NIH Public Access
Author Manuscript
J Genet Couns. Author manuscript; available in PMC 2010 January 27.

Published in final edited form as:
J Genet Couns. 2008 August ; 17(4): 327–335. doi:10.1007/s10897-008-9151-6.

Experiences of teens living in the shadow of Huntington Disease

Kathleen J. H. Sparbel[1], **Martha Driessnack**[1], **Janet K. Williams**[1], **Debra L. Schutte**[1], **Toni Tripp-Reimer**[1], **Meghan McGonigal-Kenney**[1], **Lori Jarmon**[1], and **Jane S. Paulsen**[2]

[1] College of Nursing, The University of Iowa, Iowa City, Iowa

[2] College of Medicine, The University of Iowa, Iowa City, Iowa

Abstract

Research on families with Huntington Disease (HD) has primarily focused on adult decision-making surrounding predictive genetic testing and caregiver stress. Little is known about the experiences of teens living in these families. This qualitative study explored the experiences of 32 teens living in families with HD. Six focus groups were conducted across the U.S. and Canada. Data were analyzed using descriptive qualitative analysis. HD appeared to cast a shadow over the experiences described by teens. Four themes were identified: *Watching and waiting*; *Alone in the midst of others*; *Family life is kind of hard*; and *Having to be like an adult*. These experiences highlight the need for genetic counselors, health care providers, and school personnel to be aware of issues facing teens living in families with HD. Recognizing patterns of teen experiences may help health care providers develop strategies to support coping by teens in HD families.

Keywords

Huntington disease; adolescent; qualitative research; focus groups

INTRODUCTION

Huntington disease (HD) is a noncurable, neurodegenerative condition that is inherited in an autosomal dominant manner. The HD gene (*IT15)* [4p16.3] is the only gene associated with Huntington disease and a trinucleotide CAG expansion is the only mutation observed (Warby, Graham, & Hayden, 2007). Alleles in the HD gene are classified as normal, intermediate, or HD-causing, depending on the number of CAG repeats. HD-causing alleles are further classified as reduced-penetrance (36-39 CAG repeats) or full-penetrance (40 or more CAG repeats). Individuals with 36 or more CAG repeats are at risk, but may not ever develop HD symptoms. However, those individuals with 40 or CAG repeats know with almost 100% certainty that they will develop HD symptoms in their lifetime.

The mean age for onset of symptoms is during the mid-childrearing years, when many families already have children. As HD progresses, the person with HD (pHD) requires increasing assistance with day-to-day functioning and ultimately becomes totally dependent on others for care. The majority of pHDs are initially cared for by their families at home, but many eventually require care in nursing homes or other long term care facility toward the end of life (Bergeron, 2004; Paulsen et al., 2006; Rosenblatt, Ranene, Nance, & Paulsen, 1999; Williams et al., 2007). Because the mean age of death is 15 years from the onset of symptoms, teens in these families may not only witness the onset of HD symptoms in a parent but may also be witness

Corresponding Author: Kathleen J.H. Sparbel 914 North Pine Street Davenport, Iowa 52804-4134 Phone: 563 940 0806 Fax: 309 757 9473 Kathleen-sparbel@uiowa.edu.

NIH-PA Author Manuscript
NIH-PA Author Manuscript
NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

to the disease's progression, the transition of the pHD from home to long term care facility, and death.

Studies of families with HD have focused primarily on caregiver stress in adult family members and decision-making dilemmas surrounding predictive genetic testing for the pHD and other adult family members (Cox, 2003; Sobel & Cowen, 2000; Taylor, 2004; Williams, Schutte, Holkup, Evers, & Muilenburg, 2000). Teens growing up in a family with HD have rarely been included in family studies. This may be because the caregiving research has primarily focused on adult caregivers in the home. Further, decision-making studies have also focused on adults, since current guidelines dictate that teens at risk for HD under the age of 18 are not yet eligible for predictive genetic testing.

For youth at risk for HD, the research has primarily focused on whether individuals must be 18 years of age or older to receive predictive genetic testing for HD (Fryer, 2000; Gaff, Lynch, & Spencer, 2006; Richards, 2006). Although this age designation does not apply for all genetic diseases, it is applied to adult-onset diseases, like HD, for which no known intervention is available to delay disease onset, clinical progression, or fatality. The case *against* predictive genetic testing for minors (< 18) in these circumstances most often claims that maturity of judgment in decision-making is not sufficiently developed until late adolescence (18-21) (Cauffman & Steinberg, 2000; Richards, 2006) as well as the potential for long-term psychological effects and harm (Richards, 2006; Timman, Roos, Maat-Kievit, & Tibben, 2004). In addition, the potential for genetic discrimination and other long-term consequences of the identification of genetic risk in childhood or adolescence on later insurance options, health care support, education, and employment opportunities are unknown (Shinaman, Bain, & Shoulson, 2003). The argument *for* predictive testing for minors in these circumstances introduces the possibility that psychological harm may be caused by withholding testing (Binedell, Soldan, Scourfield, & Harper, 1996; Duncan, 2005; Duncan et al., 2008). These researchers reference the paucity of research with teens and the lack of empirical evidence that predictive genetic testing in these circumstances causes harm. Instead, they highlight the potential benefits for minors, as they begin to focus on decision-making about their futures, including education, career, and reproductive behaviors. They also emphasize the potential for psychological relief from the not knowing or uncertainty (Duncan, 2005; Duncan & Delatycki, 2006; Duncan et al., 2008.; Richards, 2006).

