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ARTICLE

Experiences of predictive testing in young people at risk of Huntington's disease, familial cardiomyopathy or hereditary breast and ovarian cancer

Rhona MacLeod*[,1,2], Anna Beach[1,2], Sasha Henriques[1,2], Jasmin Knopp[1,3], Katie Nelson[1,2] and Lauren Kerzin-Storrar[1,2]

While debate has focused on whether testing of minors for late onset genetic disorders should be carried out if there is no medical benefit, less is known about the impact on young people (<25 years) who have had predictive testing often many years before the likely onset of symptoms. We looked at the experiences of young people who had had predictive testing for a range of conditions with variable ages at onset and options for screening and treatment. A consecutive series of 61 young people who had a predictive test aged 15–25 years at the Clinical Genetic Service, Manchester, for HD, HBOC (BrCa 1 or 2) or FCM (Hypertrophic Cardiomyopathy or Dilated Cardiomyopathy), were invited to participate. Thirty-six (36/61; 59%) agreed to participate (10 HD, 16 HBOC and 10 FCM) and telephone interviews were audiotaped, transcribed and analysed using Interpretative Phenomenological Analysis. None of the participants expressed regret at having the test at a young age. Participants saw the value of pretest counselling not in facilitating a decision, but rather as a source of information and support. Differences emerged among the three groups in parent/family involvement in the decision to be tested. Parents in FCM families were a strong influence in favour of testing, in HBOC the decision was autonomous but usually congruent with the views of parents, whereas in HD the decision was autonomous and sometimes went against the opinions of parents/grandparents. Participants from all three groups proposed more tailoring of predictive test counselling to the needs of young people.
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INTRODUCTION

Predictive genetic testing for late onset genetic conditions has become more widely available over the last two decades and has undoubtedly helped some individuals manage the uncertainty of their genetic situation, in spite of the limitations of genotype predicting phenotype, for example, age of onset and how symptoms will present.[1–3] Concerns about autonomous decision-making have led to practice guidelines, which include a presumption against predictive testing in minors for conditions that are unlikely to present until adulthood, and for testing to be delayed until the age when clinical interventions would commence for those conditions that may have onset in later childhood.[4] The desire to ensure that testing programmes do not cause more harm than good, has led to a considerable body of research on the psychosocial impact of predictive testing in adults, particularly for hereditary breast and ovarian cancer (HBOC) and Huntington's disease (HD). While most studies and reviews report an overall adjustment in the first couple of years post predictive testing,[5–8] distress levels may start to rise again later, presumably as the time approaches to possible disease onset.[9] Further, individuals may experience difficulties in specific areas of their lives such as family communication[10,11] and discrimination at home and in the work place or with insurance.[12] How young adults choosing to test early cope with such challenges and to what extent pretest counselling provides adequate support and preparation, has not been fully explored. Duncan *et al*[13,14] identified a number of psychological benefits and harms for minors (FAP) and young people (HD) who had been through predictive testing, some similar to those described in studies of older adults, as well as additional issues including fear of the blood sampling and impact at school.[13,14] Both Werner-Lin *et al*[15] and Hoskins *et al*[16] (2012) have recently reported studies of psychosocial impact of BrCa status in younger women (<24 and <35, respectively) as subsets of larger cohorts. They identified heightened issues for young women including the gap between testing and clinical surveillance, concerns about disclosing result to future partners and feeling under pressure to have children early. There have been no studies to date specifically looking at young people undergoing presymptomatic testing for familial cardiomyopathies (FCM). In a qualitative study investigating the impact of testing on children (<18 years) for a range of cardiac conditions with onset from childhood into adulthood Meulencamp (2008) found that children overall coped well, and that confidence in current and future treatment enhanced perceived control.[17] This current study adds to the literature by focusing specifically on young people tested for a range of later onset genetic conditions, and aims to look at the motivation of participants to be tested when young, their experiences of the counselling process and the advice they would offer to health professionals and other young adults considering testing.

