breakfast, did the things I needed to do, but didn’t really do much else, um, didn’t feel like I was living kind of thing. I had a life but I wasn’t living because I was just thinking about things that were too far ahead… Once I had the knowledge, that was it, ok, fine, you know, I can’t do anything about it, and I realised that I guess, maybe that was part of it. I’m thinking ok, well I’m not in control of my life, but then you’ve got to realise that you’re not in control of your life (laugh), you can control certain aspects, you can control your decisions, but you can’t actually control your life, you know, so and I guess that’s part of it. I realised that and you just accept that if you can, I mean, everyone’s different, but I just accepted that and thought well I’ll get on with controlling my decisions that will affect my life and do what I want, so, it makes sense you know”

**Belinda:** F: 25: HD: 21: +ve

6.2.8 The best and worst aspects of the process

When interviewing young people about their experiences of predictive genetic testing I asked them to identify the best and worst aspects of the entire testing process.
6.2.8.1 The best

Some individuals had difficulty articulating the best part of their predictive genetic testing process:

“I don’t think there is one. Eating Maccas after you get out when you haven’t eaten in a day and a half or something, you haven’t eaten in a day and you go downstairs and there’s a Maccas downstairs so you eat for the first time”

Mark:M:21:FAP:16:+ve

“Maccas… every week we got Maccas”


While others found the identification of the best part a relatively easy task:

“Finding out that I had it so there could be something done”

Harry:M:14:FAP:12:+ve

“Maybe just kind of bringing the family closer cause they were talking about it and being open about it and stuff”

Emily:F:21:FAP:14:-ve

“Probably putting everyone’s mind at rest I would say, because we all knew, we just all knew, but it was probably knowing, that was probably the best thing, just to put everyone’s mind at rest, like, mum and dad waited god what, 18 years, 19 years to find out for my sister and 15 to find out for me, like that is a huge time. I don’t think I’ll cope waiting that long to know if Natalie’s got it…so I think that was the best thing, definitely, just putting it to rest, well, not putting it to rest, but, yeah”


“I spose it gives more freedom, being able to plan a life”

Zach:M:26:HD:23:-ve
“Instead of umming and aahing, am I going to be 60, am I going to be 40, it made a completely decisive answer to it, all the anxieties floated away, they just disappeared, off they went, rightio, this is it, it will start obviously shortly, deal with it and adjust”

Oliver:M:24:HD:19:+ve

“Finding out the result, just the result, I don’t care if it was positive or negative, but actually finding out a result and directing my life, what that result can lead to, yeah, that was the best thing, like, direction of life, a final, you know, ok, this is the direction I can take, you know, never before, I felt like I was at this, you know, a T and straight, you know, turn left, turn right, you know, and I had no direction what so ever, but now I feel like I got the reigns, you know, I can direct my life, before I had no idea what to do”

Ella:F:20:HD:18:-ve

“Life’s better”

Travis:M:24:HD:20:-ve

“Finding out I don’t have it…finding out my daughter’s not going to have to go through it either”

Nina:F:23:HD:23:-ve

“Just the final result, yeah”

Troy:M:26:HD:25:-ve

“Probably just the relief… what can happen in the future, like you can sort of set yourself up more”

Poppy:F:24:HD:17:-ve

“Actually knowing that I’m not really going to be sick, so like, I wouldn’t have changed, but it’s just, it puts something to your mind at rest”

Doug:M:18:FAP:14:-ve
“Allowing me to live my life, knowing but not knowing in a way, knowing that if I live long enough, I will develop symptoms one day, um, but not knowing exactly what the rest of my life has in store for me and allowing me to accept that and just live, yeah”


“Not having to worry I suppose would be probably the best thing, knowing I’m clear, knowing I can’t pass it on”

Sally:F:25:FAP:17:-ve

“Actually like um knowing, um, knowing that my dad’s got this gene and he can pass it on to us and that we actually know, I mean deal with it rather than not knowing at all and having one of us or any of us, you know, sort of die because of this gene, and um, yeah, it’s just sort of the whole knowing thing and that you can prevent it, if something ever happens”

Emily:F:21:FAP:14:-ve

6.2.8.2 The worst

Once again, some individuals found it difficult to identify the worst aspect of their entire predictive genetic testing process:

“Can’t think of a worst thing, it’s yeah, no can’t think, there’s nothing, nothing bad come out of it so”

Doug:M:18:FAP:14:-ve

“There’s no bad thing, I think it’s a good thing cause it’s not as if we can help having it, if we had it or, so if you have it, then you just look after yourself, if you don’t well you’re lucky, you can’t put it any other way, you know”

Amy:F:19:FAP:14:-ve

For others the worst aspect of the process was apparent immediately:

“Having it”

Harry:M:14:FAP:12:+ve
“My parents, probably seeing what it did to my parents”


“Directly afterwards I was flat for about a month, I just couldn’t do anything, I had no energy to do anything…that was probably the worst aspect of it all… but I don’t, as far as my life in general, I don’t think it’s really been, I can’t think of a negative from it”

Zach:M:26:HD:23:-ve

“Telling people is just really really hard, you don’t know how they’re going to react, like when I told my grandfather that the disease had started, yeah, by the end of the conversation he was almost crying, but um, yeah, but we’re only talking about the predictive test, yeah, hardest thing, hmm, I don’t know, concern about the workplace I spose, yeah, cause it is a safety critical job, obviously it goes without saying”

Oliver:M:24:HD:19:+ve

“The worst thing was waiting for the result, and again, the worst thing was getting tested at 18, I felt like I went through 2 years of begging to be tested, 2 years of proving myself worthy enough, that I’m healthy enough to get tested, like, proving myself that I’m ok to get tested, that was horrible for me, cause, I’m human, I know that I might have the gene or not, isn’t that enough, I actually had to prove myself that I was worthy enough to find out the result, and I really didn’t like that, but I can see why that’s done, I can understand why it’s done, but um, I mean, yeah, that was the worst thing”

Ella:F:20:HD:18:-ve

“I think, before getting tested, just feuding all the time with my family, cause they didn’t know what I was doing, although dad did, um, just with brothers and sisters we always kept on arguing and we always had you know, little fights and that, and it wasn’t because of them, it was because of me because of waiting, you know, always thinking and things like that, so I think that was the worst thing about going through the whole process”

Travis:M:24:HD:20:-ve
“Facing it all I suppose, maybe, yeah, it makes it sink in a bit more, like, deeper, I don’t think it’s actually sunk in what mum’s going through at the moment and that”

Nina:F:23:HD:23:-ve

“Maybe, maybe um the stress on me family or something, me dad, me mum, me sister and that”

Troy:M:26:HD:25:-ve

“In my case the worst thing would have been that I felt that [brother] would get it more, yeah, like, I felt because I wasn’t, I thought that there’s more of a chance for him to get it sort of thing”

Poppy:F:24:HD:17:-ve

“The worst thing was the needle, that’s it, that was the worst”

Kylie:F:20:FAP:14:+ve

“I guess knowing that one day I will develop symptoms, that I do have a genetic disease and if I live long enough it will happen… I guess part of that is not knowing when, you know, that’s the worst part, not knowing when cause you don’t know if it’ll be 35, 55, 85! You know, yeah, so I guess that’s the worst part, I just try to keep it in the back of my mind… and occasionally… something will happen and I think oh god, you know, and then whatever it is has gone or whatever and it’s just forgotten again, I don’t ponder it, I don’t sit on it, but that’s the worst thing”


“The waiting times… just waiting to see what’s happened”


“I don’t know whether, whether this really relates to it or not but um, our information getting leaked out, that’s the major concern at the moment”

Emily:F:21:FAP:14:-ve
6.3 Conclusion

In this chapter I have relayed the experiences of 18 young people who have undergone predictive genetic testing for FAP or HD. I have presented their descriptions with minimal analysis. In the next chapter I interpret these experiences, identifying the harms and benefits of predictive genetic testing as described by young people themselves. I then analyse the meaning that young people ascribe to their predictive genetic tests, free from the categories of harm and benefit.
CHAPTER SEVEN: HOLDING YOUR BREATH
7.1 Introduction

“All my life I thought I was going to get this illness ... so for 19 years it feels like I’ve held my breath... and now it feels like I’m a new born child, like I can live a life that I never knew I could ... and it seems really weird to adjust to ... the whole time I thought I was going to get it and then she told me I didn’t have the gene and, even then, I wanted a second test... cause all my life I’ve just been gearing myself up to have this illness”

Ella:F:20:HD:18:-ve

In this chapter I analyse and interpret the descriptions that were conveyed to me by the 18 young people I interviewed about their predictive genetic tests. My means of interpretation is thematic analysis. I divide this analysis into two phases. As articulated in chapter one, my primary focus in this thesis is the issue of potential harm to mature young people who undergo predictive genetic testing. Therefore, the first phase of thematic analysis entails a search for aspects of the testing process that can be categorised under the themes of harm and benefit. However, after presenting the findings of this search I argue that, while useful in some ways, this method is too simplistic to further our understanding of predictive genetic testing in a detailed way. I argue that a more sophisticated and complex analysis is required. I therefore present a second phase of thematic analysis, aimed at capturing the meaning young people ascribe to the predictive genetic tests they have undergone. Here I leave behind the concepts of benefit and harm and investigate the data for the meanings that young people themselves attach to their experience of predictive genetic testing, in their own terms. Of critical importance in this second phase of analysis is the comparison of life before testing with life after testing. Finally, I bring both phases of interpretation together in order to propose what I believe to be a new way of thinking about predictive genetic testing in young people.
7.2 What are we listening for?

A clash of opinion is taking place in relation to predictive genetic testing in young people. It is thought that such testing will impact upon young people differently from the way in which it does upon adults. However, the empirical evidence available is incapable of indicating if these differences will result in greater harm or greater benefit when young people are tested. We know that such tests are being provided to young people for non-medical reasons internationally. We also know that the outcomes of these tests are not being followed systematically. There clearly exists an urgent need for empirical research to be performed concerning the impacts of predictive genetic testing in young people. This is required so that policy and practice may become evidence based. There is also a need for the initial phases of this research to be exploratory and deep so that we are able to identify a range of potential impacts and to clarify those that hold most significance to young people specifically. There is a need to listen to young people themselves. The question that remains then is this: what shall we listen for in these detailed accounts provided by young people?

In the interviews I conducted with 18 young people who had experienced predictive genetic testing first-hand, I listened for two distinct qualities initially. The first was the occurrence of positive, or beneficial, aspects of their testing experience. The second was the occurrence of negative, or harmful, aspects of their experience. This initial search for descriptions of the testing process that fit into the categories of harm and benefit constitutes the first phase of my thematic analysis, articulated below. In chapter two I also relayed the extreme lack of variety found within the range of harms and benefits that have been researched empirically thus far. I noted that, of the small range of harms and benefits that have been researched empirically, the majority are harms. Therefore, in this first phase of my thematic analysis, I search not only for the existence of both positive and negative aspects of the predictive genetic testing process, but also for a wide range of such outcomes.
7.3 Harms and benefits as described by young people

In this section I identify a range of harms and benefits associated with predictive genetic testing in young people, as described by young people who have experienced such testing themselves. I refer to aspects relating to the entire process of predictive genetic testing, not simply the time after disclosure of test results. Many of the harms and benefits I articulate below have been portrayed in quotes I presented in the previous chapter. Therefore, in order to avoid excessive repetition, I provide examples for only a sub-set of the harms and benefits I identified.

7.3.1 Harms

The young people I spoke with referred to several harms, or negative aspects, associated with the predictive genetic tests they underwent. I have divided these into two groups: harms articulated by young people who received a gene-positive test result and harms articulated by young people who received a gene-negative test result.

7.3.1.1 Harms articulated by young people who received a gene-positive test result

Young people who received a gene-positive test result described several harms associated with the predictive genetic testing process. Receiving a gene-positive test result was difficult in itself for several young people:

“[It is] something that I have for life and could kill me”

Harry:M:14:FAP:12:+ve

“I got a bit angry… um, shaky, ah, ok, very down, depressed”


“One day I will develop symptoms… I do have a genetic disease, and if I live long enough it will happen”


Others described concern about their gene-positive status affecting their ability to work:
“Well there’s a few things at work now, yeah, I’m going to be assessed by an occupational therapist”

Oliver:M:24:HD:19:+ve

Work gossip also caused concern:

“All work is yap yap yap yap, it’s just phenomenal, oh, yap yap yap yap, such and such is dying from this, dying from that, dying from this, they all just create numerous stories”

Oliver:M:24:HD:19:+ve

For some, the gene-positive test result became particularly difficult when other things were going badly:

“I had a new boyfriend at the time and it wasn’t going so well and you know, sort of, I got into a bit of depression with that, and the HD and everything all just flooded back”


Another difficulty associated with the receipt of a gene-positive test result was watching parents become upset:

“When my dad… realised I had it he went and sat in the bedroom and didn’t come out for like 2 hours and you know, when I saw that kind of behaviour from my dad… that really upset me then… when I saw what it did to my parents”


Some young people found it difficult identifying with other family members who were gene-positive:

“She goes you’re one of us now, and I’m like, I don’t want to be one of you!”

Kylie:F:20:FAP:14:+ve
And some felt distanced from their parent who is gene-negative:

“It probably did distance [things] a bit with my father, just not being able to relate”


Some difficulties were noted about the impact of testing upon friendships:

“My best friend, she did notice… I wasn’t really talkative”

Kylie F: 20: FAP: 14: +ve

“I probably wasn’t as outgoing as I was so I probably did crawl into my shell a little bit”


“I think I actually took a couple of days off after the test just to, you know, just to… take some time to myself… I don’t think I would have been able to have been much use at school, be it with other students or with my work”


And some young people felt anxious about other’s reactions to their gene-positive test result:

“Telling people is just really really hard, you don’t know how they’re going to react”

Oliver M: 24: HD: 19: +ve

“I’ve been let down by a couple of people who, you know, who are… thoughtless, just the complete thoughtlessness and selfishness”

Oliver M: 24: HD: 19: +ve

Negative impacts were associated with some means of disclosure:

“I don’t think they should have just said that over the phone”

Ali F: 24: FAP: 15: +ve
While regret about having the predictive genetic test was also described:

“I would rather of not ever known”

Mark: M: 21: FAP: 16: +ve

The waiting time for test results was particularly difficult for some:

“The waiting times, just waiting to see what’s happened”


As was the age at which testing occurred:

“I didn’t really understand it completely, like I do now”

Harry: M: 14: FAP: 12: +ve

7.3.1.2 Harms articulated by young people who received a gene-negative test result

Young people who received a gene-negative test result also described several harms associated with the predictive genetic testing process. Difficulties sometimes related to their family:

“She [Mum] sort of let me go off and do what I’ve gotta do, you know… she’s free to do what she wants now… if anything we’ve drifted apart but, I mean, our relationship, our friendship’s still exactly the same, it’s just um, we’ve drifted apart a bit.”

