

# "It was the missing piece": adolescent experiences of predictive genetic testing for adult-onset conditions

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**Purpose:** Predictive genetic testing for adult-onset conditions, such as Huntington disease, is available to at-risk adults. Guidelines consistently recommend against such testing before adulthood unless there is associated medical benefit. Guidelines, and related commentary, cite a range of potential harms that might be associated with young people undergoing testing, yet evidence is limited. Clinical practice has forged ahead, with research demonstrating that some clinicians are making their own determinations about the best interests of individual young people and are providing testing in specific cases. For the first time, this study reports empirical evidence concerning the process and impacts of predictive testing in mature minors for adult-onset conditions where no medical benefit exists.

**Methods:** First-hand accounts from nine young people have been documented. All were 16 or 17 years of age when tested. Six tested gene positive.

**Results:** Contrary to existing assumptions, the results convey a range of benefits and absence of harms flowing from testing. Some individuals reported considerable distress associated with institutional processes before testing.

**Conclusion:** The results highlight that young people from families affected by genetic conditions might possess task-specific competence relating to decision making about predictive testing. Further research and longer-term follow-up is required to study the outcomes of testing in young people.

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**Key Words:** adolescent; adult-onset condition; ethics; predictive genetic testing; qualitative research

Predictive genetic testing for adult-onset conditions, such as Huntington disease (HD) and some familial cancers, is available to adults at risk. Conversely, there is ongoing controversy with regard to testing young people, specifically those younger than 18 years, who are at risk for adult-onset genetic conditions for which there are no effective medical interventions available before adulthood.

International guidelines recommend that testing be deferred until an individual is competent to make an informed, autonomous decision regarding testing.<sup>1–5</sup> Recently revised guidelines specific to the testing of HD recommend that predictive testing not take place until an individual reaches the age of majority.<sup>6,7</sup> More than two decades since the publication of the first guidelines, considerable disagreement between commentators remains.<sup>8</sup> The ethical debate concerning predictive genetic testing in young people has become mired in a conflicting set of opinions, assumptions, and speculation, with little relevant evidence to inform these.<sup>9</sup> Two overarching concerns dominate the arguments against predictive genetic testing in young people: (i) young people lack the competence to comprehend the significance of a predictive genetic test and may regret their decision later in life, and (ii) young people who receive a gene-positive test result (the presence of the relevant family mutation) are at risk of adverse psychosocial consequences. This debate is situated within a context of knowledge that most adults at risk for HD choose not to undergo predictive genetic testing.<sup>10</sup>

There have been numerous studies reporting the effects of predictive testing for childhood-onset conditions such as familial adenomatous polyposis<sup>11–14</sup> and the effects of predictive testing in adults for adult-onset conditions.<sup>15–21</sup> Conversely, few studies have examined the impact of testing on young people, and none have specifically considered minors tested for adult-onset conditions. An international survey in 2005 showed that some clinicians are providing predictive tests to minors in particular circumstances;<sup>22</sup> however, the outcomes are poorly studied.

The current study aimed to (i) gauge the impact on young people of predictive testing before 18 years, (ii) identify factors that mediate the testing experience, and (iii) assess whether evidence exists to support the central concerns raised in the existing literature with respect to testing young people. The study addressed only self-initiated requests by adolescents, not young children.

## METHODS

Participants were recruited from two Australian states. At the time of data collection, 10 individuals had undergone predictive testing for an adult-onset condition before the age of 18 years in these two states. Nine agreed to participate. The tenth was unable to do so because he was traveling.

In one state, all the young people had instigated testing by requesting a test themselves. In the other state, some young people received a letter outlining testing options for them. In that state, predictive testing for familial cancers is offered from

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16 years at the discretion of a young person's clinical team. Participant details are presented in [Table 1](#).

The study was conducted with the approval of the human research ethics committees of the Royal Children's Hospital and Monash University, Melbourne, and the Women's and Children's Hospital, Adelaide.

Clinical staff identified individuals who fit the inclusion criteria and invited them to participate in the study on behalf of the research team. The research team then contacted interested individuals directly. Inclusion criterion was having undergone a predictive test when younger than 18 years of age for a condition that usually has onset in adulthood. Excluded were individuals who had undergone testing for a condition for which an intervention before adulthood is recommended (e.g., familial adenomatous polyposis). Participants provided written informed consent. Interviews were conducted between 2009 and 2011, mostly in participants' homes. Interviews were digitally recorded and transcribed verbatim. Transcripts were analyzed using thematic analysis.<sup>23</sup> Cocoding was used to enhance rigor.