[1]Manchester Centre for Genomic Medicine, Institute of Human Development, Faculty of Medical and Human Sciences, University of Manchester, Manchester Academic Health Science Centre (MAHSC), Manchester, UK; [2]Manchester Centre for Genomic Medicine, Central Manchester University Hospitals NHS Foundation Trust, MAHSC, Manchester, UK; [3]School of Psychological Sciences, University Of Manchester, Manchester, UK
*Correspondence: Dr R MacLeod, Manchester Centre for Genomic Medicine, Central Manchester University Hospitals NHS Foundation Trust, MAHSC, Manchester M13 9WL, UK. Tel: +44 161 276 6510; Fax: +44 161 276 6145; E-mail: Rhona.Macleod@cmft.nhs.uk
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MATERIALS AND METHODS

Participants

A consecutive series of 61 young people who had a predictive test aged 15–25 years at the Clinical Genetic Service, Manchester, for HD, HBOC (BrCa 1 or 2) or FCM (Hypertrophic Cardiomyopathy or Dilated Cardiomyopathy), were invited to participate. Thirty-six young adults (59%) agreed to take part in telephone interviews (10 HD, 16 HBOC and 10 FCM). Invited participants were all presymptomatic and a gap of at least 3 months had passed since receiving their test result.

The ages at which predictive testing is offered varies for different conditions in our Centre and is determined by a number of factors including usual age at onset, when surveillance options would be commenced and existing guidelines.[4,18] Currently, testing is usually offered from 18 years for HD and HBOC compared with 10 years or earlier for FCM. However, the focus of this study was particularly on emerging adults who had experienced predictive testing.

Interviews

The decision to conduct telephone interviews was informed by pilot work, which suggested that this approach would be more acceptable to young people used to telephone communication in other aspects of their life. Interviews were conducted by four MSc student researchers (HD by author KN; FCM by SH; HBOC by AB and JK). A semi-structured interview guide allowed flexibility to move between topics as led by participants themselves. The broad topic areas included: motivations for testing in young adulthood, impact of test result and experiences of the predictive test process. Interviews were 30–80 min in length, audio-recorded with permission and transcribed in full in preparation for analysis.

Ethical approval

Local Research Ethics Committee approval was obtained to conduct the study (LREC 10/H011/8).

Interpretative Phenomenological Analysis

The interview transcripts were analysed using Interpretative Phenomenological Analysis (IPA).[19] IPA is a qualitative method widely adopted in health psychology, that enabled account to be taken of the personal meaning of predictive testing to young people rather than seek an objectifying account. The data sets for FCM, HD, HBOC were analysed separately by the MSc student researchers who had carried out the interviews, and one of the senior authors, reflecting the individual projects and also providing a way to explore the specific condition before a cross-case analysis was conducted across the sets of results by the two senior authors (RM and LKS). Regular coding meetings were held involving all the researchers throughout the project.

The approach to analysis in IPA is ideographic, which involves focus on the individual case before the analysis slowly builds up to the group level.[20] Each transcript was read several times to increase familiarity with the data. Initial codes picked up keywords and early interpretations by the researcher. Themes encapsulating the meaning of the text were recorded. As the analysis progressed, patterns and connections were looked for between themes. Themes that clustered together were grouped under a superordinate theme that drew these together at a more interpretative level. A master table for each interview transcript included the superordinate themes, subthemes and illustrations of where examples could be found in the transcript.[20] The analysis of the data sets was an iterative process with frequent reference to the original interview data to ensure the analysis held true at the individual, as well as the group level.

RESULTS

Participants were aged between 15 and 31 years (median age 25) at time of interview with mean length of time since testing, 3.8 years (Table 1). Twenty-seven of 36 participants were female, with unsurprisingly more males represented in the FCM and HD groups than in HBOC.

For this group of 36 individuals who had chosen to have a predictive test in young adulthood, none regretted their decision. The majority of participants had accommodated to their result and it was particularly striking to observe the resilience with which those who had received a positive test result spoke of their experiences.