Adolescence is a critical period for the establishment of lifelong health-related behaviors and attitudes. It is also a time when developmental and health trajectories can be altered in positive or negative directions depending on the interpersonal contexts in which they occur (Holmbeck, 2002). To date, little research has focused on the experiences of teens growing up in families in which one of its members has HD. Research studies are now just beginning to explore teen experiences, especially in families with heritable conditions (Holt, 2006; Van der Meer et al., 2006).

Three recent studies begin to look beyond the adult experience in families with HD and highlight the need for continued exploration of teen experiences in these families (Forrest, Miedzybrodzka, van Teijlingen, McKee, & Simpson, 2007; Vamos, Hambridge, Edwards, & Conaghan, 2007; Van der Meer et al., 2006). In the first of these studies (Forrest et al., 2007), the participants included some teens, but in general were older (*M*=21). However, the authors noted a small subgroup of participants under age 18 carried out significant caregiving tasks and assumed increased responsibility for the affected parent. They caution the reader that such increased responsibility may have a detrimental impact on young caregivers' education, health, social, and emotional well being, and that the age at which a teen became aware of the inherited nature of HD could be associated with teen adjustment issues.

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

In the second study, Vamos and colleagues (2007) explored the impact of HD on family members. The research was retrospective, recalled by adults who grew up in families affected by HD. Adverse parenting experiences and the quality of family functioning were reported. Low family cohesion and expressiveness and high levels of family conflict were identified, with adverse parenting behaviors experienced from both the pHD and nonHD affected parent. The authors suggest the effects of HD on the entire family unit may be underestimated and far-reaching.

The third study (Van der Meer et al., 2006) also notes the effects on family, including children and teens. Their study focused on the presence of role reversal (i.e. child caring for parent) and parentification (i.e. when unaffected parent looks to a child as a substitute partner) in the children and teens in families with HD. Participants in this study were also adults, rather than teens, who looked back at their experiences. Being raised by a parent with HD revealed lower attachment representations in participants later in life. Unresolved and disorganized attachment representations were significantly higher if the pHD died or had psychiatric hospitalization before the participant was 18 years old.

Each of these studies underscores the potential effect of stressful life experiences for teens in families with an adult member with HD. These experiences are especially important to explore as we consider teens are already in the midst of establishing health behaviors and beliefs that will have long-term implications in terms of their understanding of health, risk, and behavior.

This early body of evidence suggests that teens in HD families may be exposed to increasing responsibilities and stressors beyond the typical developmental challenges of adolescence. In addition, adult family members may not be able to provide these teens with needed support and guidance due to the effects of HD, family stresses, or coping limitations (Vamos et al., 2007). Health care providers, teachers, and guidance counselors are often the primary adults, other than parents, teens see repeatedly throughout adolescence. The ability of these professionals to engage teens in a health-promoting process is directly tied to a readiness to hear their stories and consider the context of their lives. Yet, little is known about the experiences these teens live with and what they mean in the teens' lives.

The **purpose of this study** is to explore the experiences of teens living in families with HD by asking them, while they are still teens.

METHODS

The study presented is part of a larger sequential mixed-methods research project focused on the concerns of family members of pHDs and the strategies these family members used to deal with these concerns (Williams, Paulsen, Schutte, & Tripp-Reimer, 2001). This larger project was linked to the PREDICT-HD study and identification of participants was facilitated by collaborators in the PREDICT-HD study (Paulsen et al., 2006). The first phase of the larger project was qualitative, using focus groups of various groupings of family members, with one focus group exclusively made up of only teens. The second phase was quantitative, with the development and testing of member specific questionnaires based on the earlier qualitative findings. This article reports findings from the first phase teen focus groups. More in-depth discussions of recruitment, group composition, and considerations in planning focus groups from the larger study are reported elsewhere (Pehler et al., 2008; Williams & Ayres, 2007; Williams et al., 2007).

Participants

Site coordinators at five HD centers in the US and Canada identified teens from HD families. Inclusion criteria for teen participants included individuals 14 – 18 years of age, who spoke

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

English, and had ongoing contact with a pHD. Since this was a focused study, with a clearly defined topic and limited scope, a sample size of 20-60 was estimated as adequate according to Morse (2000). Six teen focus groups were conducted across the US and Canada from 2002-2005. The groups varied in size (2-8) for a total of 32 participants. The mean age was 15.9 years and females outnumbered male participants three to one (Table 1) (Pehler et al., 2008).