## RESULTS

The themes that emerged from the interviews with young people are presented chronologically: (i) life before the test; (ii) the battle to be tested; and (iii) living with the knowledge.

### Life before the test

In exploring the lives of young people before embarking on the predictive testing journey, four themes were identified: (i) growing up in a family with a genetic condition, (ii) living at risk, (iii) desire for testing, and (iv) preconceived ideas regarding gene status.

*Growing up in a family with a genetic condition.* Many young people spoke of the impact of living in a family with a genetic condition and the significant emotional and practical day-to-day impacts of living with an unwell parent.

**Table 1** Demographic details of participants

Pseudonym	Gender	Condition tested for	Age at test (years)	Age at interview (years)	Result
Cate	Female	HD	16	17	Negative
Amelia	Female	HD	17	18	Negative
Sophie	Female	ADCA	16	18	Positive
Madeline	Female	HNPCC	16	18	Positive
Alice	Female	HNPCC	16	20	Positive
Emma	Female	BRCA1	16	20	Positive
Zoe	Female	BRCA1	16	21	Negative
Dylan	Male	BRCA1	16	20	Positive
Luke	Male	CDH1	17	18	Positive

Negative, gene negative; Positive, gene positive.

ADCA, autosomal-dominant cerebellar ataxia; BRCA1, predisposition to a number of cancers including breast and ovarian cancer, due to a mutation in BRCA1; CDH1, predisposition to a number of cancers, in particular stomach cancer, due to a mutation in CDH1; HD, Huntington disease; HNPCC, hereditary nonpolyposis colorectal cancer.

"It wasn't until she had brain tumors and lung cancer that she started spending less time at home; she was really sick, and when we would get sick her immunity was so low she couldn't be at home. I hated it.... I cared less about school, things just didn't seem important anymore."

Emma: (pseudonym); BRCA1(condition); 16 (age at test); +ve (result); 20 (age at interview)

"I found out my mum had HD when I was 11; my parents think it deprived me of a childhood... I had things to worry about that other kids didn't and I got depressed.... it forced me to grow up quicker."

Amelia: HD: 17: -ve: 18

*Living at risk.* Young people's at-risk status had a significant impact on their outlook and sense of self.

"It was an anxiety sort of feeling, not knowing... just not knowing was the worst I guess for me ... I was just wondering all the time whether I had it."

Emma: BRCA1: 16: +ve: 20

"... She said that your mum's got it and you have a chance of getting it as well.... That was the day the clock stopped; that was the day the uncertainty began."

Amelia: HD: 17: -ve: 18

*Desire for testing.* For many young people the decision to pursue testing was motivated by a pervasive sense of uncertainty regarding their future combined with the cumulative experiences of living with and observing a parent's suffering.

"I wanted to know straight away ... it was sort of like well, I might not need to worry, but I'm going to worry anyway. It was more the not knowing that was causing anxiety rather than the prospect of cancer itself."

Madeline: hereditary nonpolyposis colorectal cancer (HNPCC): 16: +ve: 18

"I needed to know before I could do anything with my life. It was a huge part of me; it's my mum, my grandma; it's where my whole childhood disappeared to ... I realized that I couldn't do anything until I knew who I was. Because of all of this I didn't know who I was; it was like something was missing, and I needed to know."

Cate: HD: 16: -ve: 17

"After I saw what my mum went through, I think I just wanted to know whether I would have to go through that as well. I know that a genetic test doesn't mean you're going to get it, but at least it prepares you knowing that it could happen, even if it could happen to anyone."

Emma: BRCA1: 16: +ve: 20

**Preconceived ideas regarding gene status.** Despite an intellectual comprehension of the 50% risk of inheriting the gene mutation, eight of the nine participants entered the testing process with a strong belief that they were gene positive.

For some participants, that presumption arose from an underlying identification with a parent.

“I felt like I was going to go down the same path as mum ... and the fact that it runs through the women, all the women in the family that we know of have had it, I sensed that I would be next.”

Cate: HD: 16: -ve: 17

“It was a mentality thing, thinking that you’re like your mother, or like I’m my mother’s daughter, so of course I have it; it’s genetic. I know genes come from both your mum and dad, but I wasn’t shocked by the result. I was upset, but I wasn’t shocked.”

