

Original Article

Help or hindrance: young people's experiences of predictive testing for Huntington's disease

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A growing number of young people (YP) are requesting predictive testing (PT) for Huntington's disease (HD), yet there is little research in this area. The aim of this study was to explore YP's experiences of PT for HD, the impact of their result and any gaps in information or support. In-depth interviews were conducted with YP who sought PT for HD from nationally funded Genetics Services. Participants were recruited through the Grampian Genetics Service or Scottish Huntington's Association. Twelve female participants aged 17–26 years were recruited (seven below 20 years). Pre- and post-test interviews were conducted where possible. A qualitative thematic analysis suggests three main testing experiences, regardless of test result. Testing may be: (i) a journey of empowerment, (ii) an ambivalent process or (iii) a poor experience. In pre-test counselling, gaps in emotional support were highlighted. The post-test period was particularly difficult if there were unanticipated changes in family dynamics or an individual's result contradicted what they expected 'deep down'. YP's experiences of PT for HD are generally similar to those of adults, but testing may help or interfere with key issues related to this age and stage. Implications for clinical practice are outlined.

Conflict of interest

K. F. K. is a board member of the Huntington's Disease Youth Organisation but there is no conflict of interest. Z. M. and L. M. declare that they have no conflicts of interest. The funders had no involvement in the study design; in the collection, analysis and interpretation of the data; in the writing of the report; and in the decision to submit the article for publication.

K. Forrest Keenan^a, L. McKee^a and Z. Miedzybrodzka^b

^aHealth Services Research Unit, University of Aberdeen, Aberdeen, AB25 2ZD, UK and ^bMedical Genetics, University of Aberdeen, Aberdeen, AB25 2ZD, UK

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Corresponding author: Karen Forrest Keenan, Health Services Research Unit, University of Aberdeen, 3rd Floor, Health Sciences Building, Foresterhill, Aberdeen AB25 2ZD, UK.
Tel: +44 (0)1224 438161;
fax: +44 (0)1224-438165;
e-mail: k.keenan@abdn.ac.uk

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Huntington's disease (HD) is an autosomal dominant neurodegenerative disorder with onset classically between 35 and 55 years (1). Symptoms include cognitive impairment, movement disorder, personality and behavioural changes (1). Life expectancy is approximately 15–20 years from the onset. There remains no cure but there are guidelines for Standards of Care (2). Children of an affected or a gene-positive parent have a 50% risk of inheriting the mutation and developing the illness in later life.

Predictive testing (PT) has been available for adults at risk for more than two decades. Nevertheless, less

than 25% of those at risk present for PT, ranging from 3% to 25% in published reports (3). Testing often brings psychological and social challenges, but also benefits such as relief of uncertainty, information to make future plans and reproductive choices (4). In 1994, an international PT protocol was developed to outline best clinical practice to protect those at risk. These guidelines have recently been updated to take into account the findings from two decades of psychosocial research (5). Importantly, PT for HD has not led to increased rates of suicide or psychiatric illness (4). And whilst carriers may experience initial shock and upset they generally adjust to their

result within 1 year, although anxiety levels may rise in the long term (4). Nevertheless, PT for HD can have a far-reaching impact, with reactions only partially dependent on the outcome (4). And whilst non-carriers express relief some have difficulties adjusting to a good result (6).

Although the numbers of those who test remain low the age of those testing may be changing. For example, in the UK, there has been an increase in YP aged 20 years and below who test (7). Concerns have been raised about testing YP because it may do more harm than good (8). Thus, one consistent recommendation is that the minimum age of testing be 18 years, although minors who seek testing should have access to genetic counselling (5).

The little research which has been undertaken into YP's experiences (those aged 16–25 years) suggests that the benefits and harms of testing are similar to adults', and that the majority adjust and do not regret their decision (9–12), but these findings are based on small retrospective samples, usually from one genetics centre with the majority of participants receiving a 'good' result. In online forums, some YP have reported traumatic testing experiences (13), supporting studies which suggest that the process itself can engender harms and benefits (10, 12).

Little research has explored the extent to which testing impacts on developmental tasks of this life stage (10) and it is not known if YP have different information and support needs from adults (11). The aim of this study was to explore YP's prospective and retrospective experiences of PT for HD, the impact of their result and to identify any gaps in information or support in the pre- and post-test period.