Most of the teen participants (84%) were at risk for HD, while the genetic risk for a few participants (15.6%) had already been eliminated. Either the teen had tested negative as part of a diagnostic evaluation, the teen's parent or pHD was not their biologic parent or relative, or the teen's biologic parent had received a negative test result. Most pHD in these teens' families lived at home or in a nursing home, and had either received a positive predictive test for HD or had clinical manifestations of HD. Half of the teens also spoke about other extended family members with HD. Although their primary comments centered on their immediate family experiences, teens described similar observations within a larger family network.

Procedure

Teen participants were recruited through site coordinators at HD clinical and research centers who identified and contacted families with teens. Parental permission and teen assent were secured by the research team onsite prior to data collection. Each focus group was held in a small conference room at the HD centers and lasted between one and two hours. The focus group leaders, experienced advanced practice nurses with specialization in either pediatric or psychiatric nursing, moderated the groups. Teens were asked to talk about their experiences, including their concerns about the pHD and themselves. They were also asked to describe strategies they used to manage the concerns, and the effectiveness of those strategies. Focus group sessions were audio-taped, transcribed verbatim, and checked for accuracy. Institutional Review Boards (IRB) from the university and from each participating HD center approved the study.

Data Analysis

Narrative data were coded using content analysis (Knafl & Webster, 1988; Sandelowski, 2000). The team generated descriptive codes from the data, entered these into NVivo (QSR, 2000) to facilitate sorting and data management, and maintained a codebook of definitions. Differences in coding were discussed within the team until 100% agreement was reached. Codes were organized into domains and descriptive results were outlined. An incoming member of the research team conducted an independent audit of findings after initial coding of the data was completed. Focus group summaries and climate notes for each focus group detailed observations, group interactions, and main discussion points. Using an iterative process, a subset of the research team identified interpretive themes that appeared across descriptive domains, which were reviewed by the research team for clarity and consistency with the descriptive results. The audit trail, focus group summaries, and climate notes were peer-reviewed and discussed to increase the trustworthiness of the findings (Holloway & Wheeler, 2002; Morse & Field, 1995).

RESULTS

All teens shared experiences of living in a family with HD. The data portrayed a complex and often painful family environment. Teens spoke mainly of the challenging aspects of their lives. The teens were not specifically asked to address positive experiences, and very few comments reflected positive experiences or sources of support. While the nature of their comments often reflected negative aspects of their lives, the teens themselves were not negative. Instead, the

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

teens were engaging, actively supporting other group members, and particularly forthright in sharing their experiences.

The teens expressed an overall sense of living in the shadow of an overwhelming disease. This pervasive influence is reflected in each of the four themes identified in the analysis: 1*) Watching and waiting; 2*) Alone in the midst of others; 3) Family life is kind of hard; and 4) Having to be like an adult (Table 2).

Watching and waiting

Teens described their experiences of living in a family with an adult member with HD as if they were watching from and waiting in the shadow. They described an overall depressive feeling that descended and hung over their families, compromising its ability to attend to the welfare of each of its members.

Watching from the shadow of HD—Teens *watched* the disease progress in the pHD as well as the effect of that progression on other family members. One teen shared I think that the hardest thing about it has been watching my dad (pHD) suffer and knowing that there's nothing we can do… there's no cure… we have to do our best. Teens reported details of physical health issues regarding loss of motor coordination, including eating and ambulation, and corresponding safety implications for the pHD. The greatest number of their concerns involved cognitive and behavioral changes in the pHD and the impact of these changes on family life. I know she doesn't comprehend everything… it doesn't go through her brain like everyone else's …or We've noticed that his mind is getting [like] smaller… His mind is so messed up inside that everything outside needs to be perfect. A common observation was the occurrence of erratic episodes of irritability or moody behaviors in the pHD. Her emotions are up and down every day… it's like a roller coaster. Teens reported the unpredictable nature of HD and how it affected the pHD's day-to-day functioning. The big question every morning when you wake up is…is it going to be a good day or a bad day.

The teens projected their anticipation and apprehension about what may develop in the future. While it is established that HD is a progressive deteriorating condition, the timing and progression of symptoms for each individual is variable. Teens described uncertainty regarding what to expect and how to respond to the pHD's changing condition.

> You never know what the next thing's gonna be… and it's not something that anybody can tell you, and it's not something that [you know] if you're gonna be able to slow down, or if you're gonna be able to fix…

Waiting in the shadow of HD—Teens, at risk for HD, were also *waiting* in the shadow of HD. Not yet eligible for predictive genetic testing, they discussed the experience of being in the pre-testing time, not by choice, but because of their age. Only a few discussed college plans or long-term goals. Of the teens at risk for HD, only one shared a future trajectory that included career planning. Although not initiated by the focus group leader, concerns about being able to obtain predictive testing when they turned 18, were shared in each focus group. Teens reflected on whether they would get tested and what their lives would be like if they tested positive or negative.

> Well [like] the pros would be that you don't have to worry about it anymore… [like] if you get tested. And the cons are you could get it and find out about it… you feel bad, your family feels bad, all your friends feel bad… they know you will change… and that's the one thing that's stopped me from [like] thinking about getting tested.