Ella: F: 20: HD: 18: -ve

“Feuding all the time with my family… it wasn’t because of them, it was because of me, because of waiting, you know, always thinking and, and things like that”

Travis: M: 24: HD: 20: -ve

“I was thinking because I didn’t have it, is it more chance [brothers]’s going to get it?… That’s another thing I was worried about …I’d be the only one [who could care for him]”

Poppy: F: 24: HD: 17: -ve
“I was more worried that she had it, like it was a big shock...I think I sort of felt a bit guilty because she ended up having it and I didn’t”

Liz:F:17:FAP:10:-ve

For others, the difficulties related to a feeling of being lost after the receipt of their gene-negative test result. Some young people wish that they could have had more time with the counsellor following disclosure of their test result:

“Directly afterwards I was flat for about a month, I just couldn’t do anything, I had no energy to do anything”

Zach:M:26:HD:23:-ve

“I would have loved to keep catching up with her and trying to work out, ok, this is what you do from here, yeah, but I’ve been left to go off and work it out for myself and I find that really hard, really frustrating, cause what to do?”

Ella:F:20:HD:18:-ve

However, for others, the length of the counselling was a negative aspect of the process:

“Continuously going through counselling sessions when you just want to… know what’s going on”

Travis:M:24:HD:20:-ve

Some young people felt regret about the timing of their predictive genetic test:

“I wouldn’t have done year 12... I would have taken time out for myself”

Ella:F:20:HD:18:-ve

While for others, the disclosure of test results to other members of their family caused great concern:

“Our information getting leaked out, that’s the major concern at the moment”

Emily:F:21:FAP:14:-ve
7.3.1.4 A summary of harms

The range of harms that are articulated by young people who have undergone predictive genetic testing is broad. Of particular importance within this range are two points. Firstly, there are several harms associated with receipt of a gene-negative test result. This is somewhat counter-intuitive, as receipt of a gene-negative test result constitutes ‘good news’. A gene-negative test result is the outcome that young people hope for. It is thus perhaps surprising that such ‘good news’ is often also accompanied by negative, or harmful, consequences. The second point worth highlighting is that several of the harms articulated by young people could occur in relation to either a gene-positive or a gene-negative test result. Harms such as feeling distanced from a parent who has a different gene status, feuding with family members during the waiting period and feeling regret about the effect that testing had upon school are not specific to one test result only. These harms and several others are potential outcomes of the predictive genetic testing process regardless of the test result received.

Together, these two points indicate that many of the harms associated with predictive genetic testing in young people are much broader than simply reactions to a gene-positive test result. Although this may be counter-intuitive, it is vital that we understand this fact when engaging in debate about the provision of such testing to young people.

As I stated at the beginning of this section concerning harms associated with predictive genetic testing in young people, I chose to provide examples for only a subset of the total range of harms that were articulated. I did this in order to avoid repetition, as several descriptions of the harms associated with such testing were presented in the previous chapter. Therefore, I now present a complete list of the harms associated with the predictive genetic testing process, as articulated by the young people I interviewed, in Table 7.1.
Table 7.1 Harms associated with testing in young people

<table>
<thead>
<tr>
<th>Harms articulated by young people who receive a gene-positive test result</th>
<th>Harms articulated by young people who receive a gene-negative test result</th>
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<tbody>
<tr>
<td>▪ Concern about not being able to perform at work, or being fired</td>
<td>▪ Feeling flat after receipt of the result</td>
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<tr>
<td>▪ Anxiety about other people learning of the test result or gossiping about it</td>
<td>▪ Feeling lost after receipt of the result</td>
</tr>
<tr>
<td>▪ Feeling let down by friends who are unsupportive</td>
<td>▪ Not knowing how to proceed after receipt of the result</td>
</tr>
<tr>
<td>▪ Worry about how other people will react to news of the test result</td>
<td>▪ Drifting apart from parents</td>
</tr>
<tr>
<td>▪ Being treated differently by friends</td>
<td>▪ Feuding with family members while waiting for the test result</td>
</tr>
<tr>
<td>▪ Having to provide explanations about genetics to friends</td>
<td>▪ Believing that one’s own genetic test result implies a specific test result in a sibling</td>
</tr>
<tr>
<td>▪ The gene-positive test result re-surfacing when other things go badly in life</td>
<td>▪ Feeling concerned about having to look after a gene-positive sibling later in life</td>
</tr>
<tr>
<td>▪ Worry about the onset of the genetic condition</td>
<td>▪ Feeling concerned about siblings with a gene-positive test result</td>
</tr>
<tr>
<td>▪ Watching a symptomatic parent as a gene-positive individual</td>
<td>▪ Feeling guilty about a gene-negative test result when siblings are gene-positive</td>
</tr>
<tr>
<td>▪ Feeling stuck with something for life</td>
<td>▪ Wanting more contact with the counsellor after receipt of the test result</td>
</tr>
<tr>
<td>▪ Knowing that the genetic condition could be fatal</td>
<td>▪ The testing process interfering with the final year of school</td>
</tr>
<tr>
<td>▪ Feeling shocked by the result</td>
<td>▪ Feeling anxious during the waiting period for test results</td>
</tr>
<tr>
<td>▪ Feeling depressed about the result</td>
<td>▪ Feeling that the counselling sessions prior to disclosure of test results went on for too long</td>
</tr>
<tr>
<td>▪ Feeling angry about the result</td>
<td>▪ Feeling angry about test results being disclosed to other family members</td>
</tr>
<tr>
<td>▪ Feeling despair about the result</td>
<td>▪ Thinking “why me”</td>
</tr>
<tr>
<td>▪ Being too young to understand what the test entailed</td>
<td>▪ Feeling flat after receipt of the result</td>
</tr>
<tr>
<td>▪ Feeling angry that test results were disclosed over the telephone</td>
<td>▪ Feeling lost after receipt of the result</td>
</tr>
<tr>
<td>▪ Watching parents become upset</td>
<td>▪ Not knowing how to proceed after receipt of the result</td>
</tr>
<tr>
<td>▪ Feeling distanced from parents who do not share the same genetic status</td>
<td>▪ Drifting apart from parents</td>
</tr>
<tr>
<td>▪ Not wanting to identify with sick family members</td>
<td>▪ Feuding with family members while waiting for the test result</td>
</tr>
<tr>
<td>▪ The desire for time alone affecting friendships</td>
<td>▪ Believing that one’s own genetic test result implies a specific test result in a sibling</td>
</tr>
<tr>
<td>▪ Needing to take time off school</td>
<td>▪ Feeling concerned about having to look after a gene-positive sibling later in life</td>
</tr>
<tr>
<td>▪ Regret about having the test</td>
<td>▪ Feeling concerned about siblings with a gene-positive test result</td>
</tr>
<tr>
<td>▪ Waiting for the test results</td>
<td>▪ Feeling guilty about a gene-negative test result when siblings are gene-positive</td>
</tr>
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</table>

7.3.2 Benefits

In presenting the benefits, or positive aspects, associated with predictive genetic testing as described by the 18 young people I interviewed, I again divide them into two groups. Firstly, benefits articulated by young people who received a gene-positive
test result. Secondly, benefits articulated by young people who received a gene-negative test result.

7.3.2.1 Benefits associated with a gene-positive test result

Young people who received a gene-positive test result articulated several benefits associated with the predictive genetic testing process. For some young people, the actual process of predictive genetic testing created benefits for them:

“I think it was a pamphlet that was sent, or a letter … and it required my signature, not mum’s, I remember that which was of course quite surprising being under 18, it actually made [me feel] quite good and nice and important”


Many also referred to the positive impacts of finding out what their future holds:

“Probably putting everyone’s mind at rest I would say, because we all knew, we just all knew”


“I just felt relief because I knew, and I had some sense of what was going to happen to me later in my life”


“I’m glad I know”


“Instead of umming and aahing, am I going to be 60, am I going to be 40… a completely decisive answer”

Oliver:M:24:HD:19:+ve

The predictive genetic testing process improved relationships within the families of some young people:
“I think because my dad’s had the surgery, and then for the same thing to happen to me, I think that you create a bond… you do just become a lot closer… I would definitely say that we share like a special bond and we’re closer now than we were before then”


“In hindsight it’s improved the relationship between my father and I”

Oliver:M:24:HD:19:+ve

“When I told him my result was positive he gave me a hug…(laugh)… it’s very rare for him”


“I see friends of ours with their kids and they’re getting to that age where they’ve just got that… smart-mouth and… just at that age they don’t think it’s necessary to tell their parents they love them or give them a kiss before they go to bed and stuff like that…you realise when you go through that at that age, you sort of lose a bit of that attitude and realise well shit, look what’s happening here, I need to not take these things for granted”


“My mum and I talk a lot more… with this… FAP thing we can actually relate which is good”

Kylie:F:20:FAP:14:+ve

While others felt that the gene-positive test result allowed them to establish new relationships, or strengthen existing ones:

“It’s given us a level to relate to each other on… sort of getting to know each other, that was our relation level, so I guess in that way, if my test had been negative that may not have happened”

“It kind of brought us closer because my best friend, she has diabetes … and now that I’ve got this, we can sort of… you know, I’ve got something… and she’s got something”

Kylie:F:20:FAP:14:+ve

“There’s actually one girl, I never knew her… but she… didn’t play hockey for a while and… I found out later she had the same operation that dad had, so now I’ve started talking to her and we’re really good friends”

Mark:M:21:FAP:16:+ve

Some young people felt that life was improved by the predictive genetic test:

“Allowing me to live my life, knowing but not knowing in a way, knowing that if I live long enough… I will develop symptoms one day… but not knowing exactly what the rest of my life has in store for me and allowing me to accept that and just live.”


While the counselling process itself was also rewarding for some:

“When you have the counselling… it brings out anything, any problems… most people have never had counselling… so it starts off being an HD related thing, but… they talk about everything so if there’s some sort of problem in your life that’s got nothing to do with HD they can help you with that”


7.3.2.2 Benefits associated with a gene-negative test result

Young people who received a gene-negative test result also articulated several benefits associated with the predictive genetic testing process. A major benefit of being tested for those who received a gene-negative test result was finding out that are free from the condition that runs in their family:

“Finding out I don’t have it”

Nina:F:23:HD:23:-ve
“I don’t know if I could even explain it…the relief I felt”

Poppy:F:24:HD:17:-ve

“It was good knowing that… you know… I didn’t have this disease”

Doug:M:18:FAP:14:-ve

For several young people, it was a relief to know that they could not pass it on to children:

“Finding out my daughter’s not going to have to go through it either”

Nina:F:23:HD:23:-ve

Several comments were made about the freedom from worry that accompanied the gene-negative test result:

“Not having to worry”

Sally:F:25:FAP:17:-ve

“I didn’t have to worry about it any more”

Emily:F:21:FAP:14:-ve

For many, simply having an answer, and knowing what the future holds was a benefit:

“We actually know”

Emily:F:21:FAP:14:-ve

“Finding out the result, just the result, I don’t care if it was positive or negative, but actually finding out a result and directing my life”

Ella:F:20:HD:18:-ve

For some, the predictive genetic test improved relationships with family members:

“I spose I was a lot more… not a lot more, but a little bit more open to seeing [my dad]”

Zach:M:26:HD:23:-ve
“Bringing the family closer cause they were talking about it and being open about it and stuff”

Liz:F:17:FAP:10:-ve

Some felt good about the impact that their gene-negative test result had for their parents:

“Mum can breathe too”

Ella:F:20:HD:18:-ve

“They were happy, they were happy”

Emily:F:21:FAP:14:-ve

Some also felt comforted that, in their families, the condition ceased with them:

“This part of the family tree is cut off from any further”

Emily:F:21:FAP:14:-ve

Friendships improved for some young people:

“The community and the friendship… I just found out was so solid through the process”

Zach:M:26:HD:23:-ve

And many felt that their gene-negative test result had allowed them to plan for the future more:

“It gives more freedom, being able to plan a life”

Zach:M:26:HD:23:-ve

“We’ve got a wonderful life ahead of us… it’s been good since we found out”

Ella:F:20:HD:18:-ve

“You can sort of plan ahead, just for what you’re going to do”

Troy:M:26:HD:25:-ve
The test result also allowed some young people to enjoy life more:

“Life’s better… happy and cheery”

Travis: M: 24: HD: 20: -ve

“I seem to have changed a bit, just come out of me shell a bit more… you know, a bit happier and stuff”

Troy: M: 26: HD: 25: -ve

Going through the predictive genetic testing process was an empowering experience for some:

“I respected myself a lot more after going through it all… I thought well, if I can go through this whole process of getting tested for this thing, I can pretty much do anything”

Travis: M: 24: HD: 20: -ve

One young person felt that it was beneficial that she was too young for the test to have had much of an impact upon her:

“If I had of had the test done now, it would have had a bigger impact on me than what it would have six years ago because I think if you are younger you don’t really sort of have the same sort of knowledge that you have when you’re older and things can have a bigger impact on you”

Emily: F: 21: FAP: 14: -ve

7.3.2.3 A summary of benefits

The range of benefits that have been articulated by young people who have undergone predictive genetic testing is also broad. As was the case with the range of harms that were described, there are two crucial points to note about the benefits presented above. Firstly, just as it was surprising to find that there were several harms associated with the receipt of a gene-negative test result in the previous section, it is interesting to find that a range of benefits are associated with receipt of a gene-positive test result. This is also somewhat counter-intuitive, given that a gene-positive test result is
perceived as ‘bad news’. Importantly though, it seems that ‘bad news’ can also result in positive consequences. The second point to highlight is that, as was again the case in relation to the harms associated with predictive genetic testing in young people, several of the benefits articulated by young people could occur in relation to either a gene-positive or a gene-negative test result. These include feeling empowered by the testing process, feeling that family relationships have been improved and experiencing relief about knowing what the future holds. It therefore appears that the benefits associated with such testing extend far beyond obvious reactions to a gene-negative test result. Although this is counter-intuitive, once again this is a critical point to remember when engaging in debate about the provision of such testing to young people in the future.