Materials and methods

Recruitment and sampling

Participants were recruited through the Grampian Genetics Service and the Scottish Huntington's Association (SHA) between August 2011 and November 2013. Any new or previous test candidate aged 25 years and below was invited to take part by the consultant ($n = 11$) or SHA ($n = 13$). Potential participants were asked to send back a reply slip to the researcher (K. F. K.). Purposive sampling was used to recruit as diverse a range of participants as possible (14). After an explanation about the study, all participants were asked to sign a written consent form agreeing to take part.

The study was approved by the North of Scotland Research Ethics Service.

Data collection

Semi-structured interviews were conducted (by K. F. K.) with the majority of participants and lasted between 30 and 90 mins. Interviews took place in participants' homes or the genetic clinic. One participant chose to be interviewed by phone and another by e-mail. Prospective and retrospective interviews were conducted where possible. All phone and in-person interviews were audiotaped with consent and fully transcribed; confidentiality and anonymity were assured.

The interviews focussed on: reasons for seeking PT, experiences of the testing process, the impact of any result, any gaps in information or support, as well as participants' experiences of HD. One interview was stopped because of the participant's distress. Post-interview support was arranged with a health professional.

Data analysis and presentation

The analysis was conducted in two main phases. In the initial phase, a thematic analysis was undertaken using a system of open coding and the constant comparison method (15, 16). K. F. K. identified themes and categories by reading and re-reading transcripts. Different segments of the data were then grouped together into separate categories, allowing reflection on the major themes and any relationships among them. The thematic analysis was ongoing throughout the fieldwork until a point of data saturation was reached about the main themes (16).

In the later phase, the findings were also interpreted with reference to existing psychological and social theory, where issues related to adolescent development (17), family systems (18) and (dis)empowerment (19) were anticipated to arise. Thus, whilst phase 1 aimed to generate inductive themes from participants' accounts, phase 2 interpreted these accounts within a broader theoretical framework.

To illustrate the range and complexity of participants' experiences, we present five case studies for discussion. These cases have been chosen because they illustrate the main themes and categories within the whole dataset.

The participants

Twenty-four patients were invited to participate by the clinical geneticist (8 females and 3 males) or SHA (13 females). Fourteen replied and 12 were interviewed (2 were lost to follow-up). Six were recruited by the clinical geneticist and six through the SHA.

Ten participants were tested, one withdrew and the remaining participant was lost to follow-up. Seven participants took part in pre- and post-test interviews, two only in pre-test and three in retrospective interviews. Two partners were also interviewed but were not included in the analysis.

Table 1 summarises participant demographic characteristics. All participants were female and described their ethnicity as 'white.' All names and identifiable details have been changed to protect confidentiality.

Results

Our qualitative analysis found that testing was experienced by YP as either: (i) a journey of empowerment ($n = 3$), (ii) an ambivalent process ($n = 4$), or (iii) a poor experience ($n = 5$), regardless of test results. Four categories structured each account: Family background, Deciding to test or not, Experience of Pre-test Counselling and Post-Testing/Postponing testing. The length of testing was typical for the UK for nine participants – varying from between 5 to 9 months (7).

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Table 1. Demographic characteristics of patients interviewed

Age
Mean age 21 years (range 17–26)
Adolescents aged 17–19 = 7
Young adults aged 24–26 = 5
Number of participants
Married or living with partner 4
Live at home 5
Live on own 1
In supported accommodation 2
Parents of children 3
Have siblings 11
In full- or part-time education 5
Possess post-secondary qualifications 1
In full- or part-time employment 4

A journey of empowerment

Three participants described PT for HD as an empowering experience, two carriers and one non-carrier. Katie was particularly articulate about her journey of empowerment and the positive impact on her personal identity and relationship with her partner.

Katie age 24, test at 21

Family background. Katie described having a 'bad experience' of HD when she was a child because she lived with her affected mother and father who 'never coped with it'. There were issues of paternal non-disclosure and concealment which meant that she had little knowledge of HD or her risk until her mid teens. By then, Katie was a young carer – looking after the household and her siblings.

Deciding to test. In her late teens, Katie was increasingly anxious about her risk, suffering from intrusive thoughts, symptom searching and insomnia. She coped by being 'constantly busy so not to think about it' and now believes she was 'probably depressed'. Katie wanted the test to make important life choices and future plans.