Emma: BRCA1: 16: +ve: 20

For others, it was a coping strategy, preparation for the worst possible outcome.

“I just had this feeling .... I decided that if I felt like I had it then I wouldn’t get bad news, I would get news that I expected, or I’d get good news, but there wouldn’t be any bad news, something that would bring me down.”

Madeline: HNPCC: 16: +ve: 18

### The battle to be tested

There were varying reflections about the processes that each young person had to undergo before receiving a test. Many felt disempowered, perceiving that those with power over the decision had not made the effort to understand their motives.

“I thought the information was mine .... It was my issue; it was screwing up my life; it was my mother who had potentially given it to me; it was all about me; why shouldn’t I know? The thing that really got me was the fact that people who hadn’t experienced the disease, didn’t know me or my family or my situation were telling me that I wasn’t ready.”

Amelia: HD: 17: -ve: 18

“I thought it was pretty stupid that they wouldn’t let me; they don’t really know me or if I’d get depressed or not .... She just didn’t believe anything I said about how I felt; she was acting like she knew me better than I knew myself.”

Luke: CDH1: 17: +ve: 18

“There was a lot of emphasis on making it sound like how I was feeling was wrong; they almost felt like I should be feeling worried about it, I should be concerned, which I probably would’ve been if it was something that had only just been found out about and I hadn’t lived with it ... but

half my family had it. It’s the same as half of my family has brown eyes, half has blue eyes. It’s not like you need to go and see a counselor before you look in the mirror and see what color your eyes are. That is all it was to me; it was just another part of my genetic makeup.”

Alice: HNPCC: 16: +ve: 20

Some young people specifically commented on the negative impact of being made to wait.

“It was over 3 years; it was a long time to be angry. It was the waiting game, the maybe; I wanted the maybe to become yes or no; I was over maybe, even if it meant yes, it was something definite, it was something I knew, it was the end of maybe and that’s all I wanted .... It was really hard and I felt like it was unnecessary because it was just too painful all the time; I knew what I was in for, I’d watched my mum, I’d researched the crap out of it, I’d had my days where I woke up with Huntington’s and I knew it couldn’t get worse than that, even if it was positive.”

Amelia: HD: 17: -ve: 18

Amelia also described an occasion when she was asked by a clinician to imagine living with HD.

“She said “I want you to spend a week pretending that you have HD, just so you know what it’s like,” and I’m like “I have spent 3 years pretending I have HD, I don’t need to spend another week in this false reality.” It’s like they were thinking of things that I hadn’t thought of yet, and I’m like “when this is following you around like it had been following me, you have thought of everything””

Amelia: HD: 17: -ve: 18

### Living with the knowledge

In considering the impact of testing on young people, there was a distinction between immediate and longer-term effects.

**Immediate impacts of testing.** No young person reported a catastrophic emotional response to discovering his or her genetic test result. The most significant negative report was of continuing anxiety related to a gene-positive result.

“A bit sad that I had the gene, like I could get cancer at any time.”

Dylan: BRCA1: 16: +ve: 20

The young people who received a gene-negative test result reported mixed emotions.

“It was like everyone else was more excited than I was, but for me it was like I had been wrong all this time .... for the first few weeks I was in shock that I didn’t have it.”

Cate: HD: 16: -ve: 17

"She goes "you're negative," and time itself just stopped ... all of a sudden I burst into tears, happy and at the same time sad. I felt like I'd wasted all this time on something that wasn't even there .... I'd lost my childhood all because of something that didn't exist."

Amelia: HD: 17: -ve: 18

Those who tested gene positive were often unsurprised by their test result.

"I think they all expected me to really freak out ... they didn't understand my reaction. They thought it was such a big thing, but to me it was something that I had expected for a year and a bit, so it wasn't too big of a shock."

Madeline: HNPCC: 16: +ve: 18

"It kept me on the same direction I was already going. If I hadn't of had it then it would've changed my thinking .... But I was in the same position I had always been in, except there was now a confirmation that, yes, what I was thinking was right."

Alice: HNPCC: 16: +ve: 20

Gene positive or gene negative, many young people recalled responding positively upon receiving their test result.

"I felt like I was a little more complete after finding out because I actually knew, knew the answer to something that I'd assumed about myself but wasn't sure."