I now present a complete list of the benefits that were articulated by the young people I interviewed, in Table 7.2.
### Table 7.2 Benefits associated with testing in young people

<table>
<thead>
<tr>
<th>Benefits articulated by young people who received a gene-positive test result</th>
<th>Benefits articulated by young people who received a gene-negative test result</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Improving relationships with family members, including parents, siblings and grandparents</td>
<td>- Improving relationships with family members, including parents and siblings</td>
</tr>
<tr>
<td>- Relief about finding out the test result</td>
<td>- Creating a reason to spend more time with parents</td>
</tr>
<tr>
<td>- Knowing what is going to happen in the future</td>
<td>- Feeling more able to help gene-positive parents</td>
</tr>
<tr>
<td>- Removing the uncertainty</td>
<td>- Realising how strong and important some friendships are</td>
</tr>
<tr>
<td>- Establishing new relationships with people in similar situations</td>
<td>- Feeling that children are a possibility</td>
</tr>
<tr>
<td>- Feeling that a mental block has been removed</td>
<td>- Feeling more free and able to plan for the future</td>
</tr>
<tr>
<td>- Having the opportunity to deal with many issues in the counselling sessions</td>
<td>- Feeling excited about the future</td>
</tr>
<tr>
<td>- Moving through the rebellious teenage stage quickly</td>
<td>- Knowing that partners will not be burdened by the genetic condition in the future</td>
</tr>
<tr>
<td>- Realising how strong parental support is</td>
<td>- Feeling relief</td>
</tr>
<tr>
<td>- Being able to help younger siblings through the same process</td>
<td>- Drawing people together</td>
</tr>
<tr>
<td>- Putting people’s mind at rest</td>
<td>- Knowing what the future holds</td>
</tr>
<tr>
<td>- Improving and strengthening friendships</td>
<td>- Knowing that this genetic condition will not develop</td>
</tr>
<tr>
<td>- Feeling empowered by the process</td>
<td>- Eradicating one of the unpredictabilities associated with the condition</td>
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</table>

#### 7.3.3 Conclusions about benefit and harm

The harms and benefits associated with predictive genetic testing in young people that I have described above differ from the harms and benefits that have been reported in the past in two important ways. Firstly, the harms and benefits presented here constitute aspects of the predictive genetic testing process that *young people themselves* have found difficult, or pleasing. Traditionally, the starting point of
empirical research in this field has involved a theoretical list of harms or benefits devised by professionals (most often harms). However, the lists articulated above are capable of re-positioning this starting point, allowing us to begin with young people’s subjective accounts of the ways in which predictive genetic testing affects them. The second important aspect of the lists articulated above is the range of harms and benefits referred to. This range far exceeds the harms and benefits that have previously been investigated or described in the literature. There is a need to expand the way in which we think about the impacts of predictive genetic testing in young people from herein so that we may encompass a more diverse range of potential outcomes in future research and debate.

An additional significant outcome of my search for harms and benefits associated with predictive genetic testing in young people is that the consequences of testing differ greatly between individuals. For example, some young people felt that the predictive genetic testing process brought them closer to their parents, while others felt that it created distance. Developing a detailed list of potential benefits and harms, although useful, therefore does not in itself provide an indication of how individuals are likely to experience the testing process. It simply provides a range of possibilities that are then modified by the individual’s unique circumstances.

Also of importance is the finding that the consequences of predictive genetic testing in young people are not always concordant with the reactions that would be most obviously assumed to take place in response to a gene-positive or gene-negative test result. In other words, gene-positive test results do not only cause harmful consequences and gene-negative test results do not only create beneficial ones. Past literature has recognised that if young people receive a gene-positive test result, there is potential for benefit, not just harm. Some of the benefits that have been articulated as possible consequences of a gene-positive test result in the past include the opportunity to prepare psychologically for onset of the condition and the opportunity to make better informed decisions about career, reproduction and finances. However, past literature recognises little potential for harm following gene-negative test results. The concept of survivor guilt has been mentioned frequently, where young people feel guilty about a gene-negative test result in response to siblings and parents who are gene-positive, but few other potential harms are discussed in relation to gene-
The harms articulated above by young people who received gene-negative test results therefore constitute critical and new descriptions of the predictive genetic testing process.

### 7.3.4 Benefits and harms related to young people specifically

When contemplating the current debate about such testing, it is important to bring the focus back to young people specifically. The core issue I address in this thesis is the issue of potential harm to mature young people who undergo predictive genetic testing for non-medical reasons. It is therefore vital to consider which of the harms and benefits articulated above are in fact specific to young people. Two questions need to be asked. Firstly, which of the harms and benefits articulated above could only occur in young people and not adults? Secondly, which of the harms and benefits articulated above may have an increased magnitude in young people, when compared to adults? I addressed these same questions in chapter three in relation to the harms speculated on in the literature, and found that only some alleged potential harms were specific to young people. Here again, it is important to consider the question of whether the experiences of the young people I interviewed came about as they did because they were young, or whether these experiences could have happened at any stage. Such consideration can only be tentative, of course. There is no way of knowing what these young people’s experiences would have been if they had been tested later in life. Nevertheless, by considering the nature of their experiences, it is possible to point to some features that seem particularly related to adolescence.

There are both harms and benefits described above that could only occur as a result of predictive genetic testing in young people. Some of these harms include needing to take time off school and being too young to properly understand what the test would involve. Some of these benefits include moving through the stage of ‘teenage rebellion’ more quickly and being too young for the test to have any serious impact. There are also many harms and benefits that may occur when adults are tested, but that may have a greater magnitude in young people. Some of these harms include watching parents become upset, friends gossiping about the test result, feuding with family members and feeling guilt about siblings’ discordant test results. Some of these benefits include making the family more open, feeling empowered, creating a reason to
spend time with parents, feeling able to plan for the future and feeling excited about the prospect of having children.

This initial phase of thematic analysis, aimed at identifying aspects of predictive genetic testing in young people that fit within the categories of harm and benefit, has therefore demonstrated some outcomes of testing that may be different for young people, when compared to adults. This is crucial for future debates about predictive genetic testing in young people and will be referred to in detail in the final chapter.

7.4 The risks of simplicity

Searching for content that fits into the categories of benefit and harm is perhaps the most obvious means of interpreting the information conveyed to me by the 18 young people I interviewed. Those who oppose predictive genetic testing in young people focus on the potential for harm that such test provision has, while those offering support write of the benefits that may occur if testing is provided. Thus it seems logical to aim at identifying aspects of the testing process that fit within these categories of harm and benefit. However, I argue here that although this first phase of thematic analysis is a useful way to begin, it is far from the most valuable means of furthering our understanding of the experience of predictive genetic testing for young people.

When we search only for content that fits into the categories of benefit and harm, we simplify the predictive genetic testing experience. In doing so, we fail to acknowledge the complexity of life for young people who undergo such testing. The impact that testing has for these young people, and the meaning they ascribe to the experience, is broader than simply the harms and benefits that testing creates. In fact, many of the descriptions conveyed by young people do not fit neatly into either the category of harm or the category of benefit. Other descriptions could sit within either category. In searching only for content that does fit into the categories of harm and benefit, we miss the more complex, yet equally important descriptions of the impact that testing has for young people. Take the following examples:
“It reaffirmed my decision not to have children”


“It’s changed my opinion on money completely, you know… the bottom line is money can’t buy happiness, it can assist and help you along, but it does not buy happiness and it does not change someone’s status in a disease sense, and that’s the bottom line… I’ve sort of got really really funny with my money lately”

Oliver:M:24:HD:19:+ve

“There’s stuff I never imagined like, I’m going to have to get life insurance and a car”

Ella:F:20:HD:18:-ve

These quotes convey complex yet essential descriptions of the impact that predictive genetic testing has for young people, yet they do not fit neatly within the categories of harm or benefit. Thus, when we seek only to list the benefits and harms of young people’s descriptions, we blind ourselves to the more muddled descriptions that are provided alongside the tidy ones. These jumbled descriptions are not able to fit neatly into categories of harm and benefit, yet contribute to enhanced understanding in this field, perhaps even more than neatly categorised lists of benefits and harms. I therefore move now from my first phase of thematic analysis, related to aspects of the testing process that fit within the categories of harm and benefit, to my second phase of thematic analysis, aimed at reaching a more sophisticated understanding of young people’s experiences of predictive genetic testing.

7.5 Something more complex

In this second phase of thematic analysis I refrain from using the categories of harm and benefit to organise the descriptions provided to me by the 18 young people I interviewed. Instead, I articulate themes that emerged during the qualitative research process, irrespective of their relationship to the concepts of harm or benefit. My aim here is to present the themes that recurred during my interviews with young people and
then to place these themes within a context of attempting to better understand what predictive genetic testing is like for young people, as opposed to whether it harms or benefits them. In order to articulate the themes I found emerging during my conversations with young people, I divide their experiences into two parts: life before testing and life after testing.

7.5.1 Life before testing

There are three themes that emerged through my interviews with young people that relate to the time before the predictive genetic test took place. In describing these themes I make a crucial departure from the way in which predictive genetic testing in young people has been traditionally written about.

In current debates about such testing, the time after testing has taken centre stage. Existing discourse relates to what happens once testing is performed, the effects that such an experience has and the ways in which it impacts upon the lives of those tested. Thus taking time to understand the types of lives young people live before they undergo testing is, in a sense, radical. However, as young people conveyed their experiences of testing to me, I found themes repeatedly emerging that related to this time prior to their test. I therefore relay three of these themes below: (1) Risk behaviours, (2) Thinking gene-positive, and (3) Complex pasts.

7.5.1.1 Risk behaviours

This first theme relates to the types of behaviours that some of the young people I spoke with were engaging in prior to their predictive genetic test. A theme emerged where, for several young people interviewed, the time before testing was a time that involved drug use, difficulty at school, and even trouble with the police. As outlined in chapter three, adolescence is often a time where young people engage in a variety of risk behaviours as they develop and hone their individual sense of self, moving from dependency to independence. However, the frequency of risk behaviours within the group of 18 young people I spoke with surprised me.

Ella describes here how the thought of her future illness affected her behaviour:
“I mucked up a lot I think, cause I just thought I was going to get this illness”

Ella:F:20:HD:18:-ve

She then elaborates further:

“It was all a really crazy time when I was 18, you know, I was doing year 12 and I was getting genetically tested, I mean, my head was just like, this is gonna be it, this is your last year, just do anything and everything, don’t financially plan, I didn’t give a fuck honestly”

Ella:F:20:HD:18:-ve

And refers to her use of heroin as a means of coping:

“I got into drugs, I got onto heroin…it was a crutch for me”

Ella:F:20:HD:18:-ve

Nina also refers to drug use. In her case, it was amphetamines:

“I’ve been on drugs for like 3, 4 years or something… speed…and I’ve just gotten off”

Nina:F:23:HD:23:-ve

She also talks of difficulty at school, and plans to rectify this now:

“I dropped out of school of course… I want to do my year 11 and 12 by correspondence”

Nina:F:23:HD:23:-ve

Travis also talks of difficulty at school. He specifically mentions that he doesn’t believe he would have misbehaved to the extent that he did if he hadn’t been seeking predictive genetic testing for HD:

“I didn't do well at school… just misbehaved really, did what a 14 year old kid would normally do, but just times 10…I don’t think I would have done it anyway”

Travis:M:24:HD:20:-ve
Zach mentions not caring much about school, yet he and his mother have different views about the cause of this:

“I went through high-school not really caring about getting good marks or anything like that because it just wasn’t important to me at the time…my mum thinks it was because I didn’t think that I had a future because of HD but I didn’t necessarily see it that way, I just think it was just cause I really wasn’t into school or I was more into the arts side of things rather than doing really well academically”

Zach:M:26:HD:23:-ve

Troy speaks of several difficulties during the years before his test. Critically, he sees these difficulties as the factors that motivated him to pursue testing:

“I used to drink a lot and get locked up and that, and it might have been down to, you know, that in the back of my mind and stuff… and I had depression and stuff…I left school at 15 …I just sort of kept to myself a lot, in me room and stuff…smoking and stuff…then I started feeling like weird going down the street and that, so I went and seen a doctor and he put me on prozac, and that sort of, I don’t know, it seemed like it made it worse…I just started going stupid and stuff, so he tried me on all other different stuff and that…yeah, I was real shaky and just didn’t want to go out and do anything…and then if I did, you know, get drunk or whatever, I’d sort of, it’d just make me sort of get real angry and stuff…I ended up going and seeing a sort of psychiatrist and they said there was, like I had been getting panic attacks and like anxiety and stuff like that, so he put me on, like valium and stuff…but I still, I still binge dranked and all that sort of stuff…I was still getting in trouble with the police and that…I was still drinking pretty heavy and that…well that’s why I went and got the test done, like, I thought, you know, maybe in the back of my mind, you know, subconsciously it sort of, maybe that was the cause of all the shit that was going on”

Troy:M:26:HD:25:-ve

He then articulates the exact moment he decided to pursue testing:
“One night I just got locked up for drunk and disorderly, and I just sort of, I was just talking to me old man and I just said, you know, maybe it might pay off just to go and, you know, maybe it’s in the back of me head or something, just go and do it, and I just said, I just want to do it on me own, I don’t want anyone else there, and sort of done it”

Troy:M:26:HD:25:-ve

Troy then refers again to the possibility that a history of HD may cause some young people to engage in risk behaviours. Here, he gives advice on helping other young people at risk:

“I think also if um, with teenagers and stuff, if they start sort of going silly and that, you know, maybe vandalising and stuff, it might be down to seeing it in the family or something and maybe, um, take them to counselling just to find out if it is that”

Troy:M:26:HD:25:-ve

Belinda refers to difficulty with severe depression during her teenage years:

“I had depression in my early teens… at that stage I nearly got to the stage of killing myself, when I was in my early teens”


Ella also mentions multiple suicide attempts:

“If I didn’t go into hospital, I would have killed myself, for sure, there were many many, too many attempts of suicide, um, a lot of them ended up in emergency”

Ella:F:20:HD:18:-ve

Teenage pregnancies are also sometimes associated with risk behaviours during adolescence. Both Nina and Poppy had children during their teenage years:
“I’ve got two [kids], well I’m pregnant actually, I’ve got one and I’m pregnant… Charlotte’s 7 [years old]”

Nina:F:23:HD:23:-ve

“Kimmie… she’s nearly 6 [years old]”

Poppy:F:24:HD:17:-ve

Thus for some of the young people I spoke with, the time before testing often involved several risk behaviours. Importantly, some young people identified their genetic risk status, or their desire for testing, as a causal factor of this behaviour.

7.5.1.2 Thinking gene-positive

The second theme to emerge concerning life before testing relates to young people’s perceptions of what their test result would be. Although all the young people I spoke with knew at the time of testing that, logically, there was a 50% chance of inheriting the condition that runs in their family, several of them held a specific belief about the test result that they would receive. Importantly, many of the young people I spoke with spent the time prior to testing preparing themselves for a gene-positive test result, believing that they would go on to develop the condition later in life.