Experience of pre-test counselling. Katie chose to proceed slowly with testing because she wanted time to arrange her finances and be sure of her decision, 'I wanted to take a while.' Katie described a positive experience of pre-test counselling:

I think they're great people ... It made me think about things you wouldn't think about on your own.

During this time, Katie also received support from other agencies that she valued, especially advice from the Huntington's Association about end of life care, 'I was finding the visits [to mum] demanding and upsetting,' and psychological support to cope with intrusive thoughts.

Katie's family had a tense relationship with the local genetics service so she attended the majority of appointments alone. In addition, she chose not to tell her partner about testing, or her family history, reflecting that 'you're also doing your best so they don't find out the real you'.

Post-testing. Katie described the difficulty of receiving a carrier result and the time it took to adjust. A few months afterwards she had a major 'relapse' about possible symptoms and sought intervention from her counsellor, who she felt understood and was quick to respond.

At the time of interview, Katie had implemented the plans she had discussed in pre-test counselling and was considering starting a family. She was hopeful about her future and the possible benefits of clinical trials 'in the next 5–10 years'. She had also disclosed her result and family history to her partner:

My partner he's like you're a different person now ... And thinking back to before the test I am. I've not really needed them in the last year. I must be doing something right!

An ambivalent process

Four young women described seeking testing as more ambivalent, reflecting they had mixed experiences, one carrier, two non-carriers and one who postponed testing. In these accounts, genetic counselling was information-rich but there was a lack of emotional support and little control over the test process.

Laura, age 18, testing postponed

Family background. Laura lives at home with her father and mother who has late-stage HD. She was told about her mum's diagnosis by her parents as a child, noticing the onset of symptoms around the age of 13/14. Laura undertakes no personal care for her mother and there is little interaction between them. Laura's own risk was not a huge concern for her until now.

Deciding to seek testing. Laura's older sister recently tested and received a non-carrier result, but this good news 'Just made me think my turn next! So I was determined to get the test done as soon as I was 18'. She searched for information online about HD, but found different advice and 'false hope':

That was another reason why I was up for getting the test because if it did come back positive there'd be medication ... I thought I wouldn't change my mind at all.

Laura was encouraged to seek genetic counselling by her family because 'I was getting really worked up ... Everything was hitting me really hard about the disease'.

Experience of pre-test counselling. When Laura actually attended her appointment she was unsure whether she would proceed with the test or not, 'I was still 50/50 when I went in'. Laura valued the opportunity to receive accurate information about HD and discuss the pros and cons of testing – even though it was a shock to find out there was no treatment, 'It hit me really hard when they said no there's nothing'. And whilst Laura felt the information was helpful, she also reflected that:

They never really looked into the emotional side of it, which is the side I struggle with mostly ... the everyday seeing my mum and having that feeling I might get it ... [It's someone] to be there for you ... and understand the situation you're going through ... [because] as soon as I go in there I just forget about the emotion.

Postponing testing. Laura decided to postpone testing, so she can concentrate on her short-term goals, i.e. passing her exams. She does not want to ‘take the risk’ of getting bad news, questioning whether she would cope ‘with such a concrete answer’. She plans to seek help in the future if she needs emotional support, e.g. through online peer support or other counselling services.

Louise, age 18, test at 18

Family background. Louise is 18 years old and has a daughter. She lives at home with her mother, stepfather, daughter and other close relatives. Her parents separated when she was a child. Louise has little contact with her biological father. She only found out recently from her mother about her estranged father’s family history of HD and her own 50% risk.

Deciding to test. Whilst Louise ‘did not feel upset or anything’ about this news, her reaction was to seek testing and ‘go for it.’ She wants the test ‘for her daughter’s sake’ because if she is a gene carrier she wants to tell her daughter, but if she isn’t ‘she won’t have to go through what I have’.

Experience of pre-test counselling. Louise finds waiting ‘the worst feeling ever’ and would prefer to get tested ‘straightaway and move on.’ She questioned why there is so much emphasis on pre-test counselling instead of post-testing. At the same time she acknowledged ‘it’s their job to make sure you’re ready.’ Louise complies with three pre-test sessions, believing she needs to present herself as ‘ready’ and to ‘speak up’ about why she wants the test.

If she receives a bad result, Louise expects to be ‘devastated’, but her view, and that of her family’s, is not to get ‘head over heels about it’. Whilst she has been informed the average age of onset is late 30s/early 40s Louise is adamant that she will not ‘speed things up’ and plans to live her life ‘as I was going to do it’.