While they rarely discussed college or future career plans, the teens did contemplate future relationships and the possibility of having children. A few talked about already being in steady

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

relationships with individuals who knew about the family HD history. One teen was already a parent herself, while another was pregnant at the time of the focus group. However, for most of the teens, considering starting a family of their own was presented as more of a daunting decision.

> From what I've seen my family members go through, if I have it, I don't want to have kids. I don't want to risk giving it to somebody else. So it's kind of hard… It's hard to deal with that kind of stuff. If I have it, I'm not gonna put other people through it. I just won't do it.

Another issue addressed across the groups related to tying the timing of predictive testing to relationships and establishing a family, I think I would ideally wait until I, after I was, [like] …had kids or married or whatever…and, I don't know, I just don't really want to know until I have to know.

Alone in the midst of others

The teens spoke about their sense of feeling alone and isolated both within their families and among their peers. For many, the pHD's progressive physical and cognitive losses compromised meaningful personal communication and family interaction and served as a source of sadness for the teens. As one teen noted, We haven't had a conversation in the last year and a half - like an actual conversation. The ability of the pHD to participate in routine activities with the teen diminished, Well, I think it's more just like the day-to-day life [like] everything becomes a little harder. The non-HD parent for some teens was often portrayed as absent or unavailable, due to assuming both financial and care giving responsibilities for the family, which in many cases necessitated having multiple jobs. My mom doesn't get to spend as much time with us because she's always with my grandpa and helping him…

In addition to constraints on parent-teen interactions, teens shared a sense of being alone in the personal reality of their genetic risk. The teens saw this risk and their experience as uniquely different from and not understood by the non-HD parent,

> She's [the non-HD parent] coming from a different point of view… I always tell her, but Mom, you don't have the risk of getting it [HD], so you can say you understand where I'm coming from… you really don't.

Teens also spoke about their peers, and how living in an HD household influenced both peer activities and the nature of their peer relationships. For many, their family situation impeded peer interactions. While peers are typically the primary source of social support during teen years, very few of the participants felt comfortable having friends in the house. They [friends] don't want to be at my house because of my mom [pHD]… they just … they don't like to be around her…and I don't blame them. Teens also talked about how hard it was to identify with their peers. None of my friends understand, ‘cause they just don't have any experience with it [HD]… so it's kind of hard cause you can't express it to them.

Almost all participants (94%) told us they did not know any other teens from a HD family. Two of the teens had met other teens through HD youth conventions. They described the experience of this additional social support as a source of increased personal strength. Several teens commented that being able to meet other teens during the focus group was itself a positive experience.

Family life is kind of hard

Family stress, conflict, and the impact of limited community and financial resources were common topics expressed and organized under this theme. When asked to express concerns about their family member, teens also talked about the wider circle of their entire family. Teens

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

talked about how HD had affected different family members. My mom [is] working two jobs… it's kind of hard for my dad to understand her stresses, and it's hard for her to understand my dad's stresses and… it just [kind of] creates an irritable mess.

Reports of conflict, particularly between parents, were common from teens whose family member with HD was symptomatic and living at home. The effects of the ongoing stress and family conflict affected the teens and their siblings, It's just kind of a trickle-down effect. Like, she'll start out getting mad, and then it'll come to him, and then it'll get to me and to my little brother.

While some extended families were supportive, teens often described broader family conflicts. These differences sometimes related to caregiving stresses of adult siblings for the pHD, or estrangement between two adult siblings who each had HD. Particularly poignant was family separation due to lack of understanding of HD:

> I guess my uncle and his wife don't fully understand what my mom is going through. They can't because they don't live in the household, and they don't see it all the time, like me and the rest of our family do. We haven't spoken to them in like two years, because my auntie got into a fight with my mom…and because of that, we've lost that part of the family. Cause they don't understand.

In the U.S., persons with HD may be too young for Medicare, and have difficulty qualifying for supplemental or disability income. The added emotional and financial costs of HD were also noted by the teens. They expressed concerns regarding the limited resources available for the care of the pHD and shared the impact this had on their families. It's kind of hard… because he can't work and there's a lot of stuff he can't do… it's financially hard on my family.

Having to be like an adult

Teens talked about the pressure of family stress in the context of their own lives. They described feelings of being overwhelmed and wanting to have fewer responsibilities. Because of changes in the roles of the adult family members and alterations in family functioning, they often assumed adult roles and responsibilities, including care giving for the pHD. Well, I have to feed her… and do chores around the house… and watch her for my dad because she can't be alone. Taking on adult roles increased the teen's awareness of the stresses on the non-HD parent. in managing caregiving, wage earning and parental roles. I worry about my mom's health. She doesn't sleep very much, I know that's hard on her. She'll get stressed out …there are very difficult days… she has to take care of everything …things add up. Teens spoke of helping to meet the needs of both parents.

> My dad [pHD]… he doesn't eat, you have to feed him, If he's not cooperating, then she has to take care of everything else…so I try to watch over her [non-HD parent] too…I'll run her errand, I'll clean the house, cook some food, so she takes care of my dad. There's always something that needs to be done.

