

# Impact of Huntington Disease Gene-Positive Status on Pre-Symptomatic Young Adults and Recommendations for Genetic Counselors

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**Abstract** Huntington disease (HD) is an autosomal dominant, progressive neurodegenerative disorder for which there is no cure. Predictive testing for HD is available to asymptomatic at-risk individuals. Approximately half of the population undergoing predictive testing for HD consists of young adults ( $\leq 35$  years old). Finishing one's education, starting a career, engaging in romantic relationships and becoming a parent are key milestones of young adulthood. We conducted a qualitative study to explore how testing gene-positive for HD influences young adults' attainment of these milestones, and to identify major challenges that pre-symptomatic young adults face to aid the development of targeted genetic counseling. Results of our study demonstrate that 1) knowing one's gene-positive status results in an urgency to reach milestones and positively changes young adults' approach to life; 2) testing positive influences young adults' education and career choices, romantic

relationships, and family planning; 3) young adults desire flexible and tailored genetic counseling to address needs and concerns unique to this population. Findings of this study contribute to the understanding of the impact of predictive testing for HD on young adults, and highlight issues unique to this population that call for further research, intervention and advocacy.

**Keywords** Huntington disease · Genetic counseling · Predictive testing · Pre-symptomatic · Gene-positive · Young adults

## Introduction

Huntington disease (HD) is an autosomal dominant neurodegenerative disorder that causes involuntary movements (chorea), cognitive decline and psychiatric manifestations (Sturrock and Leavitt 2010). The mean age of symptom onset is approximately 45 years, and death usually occurs within 17–20 years after symptoms (Myers 2004; Ross et al. 2014). There is currently no cure for HD, and treatment focuses on symptom management (Sturrock and Leavitt 2010). Predictive testing for HD has been available for over 20 years to asymptomatic at-risk individuals. Young adults ( $\leq 35$  years old) make up approximately half of the population that undergoes predictive testing (Creighton et al. 2003; Dufrasne et al. 2011; Harper et al. 2000; Krukenberg et al. 2013; Scuffham and MacMillan 2014; Trembath et al. 2006). The most frequently quoted motivations for seeking predictive testing are family planning, planning for the future in general, and reducing uncertainty (Dufrasne et al. 2011; Scuffham and MacMillan 2014). The international guidelines for predictive testing of HD recommend pre-test counseling, a minimum one-month waiting period between presentation

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of pre-test information and the decision to have predictive testing, as well as post-test counseling, in order to ensure informed consent and minimize adverse psychosocial outcome (International Huntington Association and the World Federation of Neurology Research Group on Huntington's Chorea's 1994; MacLeod et al. 2013). Most studies and reviews exploring the psychological impact of testing positive report an overall successful adaptation in the first few years following predictive testing (Broadstock et al. 2000; Decruyenaere et al. 2003; Gargiulo et al. 2009; Meiser and Dunn 2000). However, distress levels may start to rise in gene-positive individuals as the time of possible disease onset approaches (Timman et al. 2004).

The definition of young adulthood varies, but in this study, we refer to "young adults" as individuals between 18 and 35 years of age. Arnett (2000) describes the younger years of this stage (18-late 20s) as a time of possibilities, exploration and optimism about the future. The later years (30+) are described as a time of reaching self-sufficiency, learning consideration for others, becoming a parent, and transitioning to middle adulthood (Arnett 2004; Erikson 1982). Key milestones in young adulthood include completing one's education, starting a career, engaging in romantic relationships and becoming a parent. Current literature on the impact of predictive testing for HD on young adults is limited and rarely focuses on predictive testing's impact on the milestones of young adulthood.

We conducted a qualitative study to explore if and how the knowledge of HD gene-positive status influences pre-symptomatic young adults' attainment of milestones, including education and career, romantic relationships, and family planning. We also aimed to identify major challenges that pre-symptomatic young adults face to aid the development of targeted genetic counseling for this population.

## Methods

### Participants

Eligible participants were 18–35 years of age, spoke English, had HD predictive genetic testing and had received a positive result at least 6 months before the time of study enrollment. Those who reportedly received a clinical diagnosis of HD were excluded from the study. An online recruitment survey was used to screen potential participants for eligibility, and to collect basic demographic and contact information.