Alice: HNPCC: 16: +ve: 20

"She just said the result came back positive and I was like "okay." I didn't cry. Dad was upset, Dad cried. He thought it was crap, but I was just happy that I knew."

Emma: *BRCA1*: 16: +ve: 20

**Longer-term impacts of testing.** Beyond the moment of revelation, many of the young people interviewed described their test result as having an ongoing impact on their outlook and approach to life.

For some young people, discovering their gene-positive status prompted lifestyle changes.

"I look at life a bit differently. I want to get uni over and done with; I want everything to be done quickly, just in case something happens. I want to have kids young; I want to get married young, just in case."

Madeline: HNPCC: 16: +ve: 18

Some young people described certain reminders as triggers for focusing their thoughts on their risk of future illness.

"You hear about certain people in your family getting it [cancer] and I think, you know, any time that could be me."

Dylan: *BRCA*: 16: +ve: 20

Others were worried by the possibility of passing the gene mutation on to their own future children.

"I want to have kids young, but they've suggested, just to be safe, to use IVF to find ones that don't have the genetic mutations, so I don't pass it on to my kids. I'll do that because I would feel bad, like I know my dad feels guilty."

Madeline: HNPCC: 16: +ve: 18

"I've always wanted children and obviously having the gene makes it likely that I might pass it on to some of my children .... We have always talked about it in my family as being a good thing, but you know, I can't have the guarantee that if I were to have children that they would think about it the same way."

Alice: HNPCC: 16: +ve: 20

No young person expressed regret regarding the decision to undertake testing.

"I was really certain that I wanted the test, even though it was a negative, a positive result, it didn't change the fact that I wanted it .... I definitely didn't regret getting it done."

Emma: *BRCA1*: 16: +ve: 20

"Now when I see my mum I look at her and I still get upset, but I know that's not gonna be me .... I feel like it was the missing piece, I'm now complete."

Amelia: HD: 17: -ve: 18

## DISCUSSION

This study sheds light on the experiences of young people who request a predictive genetic test for an adult-onset condition for which there is no available preventative intervention before adulthood. As well as considerations of the predictive testing journey and outcomes, young people have provided insight into living at risk. The influence of prior experiences (life before testing) on the decision to pursue testing and on experiences after testing is a key finding.

With a few exceptions,<sup>21,24–26</sup> commentary concerning testing in young people has focused on potential consequences for the individual, each considered as if they were a blank slate, with little consideration of the influence of prior lived experiences in determining areas of vulnerability or resilience. An important message from the young people in this study is that an understanding of the potential impact of testing on an at-risk young person begins with an effort to understand the experiences and motivations of that individual. Although the implications of a predictive test for a neurogenetic condition such as HD (for which no preventive treatment is available) differs from a test for a familial cancer syndrome such as *BRCA1* (where



preventive intervention exists), remarkable consistency was found in the themes that emerged from the interviews irrespective of the condition tested for.

The significance of previous experiences, or experiential knowledge, has gained recognition in the literature relating to perceptions of risk by individuals from families affected by hereditary cancers.<sup>27</sup> Abel and Browner<sup>28</sup> describe two categories of experiential knowledge: (i) embodied knowledge (arising from personal, bodily experiences) and (ii) empathetic knowledge (developed through close association with others living through a particular set of circumstances). In exploring the experiences of young people before participating in a predictive test, it was evident that most have an empathetic knowledge largely built through observation of and shared experiences with affected family members and the resulting impacts on family dynamics. However, although experiential knowledge played a significant role in many aspects of young peoples' testing journeys, it did not appear to influence all aspects. **Figure 1** conveys the influence of experiential knowledge on the testing phases.

Despite being tested at different centers and for different conditions, almost all participants were highly critical of the process leading up to their test. Young people conveyed feelings of disempowerment, lack of control, and defeated expectations. They recalled the strain of what seemed to them to be a battle with health professionals and the emotional toll of the prolonged process they endured in order to access the information they desired. Many authors have focused on the outcome of testing as the principle source of anxiety along the testing pathway, failing to consider the process before testing as a separate source of stress. Young people in this study found the demands of the testing process onerous and a source of distress above and beyond simply living at risk.

All participants reported similar reasons for wanting testing. Crucially, those who received a gene-positive result remained satisfied in their choice to seek testing and maintained that their newfound knowledge was preferable to their prior uncertainty. Given their varied backgrounds, this highlights that a strong desire for testing can emerge from a variety of personal experiences.