The teens also expressed worry about who would assume the teen's added responsibilities as they prepared to leave home after high school, Next year I'll be graduating and so… I'm going to be helping out mainly for the next year, but I'm afraid when I leave, I don't know what's gonna happen. Although teens in these groups spoke about friends, and shared anecdotes about school activities in the casual conversation before and after data collection, the comments during data collection primarily focused on how their lives differed from their friends. These comments were not expressed with bitterness, but in a matter of fact manner. Some teens also described losing the chance or time to experience being a normal teen and wished for a different balance of responsibilities. I can't really be myself, I have to be [like] an adult…But I want to be a brat [you know], I want to be myself, and I can't.

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

Each of the four themes, 1) Watching and waiting; 2) Alone in the midst of others; 3) Family life is kind of hard; and 4) Having to be like an adult, speak to the experiences of teens living in families with HD. The presence of HD in these teens' families appeared to cast a shadow over the teen's lives, and highlight the need for increased attention to these teens' rarely seen (or heard about) worlds.

DISCUSSION

The findings in this study begin to shine a light on teen experiences in families with HD and how these experiences may challenge these teens' current and future wellbeing as well as influence their current and future decision-making (Forrest et al., 2007; Vamos et al., 2007; Van der Meer et al., 2006). The teens shared experiences and concerns about both of their parents, themselves, their families, and their peer relationships. Although these experiences were separated in order to identify common themes, they were not isolated from each other in the teens' descriptions of their lives. It appeared that their experiences were overshadowed by the presence of HD and unable to be interpreted without its presence. Furthermore, the teens did not choose to share experiences from outside this shadow, despite repeated opportunities.

Adolescence is often thought of as the transitional period between childhood and adulthood (Holmbeck, 2002). Typical adolescents struggle with rapid physical changes, body image, peer relationships, and identity (Driessnack, 2006). Their wellbeing depends on the teens' developing independence, sexual identify, cognitive expansion, moral maturation, and preparation for an adult role in society (Smetana, Campione-Barr, & Metzger, 2006). For those teens living in families with HD, typical adolescent struggles remain invisible in the shadow, subsumed under the challenges HD exerts on the family as a whole. The teens shared experiences that focused not on their *own* adolescent developmental changes, but instead reflected the impact of HD on their entire *families*. Further, while typical teens often complain their parent(s) are too present, these teens talked about the absence of parental presence in their lives. They watched as the parent with HD slowly disappeared emotionally and physically and the non HD parent took on additional roles, often working overtime, or at more than one job, to make ends meet.

For many teens, peer groups typically represent vehicles for movement out of and away from the family, and as such provide a means of achieving independence and individuation (Driessnack, 2006). However, many of these teens had limited peer support and involvement. Although many had friends, the teens shared that their friends could not really understand what it was like to live with a pHD, and for some, their lives at home and with their friends were kept separate. This need for peer contact and support, while securing time to be a teen, has been reported in studies of other young caregivers (Lackey & Gates, 2001; National Alliance for Caregiving, 2005). Opportunities for interaction with other teens in other HD families may offer these teens much needed peer contact and support (Table 3).

Genetic counselors acquainted with families with HD may be in an ideal position to establish trusting relationships with family members, including teens. Understanding the context of these teens' lives is also essential for health care providers and other trusted adults, such as school counselors, who can engage teens in health-promoting behaviors and opportunities (Ginsburg, 2007). Timely, supportive interventions at this critical developmental stage may prove valuable in mitigating the effect of numerous stressors on the teen and promote a healthy progression to young adulthood. The teens in this study reported feelings of being alone even though they were often surrounded by family and friends. They reported that life at home was hard and they were having to take on adult roles and responsibilities that often interfered with normal adolescent activities. This is an important call to action for anyone encountering these teens. It is especially a call to health care providers and genetic counselors who may be focusing only

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

on the pHD and adult caregivers. The findings of this study highlight the importance of asking about teens in the family and creating opportunities to begin or continue a dialogue with them.

For most of the teens in this study, this was their first opportunity to meet other teens in similar life circumstances. It was also their first opportunity to participate in research, which many found to be empowering (Pehler et al., 2008). These data suggest that teens in HD affected families may talk about their experiences when there are opportunities and those who are prepared to listen. In addition, health care providers and genetic counselors may be in the best position to provide resource information to the teens and their parents, support parents in developing the skills to talk to their teens, and identify potential peer contacts. Only two of the 32 teens attended a HD National Youth Alliance conference where resources and network opportunities are available for teens (See Table 3).

Teens in each of the focus groups addressed the issue of predictive genetic testing and decision-making dilemmas. While adolescence is a period of development during which teens begin to make many “adult” choices, from developing relationships to contemplating life trajectories and career choices, the current guidelines for predictive genetic testing for HD specify 18 years of age as the minimum age to be given the responsibility to make this decision. Some providers find the current guidelines to be unhelpful, and suggest they may need to be updated related to current concerns and practices (Multhaupt-Buell et al., 2007). The findings from this study suggest teens 14-18 years of age are actively considering this option. While we are not currently advocating for or against lowering the age when predictive testing for HD is available, our study clearly illustrates that many teens want, and need, to talk about it. Perhaps, as we understand more about teens' experiences in these families, we will find that there are teens for whom testing should be legitimate and possible.