### Recruitment

Information about the study and a link to the recruitment survey were posted on the websites and Facebook pages of the Huntington's Disease Society of America (HDSA) National Youth Alliance (NYA) and the Huntington's Disease Youth

Organization (HDYO). HD support groups across the nation were identified from the HDSA website (<http://hdsa.org/about-hdsa/locate-resources/>). A total of 144 support group facilitators were contacted by email and asked to help distribute information about the study to their group members.

Recruitment was from January to April of 2015. One hundred and thirteen individuals accessed the recruitment survey. Nineteen individuals met the eligibility criteria and completed the survey. The 19 individuals were then contacted to schedule an interview. Four individuals did not respond to three attempts at making contact. Fifteen participants (79 %) completed the study, 14 via phone and one via an online questionnaire.

### Data Collection

A semi-structured, open-ended interview guide was created with the collaboration of five investigators (PG, JHF, LK, CES and AHK). The main questions explored 1) if and how the participants' knowledge of their HD gene-positive status influenced milestones of young adulthood—career and education, romantic relationships, and family planning; 2) participants' experiences of predictive testing process; and 3) participants' suggestions for genetic counselors.

Fourteen phone interviews, lasting approximately 30–60 min, were conducted by PG and were digitally recorded. One phone interview was not included in the study due to the audio recording's poor quality. The remaining 13 phone interviews were transcribed verbatim. Another interview was completed, by request of the participant, in the form of a written online questionnaire that asked the same main questions in the interview guide without prompts. A total of 14 interviews (13 phone interviews and one written) were used for data analysis.

### Data Analysis

The interview transcripts were analyzed using a combination of grounded theory and thematic analysis (Miles et al. 2014). The interview transcripts were analyzed both inductively by understanding themes and patterns within the data, and deductively by drawing on frameworks from previous research and theories. In the first round of coding, PG and AHK independently coded the first four transcripts. Consistency between the coders was reached before PG completed coding of the remaining transcripts. In a second round of coding, data from different participants were compared for similarities and differences to ensure inter-case consistency. The major themes were identified from comparing, contrasting and categorizing the codes. Three additional investigators, CES and LK, who have extensive clinical experience in counseling young adults with HD, and JHF, who has expertise in qualitative healthcare research, contributed to the final analysis and validation of findings.

## Results

### Characteristics of the Sample

Fourteen participants (12 females and 2 males) were interviewed. Table 1 summarizes the socio-demographic characteristics of the study sample. At the time of enrollment, an average of 4 years (range 1–10 years) had lapsed since the participants received their positive results. The names used to refer to the participants are pseudonyms.

### Urgency to Reach Milestones

The participants were very aware that their healthy years were limited, which resulted in the majority ( $n = 8$ ) choosing to

expedite the attainment of milestones of young adulthood. After learning her gene-positive status, Erica (29, F) decided to speed up the pursuit of her teaching career:

*Had I not been gene-positive, I maybe would have started a family, taken my time, worked in an office maybe ten years, and then got a doctorate and then taught in my 40s. And now it's like - you know what - teaching is the end goal, so do it, do it, do it. Work harder. Achieve it faster.*

Brian (23, M) expressed desire to establish a serious romantic relationship and start a family sooner, after learning that he was gene-positive:

*If I have children when I'm 29, then it'll probably be ok. I think I'll probably be able to raise them and be healthy.... So I'm trying to date people who are maybe a little older than me and a little more serious about it than if I would without this information [that I'm gene-positive].*

**Table 1** Socio-demographic characteristics of the sample ( $n = 14$ )

	Number of Participants (Percentage of Total)
Age (Years)	
Range	20–33
Median	29
Sex	
Men	2 (14 %)
Women	12 (86 %)
Educational Level	
High school or lower	2 (14 %)
College degree	7 (50 %)
Masters degree or higher	5 (36 %)
Employment Status	
Student	3 (21 %)
Employed	10 (72 %)
Unemployed	1 (7 %)
Relationship Status	
Single	8 (57 %)
In a relationship	2 (14 %)
Married	4 (29 %)
Number of Children	
No children	12 (86 %)
1 child	1 (7 %)
> 1 child	1 (7 %)
Country of Residence	
USA	13 (93 %)
UK	1 (7 %)
Recruitment Source	
HDSA NYA	5 (36 %)
HDYO	2 (14 %)
Support groups	3 (21 %)
Friends and family	3 (21 %)
Other	1 (7 %)