Table 1 Participants

Genetic condition	*No. of participants*	*No. of females (result of test)*	*No. of males (result of test)*	*Range age at testing (years)*
BRCA1	8	7 (3 + ve, 4 −ve)	1 (+ve)	20–24
BRCA2	8	8 (5 +ve,3 −ve)	0	19–25
HD	10	7 (4 + ve,3 −ve)	3 (2 + ve, 1 −ve)	18–24
FCM	10	5 (5 + ve)	5 (5-ve)	15–24
Total	36	27 (17 + ve, 10 −ve)	9 (3 + ve, 6 −ve)	15–25

+ve (test positive)—mutation present; −ve (test negative)—mutation absent.
Each quote in the results is followed by the gender of the participant (F, female, M, Male), age at time of testing, and the test result.

Being tested as a young adult

Not a difficult decision. Most of the participants did not perceive the choice of having a predictive test as a decision to be deliberated over, but rather something they felt compelled to do in order to obtain information about themselves and to relieve uncertainty.

> 'I knew I had to, because I knew I'd spend my whole life wondering, will I? There was never any second doubt that I was not going to get tested. I don't know, I think because I thought about HD so much that I was just going to do it anyway.'
> F, 18, test positive HD

Time for action. A key motivator for participants at risk of inheriting a gene fault predisposing to HBOC or FCM was the perception that they were doing something to alter the course of a disease that had led to the death of affected relative(s).

> 'I just thought that you know if she (mum) would have had the opportunity to have the test, then things could have been a lot different'
> F, 21, test positive BRCA2

> 'Both my uncle and my dad had died from the condition so knowing meant that things could be done. So we were just really glad to have the test and just went ahead'
> F, 16, test positive FCM

The information provided by a test was perceived by these young people as useful in helping to plan, particularly around reproductive decisions. Only 3 of the 36 individuals in this study (two HBOC, one HD) had a child(ren) at the time of their predictive test, however, thoughts about *future* children were recalled as factoring strongly in the decision to be tested.

> 'You know starting your family earlier, all those things that would have helped me plan my life a lot earlier.... so I just think it was more about me being proactive with my life choices.'
> F, 24, test negative BRCA1

> 'Its definitely been a positive experience to find out so that I can, you know, my children, make sure they if they need any treatment. If I pass it on to them'.
> F, 16, test positive FCM

Parental attitudes to testing. Differences emerged between the disease groups in terms of parental attitudes to testing. Young people at risk of FCM felt that parents and health professionals believed that testing was a good idea, and some participants recalled that this had been stated explicitly to them. Interestingly,

even where parental influence had factored significantly in the decision to be tested, participants did not find this intrusive and several indicated that they thought their parents had their best interests at heart.

> 'I think as I was quite young it was really my mum who wanted it for us. But that was the best thing to do at the time… Looking back I think I still would have had the test- its better to know these things.'
> F, 16, test positive FCM

> 'I think at the time I felt as if they wanted me to go through with the test...but it might just have been me thinking that but I felt, not pushed, but I felt as if they were more that way, for me to get checked.
> M, 24, test negative FCM

The decision to have a predictive test for young people at risk of HBOC was more of an autonomous decision, albeit with implicit parental approval in most cases.

> 'I think she (mum) was quite keen for me to move forward as well. Erm, although she doesn't put any pressure on, she just said she thought it would be a good idea for me to find out....'
> F, 25, test negative BRCA2

> 'I really feel like I did it. Not that I did it on my own, because I had like a *bloody entourage* every time I went to the hospital, but like I'd, I felt like I'd kind of achieved it on, you know I'd done it for myself. So I'm really glad I did it.'
> F, 23, test positive BRCA1

For the HD group, the decision to test at a young age, was also an autonomous one, but in contrast to the other two groups of conditions, none of the participants spoke of a parental desire for them to be tested and indeed the decision was sometimes at odds with parental opinion.

> 'My mum didn't think that I should have the test. I mean it wasn't like an argument or anything, it was just more concerned you know how would it affect me once I knew if I did have the gene.'
> F, 19, test positive HD

Expecting to be gene positive

A major theme to emerge among the HBOC and HD groups, but not the FCM group, was the expectation that the result would be positive. While participants had understood they were at 50% risk, most had adjusted this to believing it to be closer to 100%. Indeed all of the HD participants and 14/16 of the HBOC group, reported that they believed before testing that they would turn out to be gene positive. For some, it was easier to prepare for the possibility of bad news rather than go in to testing expecting good news and then be disappointed, for others it related more to their beliefs about similarities to their affected parent.