Emily describes here how she convinced herself that she was going to receive a gene-positive test result:

“I was convinced up until that day that I had it…I just think that I always seem to focus on the negative side, trying to bring myself up to be really bad and then when it’s not you go oh thank god, it’s just one of those things that I seem to do a lot”

Emily:F:21:FAP:14:-ve

Her sister, Liz, explains the same belief:

“I remember me and Emily were convinced that we’d have it…I think it was just kind of like, I don’t know, so that if we did it wasn’t such a shock”

Liz:F:17:FAP:10:-ve
Ali again describes how sure she felt about the fact that she would receive a gene-positive test result:

“I knew I would have it, I knew, because everyone has, I know it’s got nothing to do with it, but everyone’s always said from the time I was born that I was exactly like my father, I look like him, I talk like him, I’m outgoing like he is, all that kind of stuff, so I knew, I knew in my heart I swear, like I know there’s a 50-50 chance, but I knew I’d have it…I wouldn’t say I was shocked, because like I said, I expected it, I really did, I sort of prepared myself a bit for that”


Zach again refers to a similar belief about his test result, noting that he feels it is a natural reaction to hold such a belief:

“Knowing that there’s a 50:50 chance, it’s just, like it’s always in your head that yes I have it, rather than no I don’t have it…I think it might just be a natural reaction do so as well…yeah, I didn’t build it up too high, like I was more towards yes, it’s gonna come back positive rather than negative…yeah, so I thought it was more positive”

Zach:M:26:HD:23:-ve

Travis describes himself as pessimistic and suggests that this may have contributed to his belief about his test result:

“I’m a very pessimistic person, I was always saying it’s going to be positive, it’s going to be positive, you know, I’m going to have Huntington’s…I was always convincing myself because if I was going to be positive, it wouldn’t have been that much of a shock because I’ve sort of said to myself, you know, I’m going to be positive anyway”

Travis:M:24:HD:20:-ve

Oliver refers to the process of preparing himself for a gene-positive test result, just in case, but doesn’t explicitly mention a belief that he would be gene-positive:
“You secretly hope, you know, that you’re going to get a gene-negative result, but ah, you know, a 50% chance, you’ve got to really deal with it, and start to deal with it already, before the test result”

Oliver:M:24:HD:19:+ve

He then goes on to describe how he used to watch for symptoms, jumping to conclusions about his test result:

“I was 18 or 19 years old and you know, you don’t sleep and then you get on the train and you drop your keys and you drop your tickets and you drop your money ... and at that stage it is simple clumsiness and an enormous lack of sleep that causes such things but to someone with my history, I spose, you’re thinking oh is this Huntingtons, so you know, it always sort of goes through your head”

Oliver:M:24:HD:19:+ve

Ella relays a much more definite belief about a gene-positive test result. Here, she talks of her Uncle as a motivation to prepare herself for a gene-positive test result:

“It scared the shit out of him, he killed himself, so I thought, well, I’m not going to let this illness scare me at all, I’m going to gear myself up, think I’m going to have it... you know, that sort of was my protective mechanism”

Ella:F:20:HD:18:-ve

She also speaks of the reasons why she feels she set herself up for a gene-positive test result:
“Just to stop the devastation, just to gear myself up to deal with it, like, it’s such a devastating thing like you know, in a few years I’m not allowed to, like my drivers licence will be slowly cut down and I’m not allowed to get life insurance and you know, I’ve wanted to be a lawyer some of my life and go and join the army but no way would I get in and um I thought, well, you know, I’m going to get kicked back over and over again as the years go on so gear yourself up, you know, you might as well start preparing now, and yeah, I just, I wanted to give myself up for the worst, yeah, cause if the worst doesn’t happen, only good can come from there I thought”

Ella:F:20:HD:18:-ve

Again here Ella explains why she found it easier to believe she was going to be gene-positive, as opposed to gene-negative:

“You just start tricking your mind that you’ve got it because to sit back and go ok it’s going to be 50-50, your mind starts to boggle, ok if I have it this is what I’m going to have to do, if I don’t have it this is what I’m going to have to do… and you’re just constantly going back and forth in what to do with yourself and the situation comes up so, I just lent to one side, which was the worser side, and, it was sort of easier to manage”

Ella:F:20:HD:18:-ve

And again, in describing some regrets about her last year of school, she is firm about the belief she held at the time:

“In the last year I’ve gone, oh-oh, I’m going to have to do something with myself, you know, like, I sort of wasted my year 12 away so badly… I’m really upset for that, that I did that, you know, but I honestly, honestly thought I was going to get the gene, honestly”

Ella:F:20:HD:18:-ve

Poppy describes a suspicion she had about receiving a gene-positive test result, given her family history:
“I just thought I had it, obviously I didn’t have signs or anything, but, like, in my head, I just thought well, yeah, it’s a bonus if I don’t have it, cause I thought, the amount of people that had it in my family, yeah, I thought that, yeah, you’d pretty lucky to skip it”

Poppy:F:24:HD:17:-ve

Kylie refers to the day her family GP informed her that the test results were back:

“Well he just said outright one of us has got it, but we didn't know which one, but I knew it was me, I just had that feeling it was going to be me”

Kylie:F:20:FAP:14:+ve

Thus several of the young people I spoke with had started to prepare themselves for a gene-positive test result long before the testing process actually began. Some attribute this simply to their personality type, while others perceive it as a coping mechanism, helping them to prepare for their future. Either way, this shows that the test result is delivered in a context of expectation.

7.5.1.3 Complex pasts

At the beginning of this section on life before testing, I noted that in some ways this approach is in conflict with the tradition of the field. Existing discourse relates to what happens once testing is performed, the effects that such an experience has and the ways in which it impacts upon the lives of those tested. Life before testing is often overlooked. In some ways, such a tradition assumes that young people come to have predictive genetic testing with a ‘blank slate’. In other words, the test itself is the focal point, the causal factor of any changes that occur. The harms and benefits that are articulated in the literature are thus set against this blank background. An alternative way to view this conception of a blank slate is to use a numerical scale. In a numerical sense, the blank slate is equated with zero. Young people therefore present for testing at zero. The predictive genetic test is then performed and, following testing, harms contribute negative scores while benefits contribute positive scores.

It is this conception of a blank slate, or score of zero, that I wish to challenge here. I term this theme “complex pasts” as I feel that the phrases within it convey a life that is
far from blank, far from zero and highly complex. This third theme relates to the facets of life that combine to make the lives of young people at risk of a genetic condition different from the lives of young people who are not at risk of a genetic condition. The phrases I choose below are far from an exhaustive list. I choose them to demonstrate the range of ways in which the lives of young people at risk differ from the lives of other young people their age.

Growing up with family members who are sick is a part of life that several young people spoke of. Here, Poppy talks about the time when her father and step-mother explained to her what her mother had died from:

“I think it was more my step-mum, yeah, they were both there... cause I always thought it was cancer, I don’t know why, but yeah, they said oh your mum passed away from Huntingtons and there’s a good chance that you and your brother could have it. She died from HD when I was 6”

Poppy:F:24:HD:17:-ve

Poppy also speaks about the misunderstandings she had about HD when she was younger:

“When I first got told, I would have probably been about 12, when they first told me, I thought I was going to die cause they just came out and said, oh, that’s why your mum passed away, and being told I thought straight away, oh I’m going to die”

Poppy:F:24:HD:17:-ve

As does Belinda:

“From the little bit I knew I thought I was going to die, cause I just, I didn’t know anything about it really, I knew the name and I knew effectively that you eventually died from complications to do with it, but I didn’t know when or anything like that”


Ella talks of an uncle who committed suicide when she was very young:
“Yeah, he had a brother who found out he had the gene and committed suicide at 32… I was 2 years old so it’d be a good 18 years ago”

Ella:F:20:HD:18:-ve

She also speaks about her father’s experience with HD, as she was growing up:

“I was looking after him by myself, then it got too much for dad and he tried to commit suicide and jump off the roof and he broke his leg, quite bad, but, we brought him to hospital and he was just, he was really close, he was still talking and that but he, he was at that stage when, click, anything could happen, he was really getting restless and really needing smokes all the time, constant smoker, all HD people, smoke, smoke, smoke, smoke, so we needed someone else to come in”

Ella:F:20:HD:18:-ve

Nina describes the memories she has of her Aunty:

“I just remember the last, like the last year or something of her life and her dying and stuff”

Nina:F:23:HD:23:-ve

Doug also remembers his father being sick and in hospital:

“I reckon I was about 7, he was in hospital for about 2 weeks… he’s been pretty sick, he gets sick, he also has a tumour which pushes on his kidneys and makes him sick, and it’s sort of, the bowel cancer thing is not the main problem any more, it’s this tumour pushing on his kidney”

Doug:M:18:FAP:14:-ve

He goes on to talk of his dread of hospitals, which arose when his father said a heartfelt goodbye before entering hospital himself:
“I hate hospitals… ever since he, when we first moved here and he said he loved me and he missed me and all this stuff, sort of scared me off from going back to hospital ever since…cause um, yeah, the biggest change in my life was knowing that maybe my dad might not be around, he’s actually outlived, like they gave him 2 months, no they gave him 2 years when he was first diagnosed, and it’s now 14 years later so”

Doug:M:18:FAP:14:-ve

Harry remembers being confused about his father’s illness:

“I didn’t really understand it all, I just thought why’s he got it, why does he have to have all this done”

Harry:M:14:FAP:12:+ve

Travis also recalls having very little information until he was much older:

“One day mum and dad just sat me down and said look, mum’s got something called Huntingtons Disease and, and basically that was it, that’s all I knew, it was just mum had something and I didn’t know about, you know, the 50-50 passing down of the gene and that… I really didn’t sort of understand it until only a little bit before I got tested”

Travis:M:24:HD:20:-ve

Ali talks about first finding out about her risk status, and the secrecy that often surrounded the condition in her family:

“I always remember my dad or my mum or my sister talking…I was saying how did they die and my dad would always say they got really sick, you know, he didn’t want to tell me a lot, and then as I got older and probably, when I was like about 12 or 13, they sat me down and explained why they died and what it would mean for me”


Ella also recalls being aware of illness in her family from a very young age:
“I remember at the age of 3, sitting down, drawing a human being, trying to
work out some sort of cure, I remember that so clearly…every christmas
there used to be a thing for HD families”

Ella:F:20:HD:18:-ve

Troy remembers his grandmother being symptomatic:

“I sort of knew, because grandma, she had it and she was always sort of
jittery and sort of weird and stuff, yeah, so I knew there was something
wrong there, um, probably really started thinking about it, you know
realising what it really was when I was probably about 13 or so”

Troy:M:26:HD:25:-ve

Kylie again recalls having very little information at a young age:

“I knew that my grandma died of it and that my aunty and uncle had it, and
that was it”

Kylie:F:20:FAP:14:+ve

As does Zach:

“Dad tried to explain it to me but still didn’t, like, like he just told me he
had it and, and that I might get it as well”

Zach:M:26:HD:23:-ve

Belinda used to overhear conversations about the HD in her family, but only realised
the impact this had for her when she was sitting in her Biology class at school:
“Dad had the test, I was 12 I think…all I knew was that he came home and said that he had it to mum and he didn’t actually ever tell us…I overheard the original conversation, I knew there was something wrong with my grandma, but I didn’t know what it was, it didn’t have a name at the time, and then obviously somebody must have said what it was, and then I just overheard the conversation between mum and dad and then he came back with his test results, but as I said it didn’t really mean anything to me until 4 or 5 years later when I heard about genetics and, it just, the memory sprung back into the front of my mind and I knew then what he was talking about and how if affected me, but he never talked to us about it, he never directly told us what he had or anything about it so”


Troy describes the situation at home for his father at the moment, caring for his sick mother:

“It’s real hard on him, cause lately probably every 10 minutes my mum’s, she’s got this little bell thing and she’s always ringing that and he has to, you know, go and, she wants to go the toilet or, you know, wants coffees, and it’s sort of hard for him cause he can’t understand her sort of now, it’s getting bad”

**Troy:M:26:HD:25:-ve**

Oliver speaks of how long HD has been in his thoughts:

“I’ve thought about Huntingtons for 13 years, when your mother’s died of the disease, you’re absorbing information, especially at that age, you’re absorbing so much information, mentally, and just cataloguing it all up in your head”

**Oliver:M:24:HD:19:+ve**

For several young people, the desire to be tested before turning 18 brings added facets to their lives before testing. Nina relates her experience to a pressure cooker:

“It’s a big thing hanging over your head, you think about it more and like the pressure cooker, taken away or whatever, you’re not dealing with it, you’re just wondering”

**Nina:F:23:HD:23:-ve**
She recalls being frustrated by the fact that she had to wait until she was 18 years of age, noting that she would have liked to be tested after the birth of her daughter, Charlotte, at the age of 16:

“I remember ringing up when I was 17 trying to see if I could get the counselling done first, and then find out as soon as I turn 18, but no, you’ve got to wait until you’re 18 to even start the counselling process… it frustrated me…it was something that I really wanted to do too, cause it was like, I just wanted to, I don’t know why, just, I don’t know, find out so that I could set my, you know, plan my life ahead…I just wanted to get it over and done with, yeah, from when I was 16 cause I had Charlotte when I was 16 and I know from when she was born I wanted to know”

Nina:F:23:HD:23:-ve

Travis thinks he was 14 years old when he first wanted to be tested:

“I think I was about 14 when I first wanted to get the test so it would have been about, the start of high school when I knew that, you know, if I’m mum’s kid then I could end up getting it or not”

Travis:M:24:HD:20:-ve

He uses the metaphor of tattslotto to describe his experience of waiting to be tested:

“I mean, when you want to know something you want to know, it’s like tattslotto you know, if someone knows what the numbers are, and you don’t, you’re going to get pretty pissed off with them if they don’t tell you, you know”

Travis:M:24:HD:20:-ve

Ella talks in detail about her experience of wanting to be tested earlier than 18 years of age:
“I rang and she said no, you can only be 18, but there are some exceptions, if you’re pregnant, and ra ra ra, like, ideas that I thought were stupid, um, yeah, so, I went through this huge anguish, like sort of battle on myself, holding my breath more than ever, um, wondering if I’ve got this illness or not… You just want to know what way you want to direct your life, you know, how you’re going to work around, well, if you don’t have it, what you’re going to do, if you do have it, what you’re going to do, like, it’s such a big life choice, you know, and it’s sort of a ball, just this, ball game playing with you waiting til you’re 18, and I would have loved to get tested at 12”

Ella:F:20:HD:18:-ve

In describing her frustration at waiting to be tested, she refers to the absurdity she perceived in the fact that foetuses can be tested, but not teenagers:

“I wanted to crawl back up into my mum’s vagina just to get tested, you know, that’s how desperate I was, I wanted to be a foetus again”

Ella:F:20:HD:18:-ve

However, Oliver describes his desire to know as being fuelled by his natural curiosity:

“I was just curious I suppose, but you know, all the teachers and school counsellors and everything else are thrusting all these decisions upon you as you know and, you’re expected to decide and decide and decide and make the right decisions and everything else, I spose I just ah, I was always curious”

Oliver:M:24:HD:19:+ve

Thus for young people growing up at risk of a genetic condition, life is often very difficult. Sick parents, misunderstandings about genetic information that has been conveyed and sometimes a desire to be tested can all contribute to make life before testing highly complex.
7.5.1.4 Conclusions about life before testing

There has been little focus on the phase of life before testing in debates about predictive genetic testing in young people. The debate has largely been concerned with the consequences of testing and the time that follows disclosure of test results. However, several themes emerged during my interviews with young people related to this time before testing and highlighting the need to shift our focus slightly. Young people at risk of genetic conditions arrive for predictive genetic testing with a set of complex experiences and beliefs. These experiences and beliefs are different from those of other young people their age, who are not at risk of a genetic condition. Some young people may have engaged in, or be engaging in, an array of risk behaviours, some may have already begun preparing themselves for a gene-positive test result, and others may be coping with a sick parent or finding the uncertainty of their genetic status challenging to accept. It is against this background that the consequences of testing must be analysed.