### Appreciation of Time and Becoming a “Better Person”

None of the participants expressed regret for their decision to have predictive testing. Eleven participants expressed a greater appreciation of time after receiving a gene-positive result. Specific examples included identifying clearer priorities for life, living in the moment and letting go of trivial worries. After testing positive, Heidi (29, F) re-defined her priorities:

*[I focus on] spending time with my family, being with my kids more and visiting family that I haven't seen in a while—just doing things that normally I would put off until the last moment or not think of as important, when they really should be.*

The knowledge of his gene-positive status led James (24, M) to live more in the moment:

*I think I kind of live life differently. I try to enjoy life, live each day as if it's the last day. ... I just try to enjoy everything, take everything in and enjoy it.*

Fiona (30, F), who used to let the details preoccupy her, now learned to let trivial worries go.

*Knowing that time is limited makes things that would otherwise seem like bigger deals really seem like not a deal at all.*

Six participants believed that knowing their gene-positive status had made them mature faster and/or “become a better person.” Julie (23, F) previously did not care as much about where her life was heading. After predictive testing, she felt motivated to pursue an independent and successful life:

*Before knowing my results, I didn't really care about what I did with my life as much as I do now. Now after knowing, I want to succeed more than I wanted before. I want to do better for myself. ... I'm extremely independent now, and I like that.*

Kara (29, F) believed that testing positive helped her mature and become a better person:

*I actually think it [testing positive] helps me to be a better person. ... I had to basically grow up really quick. ... You hear a lot of older people say that you just realize that you gotta be happy with yourself and your life, and I learned that pretty quick.*

### **Influence on Career and Education**

The influence of testing gene-positive for HD on young adults' education and career paths falls into three categories. 1) Four participants reported no change in their education and career choices after testing positive. Three of these individuals were already on a stable career path before predictive testing.

2) Eight participants reported making minor changes to their education and career plans based on the gene-positive result. It was typical for these participants to experience a period of a) wanting to make money within a limited number of healthy years and/or b) concern for the eventual onset of symptoms leading to loss of a career, both of which made them consider curtailing pursuit of higher education and/or changing career directions. However, eventually other factors, especially personal interest, predominated and re-directed them back on the original track. Georgie (33, F) described putting her education plan on hold for a short period due to the wish to save money for the future:

*[After I tested positive,] I didn't want to go back to school, because it cost money and I wanted to make sure that I was not spending money now and I could save it for the future. But then I ended up going back anyway, because it just made sense.*

It was also typical for participants to choose a more practical field within their original career aspirations to secure a stable job with a good salary, in order to prepare for the onset of symptoms in their future. Danielle (23, F) initially was interested in pursuing research in physics. After receiving

her gene-positive result, Danielle's plan shifted to a related but more practical direction—becoming an engineer:

*I wanted to stay in school and wanted to maybe possibly do research. ... I realized that would take a long time and wouldn't be practical so decided not to do that... I ended up studying electrical engineering and that was something I need to finish in the next couple years and ideally get a job that would pay me well so that I can have the safety net.*

3) Two participants made drastic changes in career direction after testing positive. Nikki (31, F) reported that she was “pushed out of” the job that she enjoyed and of which she was proud, possibly due to discrimination. She felt forced to find another career path but eventually discovered a new interest in journalism that would give her a chance to expose abusive acts including discrimination. Megan (30, F) already had started a career in accounting at the time of predictive testing. When she returned to work after testing positive, she discovered that her career interest completely changed:

*When I went back to work, I had a very difficult time finding purpose in what I was doing, to the point where I quit my job, because I felt like I was wasting my life, and I felt like I needed to do something to help other people who are going through the same thing [living in the impact of HD]. So I have a part-time job now, and I volunteer with a local chapter [of HDSA] so that I can do something to help other people going through the same thing.*

### **Influence on Romantic Relationships**

Seven participants, regardless of marital status, felt their gene-positive status and developing HD symptoms in the future was a “burden” or a “deficit,” which made them feel less romantically desirable. Lillian (20, F) who was in a relationship with her partner since before predictive testing, felt that she could not break up with her partner because of the concern that no one else would want to be with her.