> 'I convinced myself I got it..... because I'm so like my mum, I look like her, I act like her. You know I could see myself in her even at that age... I think when you're young you do think like that.'
> F, 18, test negative HD

> 'In my mind I felt like I would have the gene'
> F, 24, test negative BRCA1

Participants having a predictive test for FCM were less speculative about the outcome before testing. The test itself was seen more as a necessary step in finding out whether further medical surveillance would be required.

Process of adjusting

Initial shock. While almost all the participants recalled feeling ready to have the test and many had gone in to the process expecting to receive bad news, the test result itself came as a shock, and was often recalled vividly in the interviews. This was particularly true for individuals receiving an HD result, but not exclusive to that group.

> 'I remember it very clearly, I think I was just shocked more than anything and then, I don't know for about two or three days I think it was just a bit weird, just trying to get my head around it all'
> F, 24, test positive HD

Several participants reflected that their youth may have made it harder to anticipate and envisage the emotional impact of the test result, including several who had received a test negative result, but felt that had the result been different they may not have felt prepared for such news.

> 'I was only 21 when I had the test done, I think it didn't seem real at the time… It wasn't until I got the test, the test result that I thought this, this would have been a huge thing actually'
> F, 21, test negative BRCA2

> 'I think that em at the time it was also, it wasn't that.. erm.. 'what happens if it's positive?' I think being sort of that age, therefore indestructible'
> M, 19, test negative FCM

Results not shared widely. Common across the disease groups, was the finding that most young people had shared their test result with only a small immediate circle of close friends and family. This may have been a consequence of discussions in the pretest counselling period, but most participants had chosen not to inform their wider support network. Participants at risk of FCM were most matter of fact in their explanations to friends and colleagues, setting it in the context of practical issues such as health and safety at work or explaining absences from school or employment.

> 'I told them I was going to have the test because I was having a day off school. It doesn't seem to affect us, we don't really bring it up.... there's not a reason to bring it up or worry about it. It's not going to come up in conversation'
> M, 16, test negative FCM

Reasons for not talking about their result with a wider group of friends included not feeling the need for more support and fearing that other people would not understand the complexities of the testing decision or trivialise their feelings about the result.

Family impact of test result. These young people expressed concern about the impact of their test result on other members of the family both relatives and partners. Indeed several described their desire to avoid causing pain to family members who may have been affected themselves and/or watched the devastating effects of the condition. For those participants receiving good news, it was tempered by the knowledge that other members of their family may be facing a very

different situation such as siblings remaining untested or with a different test result.

> ' …when I first found out I didn't want to be too happy around them because its still not the best of situations because my mum's still poorly with it so even though its good news for me, I couldn't be too happy. I'm still upset about my mum.'
> M 24, test negative, FCM

> 'When I got told I didn't, I mean there were tears of happiness, but then for my sister as well, I felt really bad for her then.... I've had times when I've really thought about it and I have got upset. The fact that my sister has got it and I haven't and I always think to myself what does she feel about me now because I haven't got it and she has.'
> F, 22, test negative, HD

Positive appraisal. The initial period of shock, and for some acute distress, was variable in duration, recalled as lasting weeks or months. Most participants then found ways of facing up to the future, and in the interviews talked openly about how they coped with the knowledge of their genetic status. This included focusing on something favourable about their situation, for example, that the illness may be a long way off or that they were better off in some way than other people who had not been tested.

> 'There are negative things but focus on the positive which helps a lot. You might not get it young. I mean I was worried that I was going to get it young like my mum but I might not'
> F, 21, test positive HD

> 'I've got a better chance than someone who hasn't got it (HBOC) because they could be walking around with cancer, whereas if I get it I'm going to get in straight away and it's going to be gone'
> F, 21, test positive BRCA1

Faith in medicine featured strongly among young people across the three groups of conditions. There were two aspects to this; belief in the ability of the medical profession to help with any problems as they arose and secondly hope that the science will lead to better treatments in the future.