The findings of this study are in keeping with those of earlier research in adults.<sup>17,21,29–35</sup> Studies exploring the impacts of informing adults at risk of either a familial cancer or a late-onset condition (e.g., HD) found that for people harboring a strong desire to know, receiving a gene-positive result was

psychologically less deleterious than the anxiety associated with uncertainty.<sup>36,37</sup> In adults, a form of self-selection has been proposed, whereby those who choose testing might have greater psychological capacity to cope with test results. It appears that this is likely to hold true for young people also.

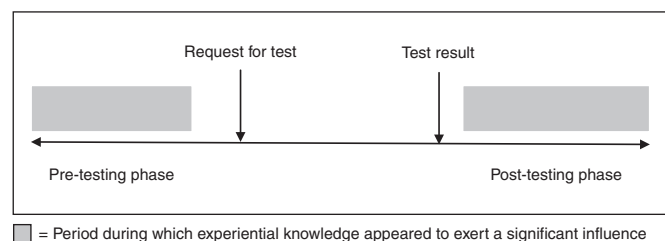
### Placing these results in the context of ongoing debate about testing young people for adult-onset conditions

Arguments concerning the competence of young people and potential harms of testing have come to dominate the ongoing debate about predictive testing in young people for adult-onset conditions.<sup>8</sup> Competence can be described as task or domain specific.<sup>38</sup> An individual may be competent to make one medical decision without being competent to make another.<sup>39</sup> Although this study did not attempt to measure participants' cognitive development, qualitative analysis of the interviews provided insight in relation to their task-specific competence for predictive testing, providing evidence of mature insight and life experience in domains relevant to decision making for predictive testing. By the time they stepped forward to request the test, most young people in this study had been preoccupied with consideration of personal risk and testing for many years. These nine young people had repeatedly considered the implications of testing for themselves and for their families and had considered the long-term implications of being gene positive. These results are in keeping with the most relevant past research, of individuals tested just after 18 years of age.<sup>11,21</sup> It is likely that many young people growing up at risk of a genetic condition have had time to understand the theoretical risk associated with being gene positive and have also witnessed and participated in managing the day-to-day reality of symptomatic disease in their families. Young people growing up in families affected by genetic disease may therefore have task-specific competence and maturity in relation to predictive genetic testing.

Our findings also provide evidence in relation to the consequences arising from a gene-positive result. The "unbearability of certainty" has been described in previous literature, asserting that knowing one will develop symptoms of a genetic condition as an adult is a greater burden for young people and is more likely to lead to psychosocial disturbance than uncertainty.<sup>40</sup> The findings of this study suggest that the opposite may be true for those who pursue testing. Before testing, many participants reported that uncertainty and anxiety associated with not knowing had become a barrier to their development and participation in everyday activities. Knowledge of gene status was considered to be empowering and allowed participants to get on with life and face key developmental tasks of adolescence and young adulthood such as establishing identity and making plans for the future.

### Conclusion

Young people in the current study had actively requested a predictive test; findings for this group may not apply across all at-risk young people, specifically those who do not request a test. The sample size is small and drawn from only two Australian states. However, in the context of current guidelines, predictive



**Figure 1** The role of experiential knowledge on phases of predictive genetic testing.

testing of minors for adult-onset conditions is uncommon, and the sample here included 9 of a possible 10 participants.

This study suggests that some young people who actively request testing might be well positioned to comprehend and cope with any challenges that arise from the testing process and outcome. For each young person, there are likely to be developmental and experiential factors that are of greater relevance than age in assessing competence and resilience. Understanding a young person's pretest trajectory is critical for predicting the impact of testing. The existing discourse may therefore have overemphasized the impact of the test result as a stand-alone phenomenon and underplayed the importance of considering the psychosocial context and previous experiences of each person.

The young people in this study demonstrated no evidence of major negative consequences associated with predictive testing, even for those receiving a gene-positive result. Several benefits associated with testing were also documented, regardless of test result. Unfortunately, a surprising source of distress identified by young people was the preparatory pretesting process designed for their protection.

This study provides neither a conclusive reason for granting all requests or for advocating a blanket ban on predictive genetic testing in minors for adult-onset conditions. Rather, the findings should assist with the development and implementation of evidence-based genetic counseling approaches. Further research, including longer-term follow-up, is needed.

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## DISCLOSURE

The authors declare no conflict of interest.

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