I’m not going to let it stop me from doing the things I want to do, or the speed that I want to. I’m just going to do everything like I normally do, as I was going to do it, and if it’s positive, when the time comes, then fine! ... I don’t want to speed things up ... I don’t want her being brought up in a rushed family ... Just make everything as normal as it can be.

Like others, Louise had not discussed her feelings with her parents or siblings about testing, ‘we don’t really speak about it’.

Post-testing. Louise found testing a long process, but was not ‘going to back down’ and was relieved to get a good result. In her post-test interview, Louise focused on how she had become homeless during the test process and the battle she had to find suitable housing. She had recently secured her first home and stressed that her priority was making a ‘normal family life’ with her partner and daughter. Testing for HD had been one more thing in a ‘busy stressful year’.

A poor experience

Five participants described testing for HD as a poor experience, four carriers and one non-carrier. The ‘waiting time’ was particularly anxious. Two young women had not anticipated the extent to which their result would impact on family dynamics and their own personal identity.

Jenny, age 19, test at 18

Family background. Jenny grew up knowing about HD because she was told by her parents as a child and witnessed the illness in a close relative. In her teens, Jenny’s mother was diagnosed with HD, but her symptoms had little impact on family life. Her own risk of HD was not a major concern. She felt she had a close relationship with both parents and a ‘loving, open family’.

Deciding to test. Prior to her 18th birthday, Jenny’s mother had a sudden deterioration in her condition that triggered a strong desire to test, ‘I couldn’t sit and watch that and not know’. As soon as she turned 18, she sought testing and believed she was a good candidate: she had knowledge and experience of HD, a supportive family, she was mature and would seek additional support.

Experience of pre-test counselling. Jenny was ‘rebellious’ with her family and genetic counsellor and did not seek her parents’ support, explaining that ‘I wanted to talk about it every day, but I didn’t want to put it on mum or dad’. Jenny also got involved with a new peer group and began to engage in risky behaviour, admitting that ‘I went off the rails for a bit ... I was taking my mind off it all’. At the same time, she was preparing herself to hear bad news, working with another professional to consider the impact of testing:

Deep down, I convinced myself I had it. I structured my life around it ... for months ... I was hard at it.

At her first visit, Jenny was informed that testing was a long process but she was more sceptical than Louise about the genetic counsellor’s role, ‘she only wants to make sure I won’t commit suicide’, and less compliant:

I just kept going, can you just test me, can you just test me? I don’t want to do this, I go through enough bloody counselling.

In this ‘year of firsts for everything’, Jenny had also started her first serious intimate relationship, but realised there was a pregnancy risk and decided she would avoid sex.

Post-testing. A year later and Jenny is still reconciling and adjusting to receiving a non-carrier result. She described an ongoing messy personal life. And whilst she continues to struggle with her mother’s worsening condition Jenny has taken on a more caring role.

I’ve to stop being so doom and gloom about my mum, it’s not the end of the world ... We’re losing her like ... (voice quietens). But

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... everyday you've got to get up and ... treat her like normal ... have a laugh ... Everything is not going to go back to normal ... So it's time I started dealing with that.

Jenny has also become her parents' confidante, leading to the discovery of a family secret that she had never anticipated, and would not have been disclosed if she had received a carrier result. Overall, Jenny reflected 'It was the worst year of my life'.

Michelle, age 19, test at 18

Family background. Michelle grew up in a single parent family with her affected mother and sibling. She first noticed changes in her mum in her early teens. They found out it may be HD when a relative was diagnosed, but her mother delayed diagnostic testing for several years. Michelle disclosed a history of self-harm, but 'is getting help for that'.

Deciding to test. Previously, Michelle had not sought testing because 'we've always said we wouldn't get tested'. But she recently changed her mind, reflecting that 'I don't do wait and see'. Michelle's anxiety significantly increased when she overheard relatives' question whether she was showing symptoms, 'then I started thinking about it all the time ... So I wanted to get tested ... [to prove them wrong]'.

Pre-test counselling experience. Michelle was referred for PT by a professional because of her anxiety, describing that 'I got tested the weirdest way ever' and how the testing process was shortened:

My anxiety got too much, I ended up taking a bad turn, so they were like I'll give you your results asap because it's not doing you any good waiting.