The majority of quotes I have used in this section on life before testing are taken from interviews I conducted with young people who underwent predictive genetic testing for HD, as opposed to FAP. I come back to this point later in the chapter.

7.5.2 Life after testing

In this section concerning life after predictive genetic testing I highlight two broad themes that emerged during my interviews with young people: (1) Identity difficulties, and (2) Living again.

7.5.2.1 Identity difficulties

Existing literature concerning the harms of predictive genetic testing in young people focuses largely upon the harms associated with a gene-positive test result. It is intuitively logical to assume that this outcome is the one that warrants the most attention and has the greatest potential for harm. However, as I spoke with young people about their predictive genetic testing experience, it became apparent that two young people in particular who had received a gene-negative test result seemed to be having great difficulty adjusting to this knowledge. It seemed that for these individuals, finding out that they would not develop the genetic condition that runs in
their family brought unique troubles, different from those associated with a gene-positive test result.

Zach describes that in one way, receiving a gene-negative result felt like an anti-climax:

“I think, um, this may sound weird but it was almost a bit of a let down as well… it was kind of like… you’ve built yourself up for something and then it just doesn’t happen”

Zach:M:26:HD:23:-ve

He then notes how, after receiving his gene-negative test result, he felt quite flat and would have welcomed some contact with his genetic counsellor:

“I know that I was meant to be contacted in regards of going down again afterwards for like a post finding out session, but that never happened, I never got a call about that, which probably would have been good…um, to just say yeah I’ve been pretty down, like, or I’ve been pretty flat over the, for a month kind of thing and, just to be told yes, that’s normal would have been nice…it was almost like ok, well you’re done now”

Zach:M:26:HD:23:-ve

Ella also explains how lost she felt after receiving her gene-negative test result:

“It took me a long time to adjust, to go ok, just accept the test and start to live a life, I mean, I’m still doing that, it takes a long time to sort of realise, oh, you don’t have the gene, I’ve sort of got to pinch myself everyday you know, to realise, and now that I don’t have it, there is no connection too, so, like, I would love it if [the genetic counsellor] would contact me and ask me how I’m going cause I really don’t know what to do with myself, I feel really really lost, and I’ve got this life to live, you know, I sort of feel like I know what I could have done if I did get the gene, I have no idea what to do now that I don’t”

Ella:F:20:HD:18:-ve
Again she describes feeling confused about what to do from now on:

“It’s a really weird thing, like, I’ve got to think oh I can do these things, I’ve got this whole life to live… I’m truly clueless, I really feel like a newborn child, you know, like, really clueless with what to do, and I find that, like, I remember going down and seeing [the doctor] and he said you know, if you’ve got the gene, you know, blah blah blah, if you don’t, that’s fantastic, that’s it, it’s over, and I thought really? is it over?”

Ella:F:20:HD:18:-ve

Ella also speaks about her difficulty in believing her gene-positive test result:

“What do I do, what do I do now, you know, like, how do I gear myself up for life now, you know, like it’s a really weird, surreal sort of feeling… it took me forever when I got the results back to accept them, like, for a good 3, 4 months… take me into the lab and show me”

Ella:F:20:HD:18:-ve

She then speaks about trying to erase the belief she previously held about being gene-positive:

“Even to this day, it’s still sinking in… I’ve just given myself up to live a life now, you know, get rid of this old thought that’s just been in my head all my life and, I mean, it’s fantastic, it is, it’s fantastic, I mean I can’t be anything but happy about it, but yeah, it is very weird, I still haven’t, I’m in another desert you know, lost… but it’s good, I mean, it’s sort of a good thing I guess, of course, yeah”

Ella:F:20:HD:18:-ve

She also then speaks of her desire for more time with her genetic counsellor, now that she has received a gene-negative test result:
“I would have loved to keep catching up with her and trying to work out, ok, this is what you do from here, but I’ve been left to go off and work it out for myself, and I find that really hard, really frustrating, cause, what to do?”

Ella:F:20:HD:18:-ve

And notes that she would like to see the same for other people who receive a gene-negative test result:

“I also would like to see support of people that know they don’t have the gene, cause they’re still being affected by HD in their life, it doesn’t mean that cause they don’t have the gene, they don’t still get affected by the thought of HD everyday, like, I think about it constantly”

Ella:F:20:HD:18:-ve

Existing literature about adults who receive gene-negative test results has demonstrated similar themes. Some authors have suggested that people at risk of HD who have held a long-standing belief that they are gene-positive might experience difficulties in coping with a gene-negative result. Empirical findings have mirrored these suggestions in some cases. Meiser and Dunn, in their summary of the literature available about the psychological impact of predictive genetic testing for HD, note that one of their more surprising findings was that some people experienced difficulties following the receipt of a gene-negative test result. They explain that this may often be related to individuals having made irreversible decisions based on a belief that they would develop HD in the future. Tibben noted that some individuals receiving a gene-negative test result for HD had to adjust to the sudden removal of the ‘Huntington scenario’. Bloch and colleagues and Huggins and colleagues have also described how, in particular, those who receive a test result that is contradictory to the consciously or unconsciously expected outcome had difficulty in adjusting to their test result.

Wahlin and colleagues describe the case-study of a woman who received a gene-negative test result for HD. They describe how she had believed she had inherited the genetic mutation, prior to undergoing predictive testing. She held this belief even though she understood that, theoretically, there was a 50% chance that she had not inherited the mutation. Following receipt of the gene-negative test result, she reported having difficulties building up her new self image. She also described how she had
planned her life around getting HD. Williams and colleagues note that individuals receiving a gene-negative test result often have difficulties redefining themselves. They describe how adults undergoing predictive testing for neurodegenerative disorders prepared themselves for the worst outcome, but not for the possibility that their genetic test results may be normal. They also note that individuals often doubt the gene-negative test result and find it difficult to let go of the prior assumptions they held about being gene-positive. Michie and colleagues performed interviews with individuals who received gene-negative test results for FAP in an attempt to understand why 40% of individuals continue to seek bowel screening following gene-negative test results. They found that many of these individuals also distrust their genetic test result.

### 7.5.2.2 Living again

For several young people I spoke with, having the predictive genetic test allowed them to begin enjoying life more. Belinda describes how, before being tested, she felt as though she wasn’t really living and how having the predictive genetic test helped her to move forward:

“I was thinking god, 40 years from now I could be shaking and doing all this and that and I’m going to be dead sort of 20 years after that sort of thing and it stopped me from living at the time effectively, I mean, I did the day to day things, I went to school I did my homework I ate dinner, ate breakfast, did the things I needed to do, but didn’t really do much else, um, didn't feel like I was living kind of thing, like I had a life but I wasn't living, because I was just thinking about things that were too far ahead, so, once I had the knowledge, that was it, ok, fine, you know, I can’t do anything about it, and I realised that I guess, well I’m not in control of my life, but then you’ve got to realise that you’re not in control of your life, you can control certain aspects, you can control your decisions, but you can’t actually control your life, so and I guess that’s part of it, as I realised that and you just accept that if you can, I just accepted that and thought well I’ll get on with controlling my decisions that will affect my life and do what I want, so, it makes sense you know”

Again, she refers to the feeling that she could live her life after her predictive genetic test:

“Allowing me to live my life, knowing but not knowing in a way, knowing that if I live long enough, I will develop symptoms one day, um, but not knowing exactly what the rest of my life has in store for me and allowing me to accept that and just live, yeah”


Travis describes how, following his predictive test, he started doing what other young people his age were doing:

“I thought well, if I can go through this whole process of getting tested for this thing, I can pretty much do anything, except for quit smoking, so that’s, I respected myself a lot more for that, and um, I just started enjoying life a hell of a lot more, I started going out to clubs and stuff and you know, just doing what a normal person of that age would do… I didn’t go out much before that, I was a bit of a home body, just staying at home, watch videos and that was it really, so yeah, my social life really picked up”

**Travis:M:24:HD:20:-ve**

He also notes that it opened the way for him to discuss the possibility of children with his partner:

“[We] just starting talking more in depth about, about um having kids and, and just our future really, just our future as girlfriend or husband and wife, whatever it was going to be, um, but I think we just talked about the future a lot more, because I always had this, this thing in my head that I don’t want to plan things while not knowing, cause, when you plan something and something doesn’t go to plan you get, you know, you get ah, disappointed, so, um, that’s what sort of changed in our relationship, we just started talking about the future a lot more”

**Travis:M:24:HD:20:-ve**

Troy notes how he seems to have come out of his shell since his test:
“Since I’ve been tested I’ve been pretty good… I haven’t been in trouble with the police or anything so…I seem to have changed a bit, just come out of me shell a bit more and… you know, a bit happier and stuff…now I want to um, start my own business, so it clears away, you know, for the next few years, now I’m not going to get symptoms in my 30s or 40s or whatever”

Troy:M:26:HD:25:-ve

Nina also explained that she has stopped using speed since being tested, feeling uncertain as to whether this was motivated by her test result or not:

“I’ve gotten off drugs since I found out…I don’t know if subconsciously it was or what, but, yeah”

Nina:F:23:HD:23:-ve

Oliver speaks about his anxieties floating away upon receipt of his test result:

“Instead of umming and ahhing, am I going to be 60, am I going to be 40… a completely decisive answer, all the anxieties floated away, they just disappeared, off they went, rightio, this is it, it will start obviously shortly, deal with it and adjust”

Oliver:M:24:HD:19:+ve

Ella describes feeling blocked as a teenager when she was not able to learn of her genetic status:

“It’s just a big block… I used to say I feel like I’m holding my breath trying to climb a big wall… a huge big, big, big brick wall and my result’s on the other side, and that’s all it felt like, just this big block in the way”

Ella:F:20:HD:18:-ve

And how she felt that she could breathe for the first time after receiving her predictive genetic test result:
“It felt like I could breathe for the first time ever when I found that out, I didn’t care what the result was, I just wanted to know, I really, by that stage I did not care the outcome, I just wanted to know which way I can make my life”

Ella:F:20:HD:18:-ve

7.5.2.3 Conclusions about life after testing
Two key themes emerged about the phase of life after predictive genetic testing, during my interviews with young people who have experienced such tests. Firstly, there can be significant difficulties associated with the receipt of a gene-negative test result. Secondly, predictive genetic testing is capable of lifting a barrier that blocks some young people and prevents them from moving forward. Of critical importance here is the finding that both young people who receive a gene-negative test result and young people who receive a gene-positive test result have experienced this feeling of being able to finally move forward. Debates about predictive genetic testing in young people need to include not only considerations of the potential benefits and harms that such testing may cause, but also considerations about the pre-existing harms that testing may help to alleviate.

A pattern that began to appear in the section concerning life before testing has continued and expanded here. The experiences of young people who have undergone predictive genetic testing for HD have been the exclusive focus in this section on life after testing. It seems that, as themes emerged from the qualitative research process, the descriptions provided by young people who have experienced testing for HD became a central focus, and the descriptions provided by young people who have experienced testing for FAP faded into the background. This was not the case in the first phase of my thematic analysis, which involved an articulation of the benefits and harms associated with such testing. However, as I have moved from that first phase to this second phase of analysis, a shift has occurred in the emergent themes. I discuss this shift in detail in the final chapter.

7.5.2.4 Emergent themes related to young people specifically
As I have noted previously, when contemplating the current debate about predictive genetic testing in young people, it is important to bring the focus back to young people
specifically. I therefore ask the same two pertinent questions here that I asked at the end of my first phase of thematic analysis. These two questions are, firstly, which of the emergent themes articulated above could only occur in young people and not adults? Secondly, which of the emergent themes articulated above may have an increased magnitude in young people, when compared to adults?

Some of the emergent themes articulated above appear to be highly associated with young people specifically. The most obvious of these is the theme concerning risk behaviours, relating to life before testing. Behaviours such as leaving school, engaging in drug use and becoming pregnant at an early age are highly specific to young people. This is not to say that adults are not capable of also engaging in risk behaviours, but the types of behaviours described, particularly those relating to school and teenage pregnancies, are particularly salient for young people and what we already know about young people’s developmental processes and quest for independence. Another emergent theme that relates to young people specifically is the theme of a complex past. Although many adults who choose to undergo predictive genetic testing may also have complex pasts, there are particular aspects of this complexity that relate solely to young people. For example, the experience of wanting to be tested and being informed that this cannot occur until the age of majority. Also the impact that uncertainty has for young people is particularly salient to the decisions they need to make about their career and reproduction.

The emergent themes articulated in this section are perhaps more important when related to the second question posed above. That is, the magnitude of these themes may be increased for young people specifically. For example, if it is common for people to develop a belief that they are gene-positive in order to cope with the possibility that this may happen in the future, the potential for harm increases in magnitude as the length of time that the false belief is held increases. Thus, it could be argued that when young people undergo predictive genetic testing there is a greater potential for benefit, as their false beliefs can be removed at an earlier age than is possible when adults are tested. Similarly, if there is the potential that a gene-positive or gene-negative test result will help people to move forward and become free from existing barriers that have been blocking them, the earlier this happens, the greater the magnitude for benefit.
This second phase of thematic analysis, aimed at identifying broad themes relating to the experience of predictive genetic testing for young people, has therefore also demonstrated the emergence of themes that may have different significance for young people, when compared to adults. This difference is also discussed in detail in the final chapter.