Members of HD testing and management teams can facilitate conversations between parents, teens, and the health care community about predictive testing and decision-making these teens will face after they reach age 18 (Duncan, 2005; Duncan et al., 2008). These opportunities may help teens offset their current risk status by increasing their sense of control over their future. In their recent study, Quaid and colleagues (2007) explored the experiences of adults who chose not to discover their actual mutation status and found that many achieved a “remarkable balance between living at risk and living with hope”. The basis for this balance may be established and influenced by experiences throughout their teen years. Longitudinal studies that follow children in HD families over time may shed considerable light on how young at risk family members approach later decision-making about predictive testing and their resultant mutation status results. However, gaining access to these young at risk family members and securing their cooperation in research studies, may be difficult if their experiences are not further understood.

As advances in genomic research continue to identify inherited risk for more diseases, it is important to explore what the experience of being at *genetic* risk means to the individual and the family. The *Family Systems Genetic Illness* (*FSGI*) model may provide one way to provide context for the findings from this study. The *FSGI* uses a family-centered approach to understand how awareness of genetic risk influences experiences, coping, and adaptation over time for all family members and relationships (Rolland & Williams, 2005) . The time trajectory extends from initial awareness of possible genetic risk, through consideration and decisions about genetic testing, and encompasses the initial and long term adaptation of living with the genetic information and, for some, the development of the condition. Using the *FSGI*, teens under 18 in HD families are in the *Non-symptomatic Awareness phase.* However, since predictive genetic testing for HD is available for persons age 18 or over, teens then transition to the *Crisis Phase I: pre-testing phase*. Teens, aware of the possible genetic risk, “may experience life as the *worried well*, living in fear or with a sense of inevitability that they will eventually develop the disease” (p.11). Yet at the same time, due to the symptom manifestations

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

experienced by the pHD, teens also find themselves in the *FSGI Long-term Adaptation phase*. Teens' lives are complicated by living in the *Non-symptomatic Awareness phase* and preparing to enter the *Crisis I: pre-testing phase*, while, as a part of the family unit, simultaneously experiencing the long-term adaptation to the reality of having a family member with clinical HD. The *FSGI* model provides one mechanism to visualize the complex lives these teens are experiencing.

Limitations

Although this study is among the first to report on teens' experiences, as expressed by teens themselves, the study has some limitations. First, only those teens whose parents gave permission were enrolled in the study. Some parents declined to enroll their teens. They stated their teen would be too reluctant to talk about HD or they feared the experience would be too stressful (Pehler et al., 2008). It is not known what other teens, not identified by or known to HD site coordinators, may have contributed to the discussion.

This study was part of a larger research project that focused on family members' concerns and the strategies used to cope with these concerns. The primary focus on concerns may have led to an over representation of negative experiences and an under representation of positive ones. Further research may identify components of teens' experiences that are beneficial and contribute to positive aspects of their lives and transition into adulthood.

Conclusion

Today, teens in HD families today are facing multiple challenges, witnessing the pHD's ongoing decline, while simultaneously considering the likelihood that they themselves will experience the same disease. Although teens are in the midst of family and friends, their experiences set them apart from those who ordinarily serve as sources of support and understanding. These insights may be of value to health care professionals who have the opportunity to explore ways in which to support these teens and their families.

Acknowledgments

Grant sponsor: The National Institute of Nursing Research; Grant number R01 07970, Janet Williams, Principal Investigator. Participants were recruited through the Neurobiological Predictors of HD grant, funded by the National Institutes of Health R01 NS400 68, Jane S. Paulsen, Principal Investigator. Portions of this content were presented as a poster presentation at the 2006 Midwest Nursing Research Society meeting, (MNRS) in Milwaukee, WI, as a paper presentation at the 2006 Council for the Advancement of Nursing Science (CANS) meeting in Washington, D.C., and as a paper presentation at the 2006 International Society of Nurses in Genetics (ISONG) meeting in New Orleans, LA. We express our sincere appreciation to Anne Letocha Ersig, Rose Marie Friedrich, Carolyn Gray, Bonnie Hennig, Mary Lou Klimek, Hillary Lipe, Elizabeth Penziner, Diane Rehak, and Ann Wellman Schwemm who participated in recruitment, planning, and conduct of focus groups.

References

Bergeron, B. Case studies in genes and disease: A primer for clinicians. American College of Physicians; Philadelphia: 2004.

Binedell J, Soldan JR, Scourfield J, Harper PS. Huntington's disease predictive testing: The case for an assessment approach to requests from adolescents. Journal of Medical Genetics 1996;33(11):912–918. [PubMed: 8950670]

Cauffman E, Steinberg L. (Im)maturity of judgment in adolescence: why adolescents may be less culpable than adults. Behavioral Sciences & the Law 2000;18(6):741–760. [PubMed: 11180420]

Cox SM. Stories in decisions: How at-risk individuals decide to request predictive tesitng for Huntington Disease. Qualitative Sociology 2003;26(2):257–280.