She also chose not to tell any of her family about testing – 'I pushed them away ... I didn't want to hurt them'. Michelle experienced another 'bad turn' several days before her result requiring professional intervention, 'I ended up flipping out and getting really angry'.

Post-test experience. Similar to Katie, Michelle described the profound emotional impact of being told that she was a gene carrier, and that she had not expected, or really prepared, to hear bad news.

Even though I did build myself up for it coming back positive I still got really really emotional. And I didn't expect it to come back positive, deep down. And when it did ... I didn't want to believe it at all.

Her initial reaction was shock and disbelief. She also wanted to tell one of her relatives, 'when I got the test result back I was like I need to tell one of them' and since this disclosure some family relationships have improved.

Six months post-testing and Michelle is trying to adjust to the bad news – 'I still think I'm dealing with it' – and an additional dilemma that has emerged:

Just now I don't know what to do ... With the result I feel like I need to do something quick, if that makes sense, because I know that I am going to develop it ... But just now I don't really have the motivation to do anything ... [voice saddens and tails off] ... I don't really know how much time I do have.

In retrospect, Michelle believes that she would have coped better if she had had family support:

I think that I could've changed it because at the time I was completely alone – I wasn't because I had the staff – but I didn't have any family. And I feel like if I'd told them, or at least told my gran, then I'd have been able to cope. I knew I had all the staff, but it's still not the same as your gran.

Discussion

This article explores YP's experiences of PT for HD, and is the first to document prospective and retrospective accounts. A qualitative analysis was undertaken, which revealed testing was experienced by YP as either: (1) a journey of empowerment, (2) an ambivalent process, or (3) a poor experience, regardless of test result.

A key finding was that a subgroup of YP experienced the testing process and their result as empowering (9,11), whether they received 'good' or 'bad' news. In particular, PT helped to establish personal identity – a key developmental task at this age (17) – and could enable disclosure in personal relationships, e.g. about being a 'pre-symptomatic' person. We also found testing is important for YP who have other identity challenges related to HD, e.g. coming to terms with being adopted or fostered, being a responsible young parent, or needing to know if you will be a carer or patient. For participants with 'complex pasts' (9), testing could be beneficial for emotional regulation (19). It could also improve interpersonal and family relationships (10).

An important finding was a subgroup of YP who sought testing when a parent's condition deteriorated, but the emotional impact of losing a parent and living at risk was 'not looked into'. Previous research has found that a significant minority of clinical genetics professionals experience discomfort in the presence of grief and loss, and also feel inadequately prepared for such patient experiences (20). Taken together these findings suggest there is a gap in pre-test counselling with regards to YP's experiences of grief and loss and any relation to seeking testing.

In contrast to adult studies (21), the findings of the present study draw attention to the isolation YP experience in pre- and post-testing and barriers to the development of a trusting patient/counsellor relationship, e.g. using defensive mechanisms such as rebellion or risky behaviour (9). For many, this isolation was compounded by a decision not to tell close relatives about testing – either through a desire to protect them, poor relationships, or because testing went against the family norm. Isolation during the test process may be particularly acute for YP who are likely to be separating from parents, and experimenting with new but fragile peer relationships (17), and concealment of risk was not uncommon. An important finding is that in retrospect

family support was preferred by participants, but it did not have to be from direct family, or throughout the whole testing process. In contrast, a few YP found the support person requirement prohibitive preferring to attend alone (22), or felt there was an unhelpful duplication of services.

The findings of the current study draw particular attention to ‘The Waiting Period’ experienced prior to receiving a test result (13), with participants describing mild to severe anxiety. Whilst the majority felt this period was too long, most complied with pre-test counselling. However, similar to other studies of YP we found participants ‘going through the motions’ or ‘playing a role’ to access testing (11). Participants also had different pre-test experiences depending upon how and when they had decided to seek testing. Like Macleod et al. (11) YP who knew they wanted the test from their first appointment felt there was repetition and inflexibility. Thus, for those who ‘have to know’ or ‘take the decision’ (23) we support Macleod et al. who suggest that pre-test counselling should focus more on the impact of the result, rather than the advantages and disadvantages of testing. In contrast, participants who were ‘evolving towards it’ (23), or who were undecided when they first attended, wanted the opportunity to have these discussions – and could change their mind about proceeding.