7.6 Conclusion: Holding your breath

The title of both this chapter and this thesis is “holding your breath”. I borrow this phrase from Ella:

“All my life I thought I was going to get this illness, all my life, and last year I found out I didn’t, you know, so, for 19 years it feels like I’ve held my breath, thinking that I’m going to get this illness, and now it feels like I’m a new born child, you know, like, I can live a life that I never knew I could and it seems really weird to adjust to, it has been really weird, like, my testing was very very prolonged and the whole time I thought I was going to get it and then she told me I didn’t have the gene and, even then, you know, I wasn’t sure, I wanted even a second test you know, cause all my life I’ve just you know, been gearing myself up to have this illness”

Ella:F:20:HD:18:-ve

I choose this phrase because it encompasses the key themes I wish to emphasise in both this chapter and this thesis.

I believe we need to begin thinking about predictive genetic testing in young people differently. In the past we have failed to notice the way in which young people at risk are living before undergoing predictive genetic testing. We have neglected to understand this phase of their lives, focussing instead on the time after testing. We have subsequently missed an opportunity to understand what it is like to go through life holding your breath.
Offering young people predictive genetic tests is not about starting with a blank slate and pouring a list of potential harms and benefits onto it. It is about understanding the lives that are already occurring for the young people involved, lives that may be filled with risk behaviours, strong beliefs about genetic status, sick parents and misinformation, and assessing the way in which a predictive genetic test would look against this background.

Going through life holding your breath is presumably difficult. For some young people it constitutes a barrier in their lives and prevents them from moving on, from enjoying life. To group all young people together, to categorise all of their lives as similar enough to be addressed by one recommendation, is to neglect the complexity of each individual life and the different impacts that testing may have upon each of the individuals who live these lives.

Crucially, there is one particular group of young people for whom predictive genetic testing may be particularly pertinent. It seems that there exists a sub-group of young people, within the broader category of young people living at risk, who are growing up convincing themselves that they are gene-positive in order to cope with the possibility that this may in fact happen in the future. Of course, living as a gene-positive person can bring with it all the harms associated with real knowledge of a gene-positive status. In these cases, a predictive genetic test may do no more than either confirm an existing belief about gene-positive status, or remove existing harms by providing knowledge of a gene-negative status. This is but one example of the nuances that can make a predictive genetic test impact differently upon different young people.

In short, I am arguing that it is time to acknowledge the types of lives that young people live prior to requesting a predictive genetic test and to treat young people as individuals, not as one group. We need to begin thinking about predictive genetic testing in young people differently.
8.1 Introduction

In this final chapter I bring together past literature related to predictive genetic testing in young people and my own empirical research findings in order to draw conclusions about future clinical practice and research. I summarise what we now know about the impact of predictive genetic testing in young people and I describe how this differs from what we knew before. I propose explanations for the current lack of empirical evidence and suggest approaches to overcome these in the future. I then consider existing guidelines and argue for revisions to these, in light of my own empirical research. I conclude that current practice concerning predictive genetic testing in young people should alter.

Firstly, I begin with a brief reminder of the major conclusions contained within the previous 7 chapters.

8.2 Summary

For more than 15 years now a debate has been taking place about the appropriateness of predictive genetic testing for non-medical reasons in young people. An obvious feature of this debate has been the existence of vehement and opposing opinions. Less obvious has been the means required for moving through the current impasse that has been reached. Existing empirical evidence about the impacts of predictive genetic testing in young people is incapable of substantiating either support for, or opposition to, such testing. Current guidelines therefore rely solely upon a desire to maximise the best interests of young people, combined with a view that caution in the face of limited evidence is the best way to do this, in order to justify their stance. My aim in this thesis was not to bring closure to the existing clash in perception that is taking place, but rather to offer a new perspective about the issue of potential harm to mature young people who choose to be tested.

Young people differ from adults in psychological and social ways. It is therefore likely that predictive genetic testing will affect young people differently from the ways
which it affects adults. However, it is not clear whether these differences will confer a greater potential for harm or a greater potential for benefit if young people are tested. Thus, although it is reasonable to separate young people from adults in discourse concerning predictive genetic testing, there is a need to scrutinise arguments used to oppose such testing in young people in order to ensure that these arguments are in fact specific to young people themselves. The most powerful arguments used to oppose predictive genetic testing in young people are those that relate to the ways in which young people are psychologically and socially distinct from adults.

My international survey of clinical geneticists provided a glimpse into current practice. The findings indicated that predictive genetic testing in young people is occurring now and that, when such tests are provided, the impacts are rarely followed up. Clinicians’ reasons for providing and refusing tests are highly varied and are driven more by the nuances of individual cases than by any one ethical principle or set of guidelines.

When young people in my study talk about the impacts of predictive genetic tests they have experienced, they refer to the entire experience of being at risk of a genetic condition, not simply the time following receipt of their test result. An analysis aimed at identifying the benefits and harms of this process, although useful in providing a subjective account of these for the first time, falls short of reaching a deep understanding of the meaning young people ascribe to such tests. Much more valuable is a broad analysis of the themes that emerge within their stories, unbound by pre-existing categories or conceptions. When listening to young people relay their stories of predictive genetic testing it is best to refrain from creating the intuitively obvious distinction between gene-positive and gene-negative test recipients. Instead, a natural distinction emerges from within their stories, separating the phase of life before testing from the phase of life after testing. This alternative organisation allows the emergence of a new way of thinking about predictive genetic testing in young people. Young people growing up at risk of a genetic condition often suffer many harms prior to their presentation for predictive genetic testing. When we understand this, it becomes clear that in some cases such testing may serve to alleviate a currently harmful situation for young people, and in this sense may create benefit.
I now enter into a broad discussion of my research findings in relation to the current debate concerning predictive genetic testing in young people. I offer a detailed examination of what these findings mean for current practice, existing guidelines and future research.

### 8.3 Key findings

Here I describe the key findings of the empirical research I have conducted. I divide these into those relating to current practice and those relating to young people’s experiences of predictive genetic testing.

#### 8.3.1 Current practice

There were nine key findings that emerged from the international survey of clinical geneticists that I conducted:

1. At least 49 predictive genetic tests have been provided to young people for non-medical reasons, in spite of current recommendations opposing such practice.
2. It cannot be assumed that predictive genetic testing in young people for non-medical reasons will cease to take place until broad consensus is reached about its acceptability.
3. There is a desire for predictive genetic testing in young people. This is evident in both young people themselves and their parents.
4. Young people who make a decision to be tested themselves may experience adverse events following the receipt of their test result, regardless of whether this is a gene-positive or gene-negative test result.
5. Clinically significant parental anxiety may occur following a gene-positive test result in immature children. This relates to how and when to disclose the gene-positive status to the child.
6. Clinicians who responded to the survey refuse requests to perform predictive genetic testing for non-medical reasons in young people more often than they satisfy such requests.
(7) Consistent follow-up is not occurring in cases where such testing is performed.

(8) The majority of clinicians who responded to the survey agree with existing guidelines, but feel that each case needs to be assessed individually.

(9) There exists great variation in how clinicians evaluate requests for predictive genetic testing in young people.

Each of these key findings represents a new piece of knowledge concerning predictive genetic testing in young people. Although some of these pieces of knowledge may have been surmised previously, there was not evidence to substantiate such assumptions. These evidence-based findings therefore constitute new insights into what is happening currently.

8.3.2 Young people’s experiences of predictive genetic testing

Following my interviews with 18 young people who have experienced predictive genetic testing for either FAP or HD, several key findings emerged. These include:

- Growing up at risk of a genetic condition causes a range of harms that occur prior to and irrespective of the undertaking of a predictive genetic test.
- The range of harms and benefits that have been empirically researched in relation to predictive genetic testing in young people represent only a small fraction of the positive and negative aspects of the testing process that young people refer to when speaking about their experiences.
- Predictive genetic testing creates harms and benefits for young people that are different from those that it creates for adults.
- Young people sometimes feel unsupported by clinicians following the receipt of their test result.

In addition to these key findings, my research also provided a deeper understanding of young people’s experiences of predictive genetic testing. Knowledge has been gained about young people’s motivations for requesting testing, the way in which young people prepare themselves for receipt of their test result and the choices young people make about whom to share their experience of testing with. Knowledge has also been gained about the influence that testing has upon young people’s future career plans,
friendships, family relationships, partnerships, school life, reproductive choices and general approach to life.

From my analysis I have drawn several major hypotheses, which are worth further investigation. These hypotheses represent attempts to explain the patterns that have emerged from the data. Since the study is a qualitative one, its function is to produce new hypotheses, rather than test pre-formulated ones. The first major hypothesis was that young people who grow up at risk of a genetic condition might often develop a belief that they are gene-positive in order to cope with the fact that this may occur in the future. This hypothesis could be tested through empirical, quantitative means.

Another major hypothesis to emerge was that receiving a gene-negative test result might create identity difficulties for young people who have developed a belief that they are gene-positive. This hypothesis could also be tested through empirical means. Qualitative methods, aimed at gaining an understanding of aspects of young people’s self-identity both before and after testing, would be valuable.

Another hypothesis to emerge from my interviews with young people was that young people growing up at risk of a genetic condition might engage in risk behaviours at a higher frequency than young people who do not grow up at risk of a genetic condition. This hypothesis is testable empirically, through quantitative means.

Yet another hypothesis to emerge was that for young people who grow up at risk of a genetic condition believing they are gene-positive, a predictive genetic test may alleviate several of the harms that existed in their lives prior to their request for predictive genetic testing. This is testable through in-depth, qualitative means.

The final two major hypotheses to emerge from my interviews with young people were that, firstly, predictive genetic testing in young people under 18 years of age may create benefits and secondly, refusing to provide predictive genetic tests to young people under 18 years of age may cause harm. These are also testable empirically through both qualitative and quantitative means.
In the next section I take these key findings and hypotheses and consider the implications they have for current practice, existing guidelines and future research.

8.4 Implications for future research

In this section I consider four aspects of research into predictive genetic testing in young people. These are (1) the current lack of empirical research, (2) possibilities for future research methodologies, (3) the need to access young people who are not coping well, and (4) the value of using research concerning tests for medical reasons to inform debate about tests for non-medical reasons.

8.4.1 Overcoming the lack of empirical research

When considering what is required as part of future research efforts concerning predictive genetic testing in young people, there is a need to also consider why so little research has been performed thus far. In other words, without a comprehensive analysis of the factors that have led to such a lack of empirical research, we cannot hope to overcome such factors in the future. Calls for more research about the impact of predictive genetic testing in young people have been made repeatedly. The question is, if the desire for research is so strong, why does there continue to be such a lack of empirical evidence some 15 years after the debate initiated?

There are at least four possible reasons for the current lack of empirical evidence: (1) Predictive genetic testing in young people does not occur; (2) Such tests are generally one-off cases performed within a clinical setting and are therefore not assessed as they would be in a research study; (3) Clinicians involved in the provision of such tests choose not to research the outcomes because it places them in a vulnerable position; and (4) The absence of a single, leading group to coordinate such research inhibits research efforts. These reasons are elaborated below.
As I outlined in the previous section on current practice, predictive genetic tests in young people for non-medical reasons are rarely performed. It is more common for clinicians to refuse such tests than to provide them. However, this does not alter the fact that there is now evidence that such testing is occurring in several countries around the world. Therefore the current lack of empirical research into the outcomes of such tests cannot be justified by a lack of opportunity for research. Predictive genetic testing in young people is occurring.

Given that predictive genetic tests in young people remain rare, it is likely that most tests are isolated cases performed within the clinical setting and do not form part of a research study. This means that the only way outcomes of such testing can be studied is through an international, multi-centre collaboration. There is no such collaboration in existence and thus the current lack of empirical evidence may be partly due to this absence. Coordination of research about many individual tests across several international centres also requires a level of knowledge and interest in researching these outcomes. In order to involve clinicians in a common goal, clinicians must perceive a need for such research, be inspired enough to collaborate and have the resources to devote to the pursuit. Clinicians may only ever provide a predictive genetic test to a young person once during their career. It is a challenge to therefore reach these individuals before they even perceive a need to be reached.

Research into the impacts of predictive genetic testing in young people requires clinicians providing such tests to offer their decisions up for scrutiny, in the knowledge that they have acted in conflict with current recommendations. This position is a vulnerable one for clinicians and even with the strictest measures for ensuring anonymity, this may prevent clinicians from taking part in a research study and drawing attention to such tests when they are provided.

Finally, the absence of a single, leading group to coordinate such research provides an immense barrier to research concerning the outcomes of predictive genetic testing in young people. A leader is required so that, in the rare event that a clinician decides to test a young person for non-medical reasons, this is done in the knowledge that there is a need to research the outcomes, a protocol in place to do this and a group of expert researchers to provide guidance. Only in this way will informed, evidence-based
decisions about predictive genetic testing in young people for non-medical reasons be possible.

The current failure to research the outcomes of tests performed in young people and communicate the findings to the broader academic and clinical communities also has consequences for the quality of the services provided. If we have no record of when, why or how such tests were provided and no indication of the impact that such tests had for the young people involved, we have no means of tailoring the process in order to provide maximum benefit and minimum harm. Given that the provision of such tests to young people is controversial because it is perceived by some to have great potential for harm, this failure to monitor and improve the process is concerning.

8.4.2 Future research methodologies

In order to move forward in the quest for empirical research concerning the effects of predictive genetic testing in young people, a specific research protocol is required. That is, a means to carry out such research that can be shared by researchers internationally. There are several options for such a protocol and both qualitative and quantitative methodology should be employed. Qualitative research methods will minimise errors in initial assumptions about the likely impacts of such testing in young people, leaving room for new conceptions to emerge. My own interviews with young people have initiated this qualitative process, but more research is required. It is important that the outcomes of testing are explored widely initially in order to learn of the range of ways in which such testing may affect young people and to generate a variety of hypotheses about these effects. Potential qualitative research methods include in-depth interviews with young people who have undergone predictive genetic tests, surveys incorporating open-ended questions, and the provision of a standard format for clinicians to write up detailed case-studies. Young people who undergo predictive genetic tests should be consulted both before and several times after testing.