> 'Having the option to have operation and avoid cancer I just think, I think it's amazing'
> F, 23 test negative BRCA1

> 'I've also got good faith in medical science…it's only going to get better the more research goes on
> M, 19, test positive HD

Knowing the result itself provided the anticipated relief for some participants, even those who received a 'bad news' result:

> 'It was like the biggest relief. You know when you've felt like you've held your breath for ages and then you go (*breathes deeply*) I can remember that feeling. It was like I hadn't breathed for ages (*breathes deeply*) right I know now'
> F, 22, test positive BRCA1

Several participants felt they had matured as a result of their testing experience and with that came a reappraisal of how they planned to live their lives. In a number of cases this had already led to them making positive life changes.

> 'I'm glad I got the test because it did make me get on with my life. I went to uni and qualified as a nurse and got on with my life now.'
> F, 18, test negative HD

View on testing process

Across the three groups of conditions, predictive test counselling was viewed as useful not in facilitating a decision, but in providing personalised information in language accessible to a young person.

> 'They didn't use massive words. They always made sure that I knew what they were saying. So I had a full understanding'.
> M, 16, test negative FCM

While participants were overall positive about the counselling process, it was not always clearly recalled and in some cases the counselling was seen as a means to an end.

Several participants had issues with the length of time between appointments and the lack of tailoring to the individual's specific situation.

> 'If people are unsure and they need to find out more information, fair enough, but if people are adamant, I'd just rather they did it sooner rather than later.'
> F, 22, test positive BRCA2

Interestingly, while advocating an individualised approach that recognised the readiness of the individual to be tested, none of the participants advocated lowering the age limit of testing to under 18 for HBOC or HD. At an early stage in the interviews, several participants commented on the issue of testing minors and this was further explored in later interviews.

> 'I just think you should you know live a bit more… you're still a teenager and still sort of finding your way in life…'
> F, 21, test positive BRCA2

> 'They are not mentally prepared; they're not an adult yet….. like a lot of them will think *oh well I'm not bothered*. But deep down inside they could be really upset and think *oh my God, what am I gonna do, my life's over*.'
> F, 24, test positive BRCA2

A few participants who had initially requested testing as a minor felt, looking back, it was better to have waited.

> 'At the time I felt quite adult, you always do don't you! You know the 'I know it all' kind of thing... I'm glad I didn't have it when I was 15.'
> M, 19, test positive HD

Limitations

While a consecutive series of young people were ascertained for the study conditions tested, the 59% who agreed to participate may reflect those people who had the best adjustment to their result and who were more likely to engage in follow-up. In addition as this was a retrospective study, some participants may have reframed their experiences for example to give a more positive account of their adaption in the post test period. The study did, however, identify areas of difficulty for individuals receiving both mutation positive and negative test results. The fact that each condition constituted a separate MSc project was both a strength and a weakness of the

study design. For reasons of consistency it may have been better for one researcher to conduct all the interviews. It did, however, help to avoid the assumption that the findings would be the same across patient groups and enabled each researcher to approach their own group of patients afresh. There was also the opportunity for the supervisors to listen to the recorded interviews and ensure they were being conducted in a similar way (for example, comfort with silences, use of prompts and so on).

The decision to conduct telephone interviews rather than face to face interviews may mean that the absence of non-verbal cues led to certain nuances of meaning being lost. However, this was a potential study group of participants who led very active lives, and indeed most had requested contact to a mobile number at a time they could talk in private. It also supports the findings that the relative anonymity of telephone interviews may be more comfortable for participants when discussing personal and sensitive issues.[21,22]

DISCUSSION

This is the most extensive qualitative study to date that has purposefully sampled a consecutive series of individuals who have had predictive testing in young adulthood for one of several genetic conditions. The focus here is not whether minors should be tested for late onset conditions, but rather to identify the needs of emerging adults going through the predictive test process.