*In a way, I feel like I can't split up with my boyfriend, because I don't feel like anybody else would want to be with someone, knowing that they've got HD [mutation] and that they're going to get that [disease] when they're older.*

Most of these participants were able gradually to see their gene-positive status as only one aspect of their identity and overcome or lessen the feeling of a “deficit” or “burden.” A couple of years after receiving her positive result, Amanda

(29, F) gained the perspective that many other people also have obstacles in their romantic relationships.

*I've got to a point where I see it [HD] as a potential thing if science doesn't fix, will happen in the future, but that future is a long time away. There are people who have way worse issues that are affecting them right now and that affects their romantic prospective and their ability to be in a healthy romantic relationships. I feel now it's just one factor in many. I don't see myself to be at disadvantage any more.*

The unmarried participants ( $n = 10$ ) universally intended to disclose their gene-positive status to their dating partners. However, fear or experiences of their being rejected by dating partners following disclosure made it difficult to determine the best time to disclose one's gene-positive status: disclosing early would not give enough time for the partner to get to know the participant, but disclosing later may cause a waste of time and emotional investment in the event the partner rejects them.

Eight of the unmarried participants became more selective of romantic partners after predictive testing. They looked for mates who could fully understand and accept HD in their future, and who respected and supported their limitations in reproductive options. Erica (29, F) is an example:

*I definitely know that my approach [to romantic partners] changed. I like to put it out there [that I'm HD gene-positive].... If they say, "hey, no problem. I admire you," then cool. But if they're hesitant, then, "okay, not strong enough. Next one." ... [I only] date people that I feel are going to be worthy and going to respect my obstacles of family planning, whatever that may be, what we mutually decide.*

At the time of the interviews, four participants were married, and all were married to the same partners as before their predictive testing. The married participants reported that their partners were fully informed in the predictive testing process. Three of the married participants felt that testing positive had made their relationships stronger. The other described no change in her relationship's quality. Fiona (30, F) described that after going through predictive testing, both she and her husband were able to recognize the importance of their marriage.

*When I was tested gene-positive, it was devastating for both of us. I think he felt it equally to me, if not more so, just given his care and concern for me. ... I think, in a way, if anything, the diagnosis allows us to know how important that is, to really nurture that positive relationship and value every day we have together.*

## Influence on Family Planning

The influence of testing gene-positive on female participants' ( $n = 12$ ) family planning decisions can be categorized into 3 groups. 1) Ten female participants, after testing gene-positive, still wanted to have children, or to have more children if they already had children before having had predictive testing. However, all wanted to avoid having *at-risk* children. They generally preferred pre-implantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to adoption or prenatal diagnosis as a means of avoiding having an at-risk child. Two participants had already gone through PGD with IVF at the time of the study, and one of them successfully conceived a gene-negative pregnancy. However, five of the participants expressed that the high cost of multiple cycles of IVF might make IVF with PGD inaccessible. 2) One female participant decided not to have children at all, not only because she wanted to avoid transmitting the mutation, but also because she did not want a child to experience taking care of a parent with HD. 3) Another female participant, Julie (23, F), was not opposed to conceiving an at risk pregnancy, if it happened. But she described that having children became less of a priority:

*Before I got tested, all I wanted was having kids. But now, whether it happens or not, I'm not gonna be upset either way. Because it's a win-win situation, having kids or not having kids. Either I have kids and have that joy, or I don't have kids and don't have to put somebody else through that [living at risk for HD].*

The two male participants who were both single had less concrete ideas for family planning compared to the female participants. The male participants deferred decisions regarding children to their future female partners, who would be the ones carrying their pregnancies. Brian (23, M) described how his future partner would play important roles in his family planning decisions:

*There are certain health implications for whoever will be carrying my child. In vitro fertilization is a medical procedure. I haven't really gone down that road and thought about it. I would need to think about it with a partner. I thought about adoption, but not seriously. These are decisions that would need to be made with a partner.*

## Concerns for Discrimination

Although participants of this study were able to adapt well to the knowledge of their gene-positive status, they pointed out some remaining concerns. Eight participants

felt that disclosing their genetic status in personal and working relationships was a major challenge they constantly faced. James (24, M) found disclosing his genetic status to be especially troublesome:

*The biggest question is when or if or how do I tell someone that I want to date about Huntington's disease and being tested. ... [Also] just in general telling people, it's very tough, because you don't know whether they are going to judge you, and how they are going to react.*

A main reason that disclosing their genetic status was difficult was the fear of discrimination. Four participants expressed worry or shared experiences of being discriminated by others due to their HD gene-positive status. Danielle (23, F) was concerned about being discriminated by employers and insurance companies:

*You have to be careful with who you talk about it because, for example, if you tell somebody and an employer finds out, they can choose not to hire you. Or insurance companies can still - they are technically not allowed, it's illegal - but they still discriminate against you.*

Nikki (31, F) reported that she lost her job soon after learning her gene-positive status. She believed this was due to discrimination.

*Once I found out that I was gene-positive, I got pushed out of my job. It was unfair and totally against GINA (Genetic Information Nondiscrimination Act), but I couldn't do anything about it although I tried. This was a very depressing and difficult time.*

### Feeling Lonely

Three participants experienced loneliness after testing gene-positive due to a lack of sufficient peer support. Brian (23, M) is an example:

*I talk to lots of people, but it's not the same as someone who - especially a young person - who's dealing with the same situation [of testing positive].... It can be lonely to know you're gene-positive.*

### Comments on the Predictive Testing Process

Eleven participants received genetic counseling as part of their testing process. Participants' evaluation of their counseling experience varied greatly. Approximately half ( $n = 6$ ) found

the genetic counseling helpful. Specific aspects of counseling that were useful include assessment of emotional readiness for predictive testing, anticipatory guidance and information about obtaining life and disability insurances. Six participants felt the testing process was a "hurdle," an "obstacle" or unnecessarily lengthy. The participants who were more likely to find the testing process to be a "hurdle" were those who 1) tested at a center where the process was particularly prolonged or involved multiple visits in addition to the pre-test and post-test counseling, 2) felt they had carefully thought through the decision for predictive testing prior to counseling, and 3) "led a busy life."

Four participants expressed appreciation or desire for long-term follow-up after their results were given. Julie (23, F) said, "I liked that I kept in contact with them since." Megan (29, F) who did not receive follow-up said, "I wish somebody would have followed up with me after I tested positive."

Four participants, two of whom had genetic counseling, brought up unmet needs for some categories of practical information—alternative reproductive options, especially PGD with IVF, and ways to obtain life and disability insurances.

## Discussion

This study explored how the knowledge of gene-positive status influences milestones of pre-symptomatic young adults—their education and career, romantic relationships, and family planning. Many findings from our study support previous research, and some findings are novel. Results can be grouped into three main categories: 1) changes in attitude and approach towards life, 2) influences on milestones of adulthood, and 3) suggestions for genetic counselors.

### Changes in Approach and Attitude Towards Life

A prominent theme of our study is that pre-symptomatic young adults have a high level of awareness that their healthy years are limited by the future onset of HD symptoms. This phenomenon has not been described in previous studies on HD gene-positive pre-symptomatic young adults. Arnett (2000) describes the ages 18–29 years as a period of prolonged exploration of the possibilities in career and romantic relationships before choosing a more definitive path. When faced with the reality of being gene-positive and a curtailed number of healthy years, pre-symptomatic young adults in our study often felt a need to expedite this exploration process. However, this urgency was not negatively framed with a focus on lost opportunities. These young adults did not settle for a career path that was convenient although of little personal interest; they chose to expedite the pursuit of their interest, taking fewer breaks on the way. Similarly, they did not settle for a romantic partner just to be able to start a family; they

chose to look for mates who were more mature and serious about settling down.

Young adults in this study experienced positive changes after testing positive, including having clearer priorities, living more in the moment, letting go of trivial worries and feeling more motivated, all of which they described as helping them mature and become “a better person.” Similar changes have been described in previous studies looking at the impact of receiving a predictive test result, positive or negative, in young people (Duncan et al. 2007, 2008). Previous studies found these positive changes to be a possible result of relief from uncertainty about their genetic status, which “constituted a barrier in young people’s lives and prevented them from moving forward” (Duncan et al. 2007). Our study suggests that the appreciation of their limited healthy years was also a contributing factor to these positive changes.