Driessnack, M. Adolescence. In: Edelman, CL.; Mandle, C., editors. Health Promotion Throughout the Lifespan. 6th ed.. Mosby; St. Louis: 2006. p. 502-522.

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

Duncan, R. 'Holding your breath': Predictive genetic testing in young people. The University of Melbourne; 2005. Unpublished doctoral dissertation

Duncan RE, Delatycki MB. Predictive genetic testing in young people for adult-onset conditions: where is the empirical evidence? Clinical Genetics 2006;69(1):8–16. [PubMed: 16451127]

Duncan RE, Gillam L, Savulescu J, Williamson R, Rogers JG, Delatycki M. "You're one of us now": Young people describe their experiences of predictive genetic testing for Huntington disease (HD) and familial adenomatous polyposis(FAP. American Journal of Medical Genetics Part C: Seminars in Medical Genetics. Jan. 11;2008 published online.

Forrest Keenan K, Miedzybrodzka Z, van Teijlingen E, McKee L, Simpson SA. Young people's experiences of growing up in a family affected by Huntington's disease. Clinical Genetics 2007;71:120–129. [PubMed: 17250660]

Fryer A. Inappropriate genetic testing of children. Archives of Disease in Childhood 2000;83(4):283–285. [PubMed: 10999855]

Gaff CL, Lynch E, Spencer L. Predictive testing of eighteen year olds: Counseling challenges. Journal of Genetic Counseling 2006;15(4):245–251. [PubMed: 16865560]

Ginsburg KR. Engaging adolescents and building on their strengths. Adolescent Health Update 2007;19 (2):1–8.

Haigh, B.; Huq, M.; Hayden, M. Huntington Disease Gene Reviews. University of Washington; 2005. Retrieved 10/08, 2005, from the World Wide Web:

Holmbeck GN. A developmental perspective on adolescent health and illness. Journal of Pediatric Psychology 2002;27(5):409–415. [PubMed: 12058005]

Holloway, I.; Wheeler, S. Qualitative research in nursing. 2nd ed.. Blackwell Publishing; Oxford: 2002.

Holt K. What do we tell the children? Contrasting the disclosure choices of two HD families regarding risk status and predictive genetic testing. Journal of Genetic Counseling 2006;15(4):253–265. [PubMed: 16850280]

Knafl KA, Webster DC. Managing and analyzing qualitative data: A description of tasks, techniques, and materials. Western Journal of Nursing Research 1988;10:195–218. [PubMed: 3394320]

Lackey NR, Gates MF. Adults' recollections of their experiences as young caregivers of family members with chronic physical illnesses. Journal of Advanced Nursing 2001;34(3):320–328. [PubMed: 11328437]

Morse, JM.; Field, PA. Qualitative methods for health professionals. 2nd ed.. Sage Publications; Thousand Oaks: 1995.

Morse J. Determining sample size. Qualitative Health Research 2000;10(1):3–5.

Multhaupt-Buell TJ, Lovell A, Mills L, Stanford KE, Hopkin RJ. Genetic service providers' practices and attitudes regarding adolescent genetic testing for carrier status. Genetics in Medicine 2007;9(2): 101–107. [PubMed: 17304051]

National Alliance for Caregiving. Young Caregivers in the US. Bethesda, MD: 2005.

Paulsen J, Hayden M, Stout JC, Langbehn DR, Aylward E, Ross CA, et al. Preparing for preventative clinical trials: The Predict-HD study. Archives of Neurology 2006;63:883–890. [PubMed: 16769871]

Pehler S, Williams JK, Sparbel KJH, Tripp-Reimer T, Schutte DL, Paulsen J, et al. Strategies for teen focus groups on sensitive topics. 2008 Manuscript submitted for publication.

Quaid KA, Sims SL, Swenson MM, Harrison JM, Moskowitz C, Stepanov N, et al. Living at risk: Concealing risk and preserving hope in Huntington disease. Journal of Genetic Counseling. Oct. 18;2007 epub ahead of print.

QSR. NVivo. QSR International; Doncaster, Victoria, Australia: 2000.

Richards FH. Maturity of judgment in decision making for predictive testing for nontreatable adult-onset neurogenetic conditions: A case against predictive testing of minors. Clinical Genetics 2006;70:396–401. [PubMed: 17026621]

Rolland JS, Williams JK. Toward a biopsychosocial model for 21st-century genetics. Family Process 2005;44(1):3–24. [PubMed: 15807075]

Rosenblatt, A.; Ranene, N.; Nance, M.; Paulsen, J. A physician's guide to the management of Huntington's disease. 2nd ed.. Huntington's disease Society of America; New York: 1999.