The waiting time was particularly stressful for YP when they felt they did not have clear information about how many pre-test appointments they were likely to attend and the time span between each. In addition, there was confusion when a YP expected the clinic to contact them about appointments, but clinic practice was for the YP to make contact. The data supports reports that genetics professionals are cautious when counselling YP for PT and may delay testing – using strategies such as prolonging the length of the testing process and waiting for YP to make contact (12). In contrast, we also observed the detrimental impact of shortening the recommended protocol. In either circumstance, if the process became a ‘battle to get tested’ it added to distress and had a negative impact on relationships (12).

The findings of the present study draw attention to individuals who discover their family history ‘out of the

blue’ and seek testing straightaway. YP who received good news in this category were able to adjust and move on, but those receiving bad news found it hard to cope, especially if they were living with a recently diagnosed parent. In this subgroup, testing could also be experienced as one more risk in a multitude of daily challenges facing impoverished YP.

Like adult studies, YP who received results which contradicted what they expected ‘deep down’ found it harder to adjust (24), whether they were a carrier or non-carrier, describing feelings of being stuck, lost and not knowing ‘what to do,’ similar to a mid-life or existential crisis (9, 25, 26). As the majority of participants expressed they ‘lived like gene-carriers’ there were complex consequences of receiving a good result (6, 9). In contrast, we also found YP who did not believe ‘deep down’ that they were carriers – testing to ‘prove others wrong’ or proceed with having children. Consequently, testing positive was unexpected and created more uncertainty (4, 9, 25) – and was a double bind if the YP had little psychological or material resources.

A final important finding was that some YP had not anticipated the extent to which their result could impact on family dynamics and family relationships. In particular, how testing could lead to changes in family roles such as becoming a young adult carer, changes in family membership and changes in family communication, e.g. who speaks to whom and levels of openness (18). The complex and far reaching impact of PT on family dynamics and family communication may be another aspect that YP have little personal experience of and find hard to anticipate.

Strengths and limits

A considerable strength of this study is that it reports the retrospective and prospective experiences of YP who have sought PT for HD. Offering a range of options for taking part and recruiting through the Grampian clinic and SHA meant we were able to recruit more diverse participants – including YP who were candid about their adjustment difficulties. A limit of the study is that we only identified three young men to invite,

Table 2. Implications for practice

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- As well as testing YP may be seeking support to deal with emotions such as anger, intrusive thoughts or grief and loss. Refer elsewhere if necessary, e.g. Huntington’s Association, Psychology or Peer Support.
 - Explore understanding of potential impact on family dynamics and family communication whether receive a carrier or non-carrier result.
 - Explore YP’s beliefs about their genetic status and discuss potential impact of an unexpected result.
 - Discuss whether testing will help or hinder in dealing with identity issues, such as becoming a carer or becoming pre-symptomatic, being adopted or fostered, being a responsible parent.
 - Encourage use of available family support before and after testing, this need not be through nuclear family e.g. aunt or uncle, grandparents, cousins.
 - Avoid duplication of services if young person also receiving support elsewhere.
 - Be clear about whose responsibility it is to make further appointments i.e. the young person or clinic.
 - Provide clear and engaging written and verbal information about PT protocol, especially number of pre-test appointments and time span between each – signpost to resources such as HDYO www.hdyo.org.
 - Consider if it at increased risk of crisis in time between venepuncture and result and facilitate additional support or intervention.
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none of whom wanted to take part, even though similar numbers of young men and young women access testing in the UK (27). Future studies should explore any differences in adjustment related to gender and age, e.g. between mature adolescents and young adults in their 20s. Future research is also needed to explore the long-term outcomes for YP who seek PT for HD and any interventions that might help gene carriers in the intervening years before diagnosis. We outline key recommendations for practice in Table 2.

Conclusions

Whilst YP's experiences of PT for HD are generally similar to those of adults, testing helped or interfered with key issues related to this age and stage, e.g. establishment of personal identity, separation from parents and the development of peer/intimate relationships. Most YP felt that the pre-test period was too long but those who had clear and engaging information about the PT protocol and the likely number and interval between appointments coped better. YP who were separating from parents and/or had traumatic experiences of growing up with HD could be very isolated during the test process. If YP sought testing when a parent's condition had significantly declined, the impact of grief and loss may need additional consideration. In the post-test period, the most vulnerable YP were those who had received results which contradicted what they felt 'deep down', and those who did not anticipate the impact testing could have on family dynamics. Future research is needed to explore the long term outcomes for YP who seek PT for HD and any interventions that might help in the post-test period.

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