### **Influence on Milestones of Young Adulthood**

The knowledge of being gene-positive for HD influenced the young adults’ age-specific milestones—education and career paths, romantic relationships, and family planning. To our knowledge, our study is the first to explore the influence of HD genetic status on education and career paths of young adults. It is typical for young adults to take time trying different majors in school and taking on several different jobs before committing to a career that best suits their interests and skills (Arnett 2004). In addition to considering personal interests and skill profile, the young adults in our study also had their genetic status to take into consideration when choosing a career. Having testing at a young age means that the young adults were often still at the early phase of career exploration when they learned their gene-positive status, which allowed them to make adjustments to their career path. Results of our study suggest that, although it did play a role for most young adults, genetic status rarely dramatically changed young adults’ career direction. Though uncommon, a drastic change in career *direction* may happen as a result of a drastic change in career *interest* after testing positive. However, the knowledge of their gene-positive status made the young adults think about practical aspects of their developing career, such as how long it would take to reach a stable stage, the ability to make stable financial gain, and the possibility of being forced to retire once symptoms begin.

The impact of testing positive on romantic relationships differed depending on the young adults’ relationship status. The unmarried participants intended to disclose their gene-positive status to dating partners. In the limited literature about the impact of being HD gene-positive on dating, Klitzman and Sweeney (2011) point out that one reason gene-positive older adults look for a prospective mate is in anticipation of needing a caregiver. Young adults in our study, however, did not emphasize the intention of looking for a caregiver, perhaps

because the onset of symptoms and becoming dependent on a caregiver is still far into the future. Instead, young adults focused more on the partner’s ability to appreciate their identity and personality beyond HD and the partner’s willingness to support them through obstacles that might arise as they build a family. In addition, young adults in our study identified that when and how to disclose genetic risk to a dating partner was a complicated and difficult issue. Similar observations have been made by Klitzman and Sweeney (2011) in a non-age-selected population of HD gene-positive individuals. The timing of disclosure was especially challenging for young adults of our study because of their urgency to settle into a stable relationship and start a family. Moreover, Klitzman and Sweeney (2011) described that one potential result of wanting to avoid disclosure for an older adult is choosing not to date. This was not found to be a common phenomenon in our sample of young adults. Perhaps this was due to the hopeful nature common for this age group as Arnett (2004) points out. Although faced with the potential of stigma and rejection, young adults have not given up the hope of finding a meaningful, loving relationship.

The four married adults in our study described the impact of testing positive for HD to be either positive, making the relationship stronger, or neutral, resulting in no change in the relationship’s quality. This finding is in agreement with those of previous studies regarding the impact of genetic testing for HD on committed romantic relationships in non-age-selected groups. Decruyenaere et al. (2004) found that five years after testing positive, most couples reported no change or an improvement in the quality of their relationships. Richards (2004) also found that five years after testing, most couples reported that receiving a positive predictive test result had little or no adverse effect on their relationships.

Regarding reproductive choices, we observed that young adults experienced an overall decrease in their desire to have children after testing positive, due to the wish to avoid transmitting the mutation to a child, the perceived moral obligation to stop HD, and the desire to avoid putting a child through the experience of taking care of an affected parent. Similar thoughts have been described of a group of gene-positive adults who had chosen not to have children (Quaid et al. 2010). Our study shows that adoption and prenatal diagnosis were not preferred by the young adults as ways to avoid transmitting HD, consistent with prior studies (Creighton et al. 2003; Evers-Kiebooms et al. 2002; Maat-Kievit et al. 1999; Richards and Rea 2005; Simpson and Harper 2001; Simpson et al. 2002). Young adults from our study showed a strong interest in exploring and utilizing PGD with IVF, which is in contrast to previous observation of low uptake of PGD by a gene-positive population consisting of both young and middle-aged adults (Richards and Rea 2005). We speculate that this contrast may be due to 1) an increase in the overall utilization of assisted reproductive technologies for prevention

of genetic diseases in recent years (Collins 2013), 2) that young adults may be especially aware of and accepting of assisted reproductive technologies compared to older adults, and 3) that young adults may possess a particularly strong desire to avoid transmitting HD compared to the non-age-selected gene-positive population. However, the high cost and lack of insurance coverage of IVF pose a major obstacle in young adults' access to this alternative reproductive method. If the cost of IVF were lowered or insurance coverage increased, even higher interest and uptake of this technology may be observed. This trend has been described in Belgium where PGD was covered by public health insurance beginning in 2003 (Decruyenaere et al. 2007).