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

Sandelowski M. Whatever happened to qualitative description? Research in Nursing & Health 2000;23 (4):334–340. [PubMed: 10940958]

Shinaman A, Bain LJ, Shoulson I. Preempting genetic discrimination and assaults on privacy: Report of a symposium. American Journal of Medical Genetics 2003;120A:589–593. [PubMed: 12884445]

Smetana JG, Campione-Barr N, Metzger A. Adolescent development in interpersonal and societal contexts. Annual Review of Psychology 2006;57:255–284.

Sobel SK, Cowen DB. Impact of genetic testing for Huntington disease on the family system. American Journal of Medical Genetics 2000;90:49–59. [PubMed: 10602118]

Taylor SD. Predictive genetic test decisions for Huntington's disease: Context appraisal, and new moral imperatives. Social Science & Medicine 2004;58:137–149. [PubMed: 14572927]

The Huntington's Disease Collaborative Group. A novel gene containing a trinucleotide repeat that is expanded and unstable on the Huntington's disease chromosomes. Cell 1993;72:971–983. [PubMed: 8458085]

Timman R, Roos R, Maat-Kievit A, Tibben A. Adverse effect of predictive testing for Huntington disease underestimated: Long-term effects 7-10 years after the test. Health Psychology 2004;23(2):189–197. [PubMed: 15008664]

Van der Meer L, Timman R, Trijsburg W, Duisterhof M, Erdman R, Van Elderen T, et al. Attachment in families with Huntington's disease: A paradigm in clinical genetics. Patient Education and Counseling 2006;63:246–254. [PubMed: 16427763]

Vamos M, Hambridge J, Edwards M, Conaghan J. The impact of Huntington's disease on family life. Psychosomatics 2007;48:400–404. [PubMed: 17878498]

Warby, SC.; Graham, RK.; Hayden, MR. Huntington disease. Jul 19. 2007 Retrieved November 19, 2007 from GeneReviews at URL http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=pibbn9eKgaEuy&gry=&fcn=y&fw=ojaX&filename=/profiles/huntington/index.html

Williams JK, Ayres L. “I'm like you': Establishing and protecting a common ground in focus groups with Huntington disease caregivers. Journal of Research in Nursing 2007;12(6):655–664.

Williams JK, Hamilton RJ, Nehl C, McGonigal-Kenney M, Schutte DL, Sparbel KJH, et al. “No one else sees the difference”: Family members' descriptions of symptoms in persons with preclinical HD and attempts to manage these symptoms. American Journal of Medical Genetics, Part B 2007;144B(5): 636–641.

Williams JK, Paulsen JS, Schutte DL, Tripp-Reimer T. Family health after predictive HD testing. 2001 Grant NR07970, funded by the National Institutes of Health, National Institute of Nursing Research, from http://crisp.cit.nih.gov.

Williams JK, Schutte DL, Holkup P, Evers C, Muilenburg A. Psychological impact of predictive testing for Huntington disease on support persons. American Journal of Medical Genetics, Part B (Neuropsychiatric Genetics) 2000;96:353–359.

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

Table 1

Characteristics of Teen Participants

Characteristic	Number	Percentage of Total
1. Age (Mean = 15.9)		
14-15	13	40
16-18	19	60
2. Gender		
Female	23	72
Male	9	28
3. HD Risk Status		
Not at Risk	5	15.6
At Risk	27	84.4
4. Living arrangements of pHD		
In Teen's Home	25	78.1
Outside of Teen's Home	7	21.9

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

Table 2

Themes of Teen Experiences

Watching and Waiting

- Watching from the Shadow of HD
  - ○ Observing pHD deterioration while being unable to change disease progression
  - ○ Observing effect of health changes on family o Anticipating the future course of disease in pHD
- Waiting in the Shadow of HD
  - ○ At risk for HD but in pretesting time due to age
  - ○ Reflecting on future decision-making regarding genetic testing
  - ○ Little discussion of future plans beyond genetic testing

Alone in the Midst of Others

- Isolation in the Family
  - ○ Loss of Meaningful Communication with pHD
  - ○ Decreased availability of non-HD parent
- The personal reality of genetic risk
- Separate from Peers
  - ○ Impeded social interaction
  - ○ Lack of peer understanding and support

Family Life is Kind of Hard

- Family Stress
- Conflict in the Home
  - ○ Between parents
  - ○ Among family members
- Financial Strain

Having to be like an Adult

- Caregiving of the pHD
- Caring for the non-HD parent
- Assuming home management responsibilities
- Wanting to have less responsibility

NIH-PA Author Manuscript

NIH-PA Author Manuscript

NIH-PA Author Manuscript

Table 3

Huntington Disease Resources for Teens

The Huntington Society of America
National Youth Alliance (NYA)
http://get-me.to/NYA
The purpose is to provide a support network for youth with Huntington Disease in their lives and bring attention and understanding of Huntington Disease in the community
Gray, A. (2003). HD and me: A guide for young people. http://endoflifecare.tripod.com/livingwith he/id17.html. This 58-page book directed at young people living in a family with HD offers strategies for coping with daily challenges.
Hennig, B (2004) Talking to Kids About Huntington Disease
www.talkingtokidsabouthd.com/book.htm
A 45-page book designed for caregivers of children and teens who have a loved one affected by HD.