Standardised quantitative measures should also be utilised. These could incorporate questionnaires specifically concerning predictive genetic testing or take advantage of already existing, validated psychological instruments or utilise a combination of both. The qualitative phase should inform the specific outcomes that are measured by
quantitative methodologies. For example, the range of harms and benefits that have emerged through my interviews with young people can be used to direct more large-scale quantitative methods of research. Similarly, the hypotheses that were generated through these interviews can be used to design both qualitative and quantitative research in the future. These quantitative measures should also be administered both before and several times after testing.

Both qualitative and quantitative research methods used to assess the impact of testing in young people must include both harmful and beneficial outcome measures. Until now, there has been a trend to measure mainly the harmful consequences of predictive genetic testing in young people, in order to either support or refute assertions about potential harm in current literature. If a balanced understanding of the ways in which a predictive genetic test may influence the life of a young person is to be achieved a range of both positive and negative outcomes need to be sought.

In addition to these measures aimed at assessing the impacts of predictive genetic testing in young people, my own research has identified a need to assess the lives that young people live prior to their predictive genetic test. Young people who are growing up at risk of a genetic condition need to be invited to participate in research that utilises the same methodologies outlined above, but that specifically focuses on the types of harms that exist prior to predictive genetic testing. Importantly, such research bypasses many of the factors that may have been limiting past research efforts, such as a low number of young people undergoing such tests and clinicians feeling vulnerable about researching the outcomes of their decisions to provide testing.

**8.4.3 Accessing young people who do not cope well**

During my interviews with young people I visited a home where three sisters who had undergone predictive genetic testing for FAP lived with their parents. Two of these sisters had agreed to participate in an interview with me, but the third sister had declined. During my interviews with these girls it became apparent that their sister who had declined to participate in an interview was the only sibling to have received a gene-positive test result. It also became apparent that she had experienced serious difficulties coming to terms with this information. Her sisters explained that they were
not at all surprised by her decision to decline my invitation to participate in an interview as she “hates anyone talking about it”. We must remain mindful of the fact that individuals such as this young woman are often the ones who we consistently fail to reach through traditional research methods. If future research is to be balanced and rigorous, these difficulties must at least be acknowledged and ideally be overcome. The initial step in this process should entail an attempt to survey young people who decline to participate in research concerning the impacts of predictive genetic testing. For example, it may be that although young people within this group do not feel comfortable participating in in-depth interviews, they are willing to complete an anonymous survey.

In order to target young people specifically who do not cope well with the receipt of their predictive genetic test result, several means must be employed. Invitations sent to young people should explicitly describe our need for learning of the worst experiences of predictive genetic testing specifically. Such invitations must also normalise these types of reactions, explaining that many people find receipt of their predictive genetic test result difficult. Research methodologies must be designed specifically for this group and must be non-threatening. Possibilities include anonymous open ended surveys, an on-line forum where young people can hear of others difficulties as well as documenting their own, and written forms of qualitative research such as a series of letters or e-mails that are written between researchers and participants.

Any research methodology specifically targeting a group of young people who are not coping well with the receipt of their genetic test result must incorporate an offer of expert counselling at no cost.

Research that aims at assessing the lives of young people prior to their predictive genetic test may be prove particularly useful in understanding why some young people find coping more difficult following the receipt of their genetic test results. Ideally, research should ultimately be aimed at identifying this group of young people prior to their undertaking of predictive genetic testing.
8.4.4 Medical and non-medical tests

As part of my own empirical research I made a decision to combine the experiences of young people who were tested for FAP (a test performed for medical reasons) with the experiences of young people who were tested for HD (a test performed for non-medical reasons). I did this despite the fact that my primary focus was predictive genetic testing for non-medical reasons. My justification was based on an assumption that the existence of a medical benefit would not negate the potential for harm when mature young people undergo predictive genetic tests, just as the absence of a medical benefit would not negate the possibility for benefit. However, as I noted in the previous chapter, the more in-depth my thematic analysis became, the less I utilised descriptions provided by young people who were tested for FAP and the more I utilised descriptions provided by young people who were tested for HD.

The experience of predictive genetic testing for FAP is often heavily influenced by the medial surveillance that accompanies a gene-positive test result (or that ceases to be required following a gene-negative test result). Thus, young people’s descriptions of their experiences of testing for FAP often focus upon the positive and negative aspects of the provision (or absence) of medical surveillance such as colonoscopies and surgery. In other words, for young people who undergo predictive genetic testing for medical reasons, the meaning they ascribe to the test is often centred around this medical outcome. It is therefore fundamentally different from the experiences of young people who undergo tests for non-medical reasons. It is not the range of benefits and harms that alters in these cases, but rather the young people’s focus within this range. For example, in the face of a gene-positive test result for FAP, a major benefit articulated by young people was that they would be able to prevent cancer from developing. This, understandably, often overshadowed other benefits such as relief about knowing their genetic status or feeling able to plan life more effectively.

In debating the provision of predictive genetic tests to young people for non-medical reasons we must be mindful of the limitations of using empirical evidence relating to tests provided for medical reasons in order to draw conclusions about tests provided for non-medical reasons. The outcomes of predictive genetic testing for FAP constitute the largest body of empirical research related to such testing in young people. My
research makes it clear that this research can assist in answering some of the questions related to predictive genetic testing in young people for non-medical reasons, but that there are serious limitations to these.

8.5 Implications for existing guidelines and current practice

Guidelines concerning predictive genetic testing in young people have existed for more than 15 years. Within these published recommendations there is general consensus that, where medical benefit is not an outcome of such testing, testing should be deferred until young people are autonomous. As I have shown, the published empirical evidence available about the impacts of predictive genetic testing in young people is not strong enough to justify such prohibition. Authors of existing guidelines therefore justify their recommendations with the assertion that caution in the face of limited evidence provides the best means of maximising young people’s best interests. I argue that, in light of my own empirical research findings, such a cautious approach may sometimes do more harm than it aims to prevent. I suggest that current guidelines should become less cautious, and more open to decisions based in individual circumstances. There are five specific changes that I recommend.

Firstly, guidelines should not dissuade clinicians from providing predictive genetic tests to mature young people who request testing themselves. There is not sufficient evidence to support such a recommendation. Instead, guidelines should encourage clinicians to make case-sensitive decisions about mature young people’s requests for testing. Guidelines should also encourage clinicians to learn about the existing aspects of young people’s lives at the time they present for testing and to compare the potential impacts of a predictive genetic test with these pre-existing facets of their lives. Predictive genetic testing not only has the potential to create harms and benefits in the lives of young people, but also to alleviate existing harms in their lives.

Secondly, guidelines should refer to a broader range of potential outcomes of predictive genetic testing in young people. Existing guidelines present only a narrow range of benefits and harms. Guidelines concerning predictive genetic testing in young
people should provide clinicians with a detailed understanding of the issues involved and an insight into the potential impacts such testing may have. A wider range of potential outcomes of such testing should therefore be incorporated.

Thirdly, guidelines should provide a more balanced account of the potential outcomes that predictive genetic testing in young people may have, including the many non-medical benefits that are possible. Existing guidelines focus heavily upon the potential harms of such testing but devote little space to a delineation of the potential non-medical benefits. This is biased and should change.

Fourthly, guidelines should articulate the severe lack of empirical evidence currently available about the impacts of predictive genetic testing in young people. Guidelines should inform clinicians of the importance of systematic follow-up in cases where they choose to provide such tests and also educate them about the need for this to be both pre-test and post-test follow-up. Guidelines should encourage broad communication about cases in which predictive genetic tests are provided to young people.

Finally, guidelines should advise clinicians about how best to provide predictive genetic tests to young people, when they choose to do so. Young people are different from adults. They have different communication styles, different social networks, different home environments, different motivations and sometimes, different capabilities for reasoning. Guidelines should offer guidance to clinicians about talking with young people, counselling them, relating to them, listening to them and educating them.

I refrain here from drawing conclusions related to immature young people specifically as my primary focus in this thesis was the issue of potential harm to mature young people who choose to undergo predictive genetic testing for non-medical reasons. The harms and benefits associated with such testing in immature young people are different and require independent research. The issue of (future) autonomy also holds particular significance to this younger group.
I now come to the final section of both this chapter and this thesis. Here, I discuss the implications of my own empirical research for the ongoing ethical debate about predictive genetic testing in young people.

8.6 Implications for the ethical debate

The outcomes of predictive genetic testing in adults for non-medical reasons have been thoroughly studied. Where testing is performed in the setting of a protocol with pre-test and post-test counselling, there have not been the rates of adverse consequences that some professionals predicted would occur prior to the availability of such testing. Overall, the empirical evidence about predictive genetic testing in adults for non-medical reasons demonstrates largely beneficial experiences for those involved. The available evidence does not indicate that, for those who seek testing and who receive counselling as part of this process, the testing process is generally harmful.

Prior to performing my own empirical research concerning predictive genetic testing in young people, several certainties existed. There existed strong theoretical arguments to suggest that some young people may benefit from predictive genetic testing for non-medical reasons. There was extensive empirical evidence demonstrating that when adults undergo similar testing it is generally a beneficial process for them. There was no empirical evidence to suggest that if young people are provided with predictive genetic tests for non-medical reasons there will be greater potential for harm than when adults are provided with such tests. There was also some initial empirical evidence to suggest that young people who undergo predictive genetic testing for medical reasons cope well with the process. Despite the evidence, guidelines continued to advise against predictive genetic testing in young people for non-medical reasons, unless circumstances were extraordinary. This was justified by a conservative stance in the face of minimal evidence.

Following the empirical research that I have performed, several additional findings have emerged. There is now empirical evidence to demonstrate that some young
people have a strong desire to undergo predictive genetic testing for non-medical reasons before they reach the age of 18 years. There is also empirical evidence to demonstrate that clinicians sometimes choose to provide predictive genetic tests to young people for non-medical reasons, even though this conflicts with current recommendations. Most importantly though, there is empirical evidence to suggest that predictive genetic testing in young people can be beneficial, rewarding, empowering, appropriate and capable of helping young people to move forward in their lives. There is also evidence demonstrating that predictive genetic testing in young people can be difficult, stressful; that it can make school challenging and cause young people to feel lost. This empirical evidence provides an insight, for the first time, into what it is like for young people to experience predictive genetic tests for non-medical reasons. It shows us that the impacts of such testing for young people are different from the impacts of such testing for adults. However, different does not equate to worse.

There is also now empirical evidence to provide an insight into what the experience of waiting for a predictive genetic test is like for young people who are informed that they must be 18 years of age before their request can be satisfied. Young people often find this experience frustrating, disempowering and belittling. It can make them feel angry, isolated, blocked and unable to proceed through the tasks of adolescence. It may also fuel their engagement in risk behaviours.

In addition to this new empirical evidence about the effects of predictive genetic testing in young people, we now have empirical evidence about what life can be like for young people prior to their request for predictive genetic testing. We are now able to understand how difficult life can be growing up at risk of a genetic condition and the range of harms that may exist within young people’s lives prior to their request for predictive genetic testing.

The desire our society has to protect young people from harm is strong. This is appropriate. Accordingly, making an argument for the commission of an act when the consequence may be harm to a young person requires much weight. Much more weight, it may seem, than an argument for the omission of that same act. However, the trouble with such logic is that the omission of an act also has potential for harm. Omissions can create harm overtly and can also prevent benefits. Deciding to refrain
from offering predictive genetic tests to young people initially, unless it was medically necessary, was a logical start. It was based on a desire to protect young people, combined with a belief that not providing testing would always have less potential for harm than providing testing. It was the appropriate way to begin, until more empirical evidence became available. It is no longer appropriate, logical or ethical.

Offering predictive genetic tests for non-medical reasons to young people who request testing has potential for harm. But it also has great potential for benefit. Importantly, predictive genetic testing in young people offers the possibility of alleviating some of the already existing harms that occur in the lives of young people living at risk of a genetic condition. These possibilities for harm and benefit are different from those that are possible when adults undergo similar testing, but they are not necessarily worse. There is no longer adequate justification for refusing to allow mature young people to undergo such testing when we routinely offer it to adults. In current debates and existing guidelines, the impact of refusing to provide young people with predictive genetic tests and the harm that this may create is critically absent.

Young people who request predictive genetic testing for non-medical reasons should be considered as appropriate potential candidates for such testing. Clinicians should be encouraged to consider such requests seriously, yet educated about the ways in which such testing may impact differently upon young people from the ways in which it impacts upon adults. Clinicians should not be encouraged to provide tests to any young person presenting to them, but rather to engage in pre-test evaluation in order to determine if testing is appropriate in each case. Tests that are provided should be offered as part of a formal protocol incorporating pre-test and post-test counselling. Ideally, all cases where testing is provided should be followed-up systematically, and empirical evidence should be constantly reviewed, with the possibility of ceasing the provision of such testing should evidence support this action.

Some mature young people at risk of a genetic condition are going through life wondering what their future will entail, feeling blocked and holding their breath. For young people within this group who request predictive genetic testing, providing a test within a formal counselling protocol may be the most ethical way we can possibly proceed.
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Appendix A: Interview Theme List

1. **Family Life**
   - Pedigree

2. **Education and Employment History**
   - What can you tell me about your education – where you went to school, how long you were you at school, any further education?
   - And what about work –are you working at the moment?
   - If so, is this what you’ve always done?
   - What other sorts of jobs have you had, if any?

3. **Spare Time**
   - What sorts of things do you like doing in your spare time?
   - Who do you do these with?
   - Who are the important people in your life – Friends? Family? Partner?

4. **HD – knowledge and understanding**
   - What have you seen of HD?
   - Where did you learn about HD?

5. **Your Family’s experience of HD**
   - How do your family talk about HD? (Do they ever talk about it?)
   - Is it often the same person who brings it up?
   - When people in your family talk about HD, how do they seem?
   - Do you think different members of your family feel differently about HD?
     What make you say that?

6. **Genetics – knowledge and understanding**
   - What have you heard about how HD can be passed on through families?
   - What does ‘genetics’ mean to you?
   - Do you think of the test for HD as a genetic test, or just a blood test like any other?

7. **Your experience of HD testing – immediately before and after**
   - What had you seen of HD at the time you first made contact with the counsellor?
   - Can you remember how you felt when you first came in to talk about predictive testing?
   - Can you tell me a bit about the process that led you to decide to have the predictive test?
   - Was your decision to have the predictive test influenced by anyone else in your family?
   - When did you have the actual blood test?
   - Can you remember how you felt the day you came in for the blood test?
   - Do you remember having any feelings about how the result might go?
8. Other people’s reactions
   - Who else knew that you were going to have the predictive test?
   - How were they leading up to the time of the blood test?
   - Who knows of your predictive test result?
   - What were their reactions like?