Importantly, none of the participants across the groups in this study expressed regret over their decision to be tested when young. Our findings support those of Duncan *et al*[13] who found that predictive testing can be an empowering experience for young adults. In our cohort, testing commonly led to positive reappraisal including the perception of control over the course of future events, a finding noted previously among teenage unaffected carriers of FCM[17] and young female BrCa carriers.[15,16] However, a new finding was that participants felt that their lack of emotional experience at the time of testing had made it difficult for them to rehearse the possible psychological impact of a test result. We provide recommendations for genetic counselling practice below (see Table 2, 'Practice points'), including the importance of follow-up in addressing ongoing emotional support. This is important to address if we are not to lose some of this particular age group to follow-up. There is some evidence in the HD literature that individuals who drop out of follow-up may be among the group of patients with greatest needs as they start to develop symptoms.[9] Other authors have also advocated the particular importance of genetic counselling follow-up for individuals tested young for BrCa[16] and FAP.[14] In our Centre individuals at risk of HD and HBOC are among those kept under long-term follow-up through our genetic family register service.[23–25] Another strategy would be to have clinics specifically for younger people and for genetic counsellors to utilise the peer support potential this may offer.

This study provides new evidence for the varying role of parental and family influence on the choice to be tested, depending upon the nature of the condition. While HD participants had in some cases been tested against parental or family advice, BrCa participants were aware of parental views in favour of testing, and many FCM participants saw their parents' views as pivotal. However, most participants felt they had been allowed to make an autonomous choice and were comfortable with the influence of family and professionals. This is in contrast to the findings of others where parental influence was perceived as having interfered with the young person's autonomy.[14,15]

There is substantial evidence that a key concern for adults undergoing predictive testing is the potential impact on their loved ones.[6,7,26] Previous studies of young females with a BrCa mutation have identified concerns about their future health on their children's well-being.[15,16] In this study, it was striking that most participants, including the youngest FCM testees, had anticipated how their family members might react to their test result and sought ways to avoid causing them distress. This caretaking response often extended towards the well-being of future children. This is of particular interest to genetic counsellors who may underestimate the extent to which this would be a concern for a young person who may be single and without children at the time of testing.

Table 2 Practice points for predictive testing in young adulthood

- Acknowledge and utilise young person's existing knowledge of testing
- Explore information needs around future choices, for example, reproductive decision-making.
- Explore/engage with parental influence
- Explore expectation of test result
- Elicit available support for the immediate post test period
- Consider taking blood sample early in session if young person afraid of needles
- Provide opportunities to participate in research where these exist
- Agree early follow-up contact to offer support around disclosure of test result to family

Although the study did not set out to explore attitudes to predictive testing of minors, it is of note that participants tested for HD or HBOC did not advocate lowering the age of testing before age 18. Interestingly this included a small group of participants (HD and HBOC) who had initially requested the test as a minor. This finding emerged in the course of interviewing participants about their experiences and would be an interesting topic for further investigation. For example a retrospective study of young people requesting–but not going ahead with–a test as a minor, could usefully explore how they felt about such a decision as an older adult and with the benefit of hindsight.

Throughout the research interviews, participants often expressed the need for counselling to be more tailored to young people. We advocate that professionals providing predictive testing should, while being guided by existing protocols, be flexible in order to give attention to those issues which may feel more relevant to young people at the time of testing, while ensuring that follow-up provision is in place so that emerging concerns can be addressed at relevant life stages in the future. Genetic counsellors should also be aware of factors that may disengage young people from discussions around predictive testing. For example, where the young person has a fear of needles it may be wise to offer to take the blood sample earlier in the session, or consider saliva sampling as an alternative.[14]

Our findings suggest that individuals choosing to test young may be a select group and not typical of either their peer group of at-risk individuals or indeed older adults choosing to have a predictive test. It is important to also emphasise that many at-risk individuals choose not to be tested young. However, it is clear that the decision-making process was not the challenge for these young people, and prolonged discussions of pros and cons of testing may be less helpful than support around the result itself and longer term follow-up. Future prospective studies could challenge the considerable energies of this group to look at effective ways of meeting their needs in the pre and post test periods, for example, exploring different models of delivering post test support. It would be interesting to explore the additional information resources and

support that young people access including online support such as the Huntington's Disease Youth Organisation (HDYO) website (www.enHDYO.org).

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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