### Suggestions for Genetic Counselors

This study asked the young adults to reflect on their predictive testing process and provide suggestions for genetic counselors. We found that the appraisal of their genetic counseling experience varied drastically among young adults, with great appreciation for counseling on one extreme and seeing counseling as a hurdle on the other extreme. Young adults' preferences for the length, depth and content of genetic counseling varied greatly, and depended on many factors, including the young adult's level of eagerness and readiness for predictive testing before coming to counseling, their lifestyle, and whether their need for particular types of information (such as information about PGD and IVF, and life and disability insurances) were met. These findings point out the importance of providing flexible and tailored genetic counseling for young adults. Another recent study of young people's (age 15–25 years) experience with predictive genetic testing for adult-onset genetic diseases including HD also identified the same need for tailored genetic counseling (MacLeod et al. 2014). These findings also suggest the need for more in-depth studies that investigate how to tailor genetic counseling of HD predictive testing for young adults. In addition, to make information and resources regarding PGD and IVF, and life and disability insurances more accessible to gene-positive young adults, the effort of professionals and organizations beyond genetic counselors is also essential. HD support groups and advocacy organizations are recommended to develop informational materials on these topics and keep inventories of such resources.

Our study found that long-term follow-up counseling after delivery of positive results was also a common request of young adults. Follow up visits may be opportunities to discuss the common concerns identified in gene-positive young adults, including worries about discrimination and loneliness. Results of our study suggest that discrimination in the social, employment and insurance settings is a common concern of gene-positive young adults. In a quantitative analysis of 293 individuals who were at risk or gene-positive for HD,

Bombard et al. (2011) found that although genetic discrimination is experienced by all age groups, younger individuals are more likely to experience discrimination in the insurance setting. No statistically significant association was found between being young and increased experience of discrimination in the employment setting (Bombard et al. 2011). However, not experiencing increased discrimination in the employment setting does not imply that discrimination in the employment setting is not a main concern of young adults. Young adults have less experience in professional interactions than older adults; they may need more information and guidance on protecting themselves from genetic discrimination in the employment setting.

Another common concern identified in our study is the feeling of loneliness attributed to a lack of understanding and support from peers. Genetic counselors are recommended to keep updated with information of peer support groups and resources targeted to young adults, and be ready to provide such information to their young adult patients who test positive. However, the efforts of genetic counselors alone may not be enough, as peers outside of the HD community may not be sufficiently educated and experienced regarding HD to provide adequate and sustained support for gene-positive pre-symptomatic young adults. Therefore enhancing social awareness and education about HD are also crucial in addressing the issue of loneliness.

### Study Limitations

Our study is based on a small sample of self-selected, highly educated young people, primarily female. Participants who choose to take part in research may also tend to be those who are coping best or who are most troubled. Therefore, the experiences conveyed may not be representative of the general population of pre-symptomatic gene-positive young adults. The experiences described by the study sample were retrospective, and are thus vulnerable to recall bias. Future studies would be strengthened by the inclusion of baseline measures in addition to post-test measures, by sampling a wider population, or by including a control group of untested at-risk young adults or gene-negative young adults.

### Conclusion

This qualitative study of HD gene-positive pre-symptomatic young adults found that 1) knowing one's gene-positive status results in an urgency to reach milestones of young adulthood and positive changes in young adults' approach to life; 2) testing positive influences young adults' education and career choices, romantic relationships, and family planning; and 3) young adults desire flexible and tailored genetic counseling to address needs and concerns unique to



this population. Findings of this study contribute to the understanding of the impact of predictive testing for HD on young adults. They highlight issues unique to the gene-positive pre-symptomatic young adult population that call for further research, intervention and advocacy from professionals within the health and social systems.

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### Compliance with Ethical Standards

**Conflict of Interest** Andrea K Hanson-Kahn facilitates a support group for individuals with HD for which she receives a stipend from the HDSA. Authors Ping Gong, Joanna H Fanos, Lauren Kory and Carly E Siskind declare that they have no conflict of interest.

**Human Studies and Informed Consent** The project was approved by the Institutional Review Board of Stanford University. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

**Animal Studies** No animal studies were carried out by the authors for this article.

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