9. Outcomes of testing – the bigger picture
   - Now I’d like you to think about how you felt a year before the test, and compare that to how you feel now. I’m wanting to look at the impact of the test on all the different parts of your life.
   - Are there any major differences in your relationships with friends or partners? Can you give me an example or a story?
   - Are there any major differences in the way you spend your spare time? Any examples?
   - Are there any big changes in your family life and family relationships? What sort of changes?
   - Are there any major differences in your work life? Can you give me an example?
   - Are there any big differences in your feelings about yourself?
   - Have any of your plans changed about your future?
   - Are there any big differences in how much you see your Dr/ GP/ Specialist or any types of medial check-ups?
   - Why do you think the HD test has resulted in these changes?
   - Overall, what was the best thing about having the predictive test for HD?
   - What was the worst thing?
   - If you could go back in time to the period leading up to your blood test, would you have done anything differently?
   - What about the time immediately after you received your results, would you have done anything differently then?
   - Is there anything you would have liked other people to have done differently throughout your testing process?
10. Other people and genetic tests
- If you knew someone who was contemplating a predictive test for HD, what would you say to them?

11. Testing of people under 18 years of age
- If the HD predictive test had been available to you as a teenager under 18, would you have liked to have it earlier than you did?
- What do you think would have been the good and bad parts about having the predictive test as a teenager under 18?
- Do you think people under the age of 18 should be able to have the predictive test for HD?
- What do you think about parents being able to test their very young children for HD, for example under 5 years? What about 5 – 10 years?

12. Other medium
- I’d like you to do one last thing please. I’ve got some paper and coloured pencils here and I’d like you to draw a person with HD for me. Once you’ve finished, I’ll get you to explain what you’ve drawn to me. I’m going to leave you to do that for about 5 – 10 minutes. It’s not meant to be a work of art – just a quick sketch OK?

13. Anything Else?
- Is there anything else that you think I should know about your experience of testing for HD or just about the issue in general?

14. Closure
- Here are my contact details. You are welcome to call me to talk about our interview today or anything else related to what we’ve talked about. Some people find that doing an interview like this can stir up some feelings that they hadn’t felt before, or hadn’t felt for a long time. If you find that this happens and you’d like to meet with one of our genetic counsellors to have a chat about it, I can arrange this for you easily, so please just let me know.
- Thank you for helping us with our project. Here’s a double movie pass for you to say thanks.
Appendix B: CD Script

CLIP 1: INTRODUCTION

1. Hi. My name’s Rony and I’ve sent you this CD to tell you about an incredibly important research project that we’re conducting here at the Royal Children’s Hospital.

2. Our research project is about young people and genetic testing. We’re hoping to talk to about 20 people who have had genetic tests about what the experience was like for them.

3. Our project is being run not only by the Royal Children’s Hospital, but also by Genetic Health Services Victoria. And we’d like to invite you to take part!

CLIP 2: WHO’S INVOLVED?

4. The research project forms part of my studies at the Hospital, and there are also two other people involved. The first person is Martin Delatycki – he’s a medical geneticist at the Genetic Health Services Victoria. And the second person is Lynn Gillam – she’s a lecturer at the University of Melbourne.

CLIP 3: WHY YOU?

5. We’ve contacted you about our project because our records show that sometime over the past few years, you had a genetic test here at Genetic Health Services Victoria, which is at the Royal Children’s Hospital. And, what’s more, you had the test when you were under 18 years old.

6. So, given that we want to know what genetic testing is like for young people, we think you’d be the best person to talk to about it!

CLIP 4: WHAT WOULD YOU NEED TO DO?

7. So, what will you actually need to do, if you decide to take part in our project? Well, you’d need to be interviewed, by me, for about an hour. The interview can happen anywhere that suits you – so this might be at your home, or it might even be here at the Royal Children’s Hospital. Whatever is easiest for you.

8. During the interview, I’ll ask you questions about your genetic test. These will be things like “What was the hardest part about having the test” and “What advice would you give to other young people having the test?” I’ll also tape record the interview so that I don’t have to take many notes. Later on I’ll type up the interviews, using the tape.
CLIP 5: CONFIDENTIALITY

9. I also need to tell you that everything we talk about during the interview will be completely private. Unless it’s required by law, the only people who’ll get to hear the interview, apart from you and me, will be Martin and Lynn – the other two researchers. This means that no-one else will have access to the recorded interview – not your parents and not your doctor.

10. And if we talk or write about the results of our study, we won’t use anyone’s names.

11. The tapes with the interview on them will be kept in a locked cabinet at the Royal Childrens Hospital, and the typed up interviews will all be kept on a computer in the same office, that is protected with a password. Both the tapes and the typed up interviews will be kept for 7 years and then destroyed.

CLIP 6: PROS AND CONS

12. I understand that we’re asking you to give up an hour of your time just to talk to me, and that you don’t really get any direct benefit from doing this. But the information you can give us will be so important in making decisions about genetic testing in the future. The more we know about people’s thoughts about the tests they’ve had, the better we can make testing for other young people like you in the future.

13. The other thing is that no-one has ever done a research project talking to young people themselves about this issue before – only to their parents or their doctors. So this is your chance to tell us what you think.

14. Sometimes people find that doing an interview like this can raise feelings that they hadn’t thought about in a while. If this happens to you and you’d like to talk about these some more after the interview, we can easily arrange a meeting with a genetic counsellor for you as well.

CLIP 7: WILL I FIND OUT ABOUT THE RESULTS?

15. If you decide to take part in our research project, we’ll send you out a letter when it’s finished to tell you what we found. The project might not be finished for about a year though.

CLIP 8: CONCLUSION

16. Thanks for listening! If you are interested in taking part in our project, all you need to do is get in contact with me – you can do this by sending me a text-message, emailing me, or calling me. All my details are on the sheet of information that I sent you with this CD. I hope to hear from you! Thanks!
Appendix C: Letter of invitation

Royal Children’s Hospital, Melbourne

Dear __________

My name is Rony Duncan. I am a PhD student at the Murdoch Childrens Research Institute and I have asked the staff at Genetic Health Services Victoria to send this letter to you. Please note that your details have not been given to me and the only way I will have any details about you is if you contact me yourself.

I would like to invite you to take part in an important research project that we are conducting at the Royal Children’s Hospital. Our project is about young adults and genetic testing. We are hoping to talk to at least 10 young adults (between the ages of 18 and 25) who have had a genetic test for Huntington Disease, so that we can find out what the experience was like for them. I am hoping you might be interested in participating.

I have included some written information about the project with this letter. Once you know a bit more about our project, if you are interested in taking part, all you need to do is get in contact with me. You can do this by:

- Giving me a call at work on 8341 6245, or
- Calling or text-messaging my mobile on 0422 838 122, or
- Sending me an e-mail at rony.duncan@mcri.edu.au

Thanks,

Rony Duncan B.SC (Hons)
You are invited to participate in a Research Project that is explained below. Thank you for taking the time to read this Information Statement. It is 6 pages long. Please make sure you have all the pages.

For people who speak languages other than English:
If you would also like information about the research and the consent form in your language, please ask the person explaining this project to you.
What is an Information Statement?

These pages contain information about a research project we are inviting you to take part in. The purpose of this information is to explain to you clearly and openly all the steps and procedures of this project. The information is to help you to decide whether or not you would like to take part in the research.

Title of the Project

Predictive Genetic Testing in Young Adults

What is the Research Project about?

Our project is about genetic testing and young adults. We are hoping to talk to young adults (between the ages of 18 and 25) who have had a genetic test for Huntington Disease, about what the experience was like for them. We would also like to hear what these people think about genetic testing in people under the age of 18. At the moment there are only a few genetic tests that people under the age of 18 can have. Unfortunately, there has not been much research done in this area, and we really don't know how people who have had genetic testing feel about testing in children and adolescents. The opinions of people who have had genetic testing are critical in deciding what recommendations to make in the future.

As part of our project, we are hoping to interview at least 10 people (between the ages of 18 and 25) who have had a genetic test for Huntington Disease. The interviews should take about an hour and will be tape-recorded.

Who are the Researchers?

The people working on this project are:

- Rony Duncan. Rony is a PhD student at the Royal Children's Hospital and this research project forms part of her PhD project. Rony will be doing all the interviews.

- Associate Professor Martin Delatycki. Martin is a Medical Geneticist at Genetic Health Services Victoria. Martin supervises Rony's PhD.

- Dr Lynn Gillam. Lynn is a Lecturer in Ethics at the University of Melbourne. Lynn also supervises Rony's PhD.
Why am I being asked to be in this research project?

You are being asked to take part in our research project because you had a predictive genetic test at Genetic Health Services Victoria (at the Royal Children's Hospital) for Huntington Disease when you were between the ages of 18 and 25. There is no problem with the genetic test that you had or with the results of that test, we would simply like to talk to you about what the testing process was like. We are contacting all people who fit this category.

What do I need to do to be in this research project?

If you agree to take part in our project, Rony will interview you for about 1 hour. Rony will speak to you over the phone before the interview to work out a time that suits you. The interview will take place at the Royal Children's Hospital or at your home. The interview can be done during the day, during one evening or even on the weekend if you prefer. During the interview, Rony will have set questions that she will ask you but there will also be chances for you to talk about other parts of the genetic testing experience if you want. An example of a question Rony might ask you is “Can you tell me what the hardest part about having the test was?” Also, Rony will tape-record the interview so that she can type it up on a computer later. Although Rony will be the person who conducts the interview, there may also be another professional sitting in the room at the time. This person will be a clinical geneticist, a genetic counsellor or a case-worker. This person will not take part in the interview but will be an observer.

Is there likely to be a benefit to me?

There is no medical benefit to you for participating in our research project. We hope that you will appreciate the chance to have your views heard and to talk about your experience of genetic testing.

Is there likely to be a benefit to other people in the future?

We hope that the information we get from our interviews will be of great help to the people who make decisions about genetic testing in young people. The more information these professionals have about how genetic tests affect young people, the better decisions they can make about which tests to offer young people.
What are the possible risks and/or side effects?

The only risk is that you might get upset or distressed during the interview. This is because genetic testing and the topic of future disease can be sensitive issues. If you do become upset during the interview, Rony will stop the interview. You can then either stop for a while and then continue the interview, stop the interview and make another time to finish it, or stop the interview and decide not to take part in the project any more. Any one of these options is fine and it is completely up to you to decide. Rony has completed a course in counseling and she will be able to offer you support if you do become distressed. Another professional, such as a genetic counsellor or a clinical geneticist will also be available to offer support. Rony can also suggest some people for you to talk to at another time after the interview if you would like, including a qualified genetic counselor.

What are the possible discomforts and/or inconveniences?

The main inconvenience to you is the time that it will take to travel to and from the Royal Children's Hospital if the interview takes place there and the time it will take to do the interview. The interview should go for about 1 hour but this may change from person to person depending on how long you and Rony discuss each question.

What will be done to make sure the information is confidential?

All of the information that we collect about you during the interview will be kept completely private and the only people who will be allowed to see it will be the people working on this project (unless otherwise required by law). Any written information that we have about you will be kept in a locked filing cabinet at the Royal Children's Hospital and any information that we have on the computer will be protected with a password. The information we collect will be kept for 7 years and then it will be destroyed (deleted or shredded).

If we talk or write about the results of our project we will not use your real name. But there are not that many young people in Victoria who have undergone predictive genetic testing, especially for the more rare genetic conditions. Sometimes in the papers we write, we also give information about the age of the person and maybe even how many brothers or sisters he or she has. This means that even though we won’t use your name, someone who knows you (for example, one of the Doctors who has met you) might be able to work out who you are when we talk or write about the project. We will do our best to make it difficult for anyone to work out who we are talking about.

Will I be informed of the results when the research project is finished?

Yes. When our project is finished, we will send you out a letter that explains what we found. The letter we send you will not be about you individually; it will be about what we found about the whole group together. Our project may not be finished until the end of the year so you may not get this letter until then.
You can decide whether or not to take part in this research project.
You can decide whether or not you would like to withdraw at any time without explanation.

You may like to discuss participation in this research project with your family and with your doctor. You can ask for further information before deciding to take part.

If you would like more information about the study or if you need to contact a study representative in an emergency, the people to contact are:

**Name:** Ms Rony Duncan  
**Telephone number:** 8341 6245 (between 9am and 5pm, Monday to Friday)  
0422 838 122 (after hours or in an emergency)

**Name:** Dr Martin Delatycki  
**Telephone number:** 8341 6284 (between 9am and 5pm, Monday to Friday)  
9345 5522 (and ask for Dr Delatycki to be paged, in an emergency)

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**What are my rights as a Participant?**

1. I am informed that except where stated above, no information regarding my medical history will be released. This is subject to legal requirements.

2. I am informed that the results of any tests involving me will not be published so as to reveal my identity. This is subject to legal requirements.

3. The detail of the procedure proposed has also been explained to me. This includes how long it will take, how often the procedure will be performed and whether any discomfort will result.

4. It has also been explained that my involvement in the research may not be of any benefit to me personally. I understand that the purpose of this research project is to improve the quality of medical care in the future.

5. I have been asked if I would like to have a family member or a friend with me while the project is explained to me.

6. I understand that this project follows the guidelines of the National Statement on Ethical Conduct in Research Involving Humans (1999).

7. I understand that this research project has been approved by the Royal Children’s Hospital Ethics in Human Research Committee on behalf of Women’s and Children’s Health Board.

8. I have received a copy of this document.
Appendix E: Letter sent to parents of potential participants

Royal Children’s Hospital, Melbourne  Murdoch Childrens Research Institute

Dear _______________

My name is Rony Duncan. I am a PhD student at the Murdoch Children’s Research Institute and I have asked the staff at Genetic Health Services Victoria (GHSV) to send this letter to you. Please note that your details have not been given to me and the only way I will have any details about you is if you contact me.

I would like to tell you about an important project that we are conducting at the Royal Children’s Hospital. We are hoping your child, (name of child) might be interested in participating. Our project is about young people who have had genetic tests. We’re hoping to talk to young people between the ages of 12 and 25 who had a genetic test when they were under the age of 18. We’d like to find out what the experience was like for them.

We will be sending a letter out to (name of child) this week with some information about our project. (name of child) might like to discuss the project with you before deciding whether or not to take part. If (name of child) does decide to take part, one of you simply needs to get in contact with me. I have included some written information about our project with this letter for you as well. If you have any questions or comments, please feel free to get in touch with me. My contact details are as follows:

- Work phone number: 8341 6245
- E-mail address: rony.duncan@mcri.edu.au
- Mobile phone number: 0422 838 122

Warm regards,

Rony Duncan B.Sc (Hons)
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