Life experiences of individuals with hereditary hemorrhagic telangiectasia and disclosing outside the family: a qualitative analysis

Leigh Ann Higa¹ · Jamie McDonald^{2,3} · Deborah O. Himes³ · Erin Rothwell^{1,4}

Received: 21 May 2015 / Accepted: 27 August 2015 / Published online: 4 September 2015
© Springer-Verlag Berlin Heidelberg 2015

Abstract Hereditary hemorrhagic telangiectasia (HHT; OMIM 187300) is a disorder that affects 1:5000–1:10,000 people worldwide, with an estimated 60,000 affected individuals in the USA. Approximately 50 % of patients with HHT experience potentially life-threatening health complications such as stroke, brain abscess, or heart failure. However, the most common symptom is spontaneous and frequent nosebleeding. HHT is a hereditary condition with significant health consequences, but little is known about how individuals cope with HHT on a daily basis and how individuals share information about the disorder with social groups outside of the family. The objectives of this study were to improve understanding of the daily experiences of patients with diagnosed HHT and to investigate how they disclose their diagnosis to various social groups (friends, dating partners, employers, and coworkers) outside of their biological family. Adult patients seen at a university HHT clinic and who had been diagnosed with HHT for at least 6 months were recruited by mail. Participants completed semi-structured telephone interviews ($n=19$). A qualitative content analysis of interview transcripts identified four major categories: (1) the emotional impact of HHT, (2) the social impact of HHT, (3) concerns for

current and future health related to HHT, and (4) social context drives disclosure of HHT. Participants reported that although HHT was a manageable hereditary disorder, the symptoms negatively affected their daily life. It is important for health care providers to understand how individuals with rare genetic disorders are managing.

Keywords Hereditary hemorrhagic telangiectasia · Self-disclosure · Coping · Social groups

Introduction

Hereditary hemorrhagic telangiectasia (HHT; OMIM 187300) is an autosomal dominant genetic disorder found in all ethnic groups that affects 1:5000–1:10,000 people worldwide, with an estimated 60,000 affected individuals in the USA (McDonald et al. 2011). HHT is more common than many more widely recognized disorders such as hemophilia A (with an incidence of 1:10,000). HHT is a multisystem disorder that results from arteriovenous malformations (AVMs), the abnormal direct connection of arteries to veins (McDonald et al. 2011). Normally, arteries and veins are separated by networks of smaller blood vessels (capillaries). AVMs, or telangiectases as typically called when they are small, pose significant risks for hemorrhage as well as shunting of blood through these abnormally dilated vessels. Multiple genetic subtypes of HHT have been described, but 96 % of individuals who meet consensus diagnostic criteria for HHT (Shovlin et al. 2000) have a mutation in either the endoglin or ACVRL1 gene (HHT type 1 or HHT type 2). These two common types of HHT are indistinguishable clinically and put patients at risk for similar symptoms and complications. The penetrance of symptoms approaches 100 % by age 40 in those affected; the average age at first symptom is 12 years but ranges from

✉ Erin Rothwell
erin.rothwell@nurs.utah.edu

¹ Graduate Program in Genetic Counseling, University of Utah, 10 South 2000 East, Salt Lake City, UT 84112, USA

² Department of Radiology, University of Utah, Salt Lake City, UT, USA

³ College of Nursing, Brigham Young University, Provo, UT, USA

⁴ College of Nursing, University of Utah, Salt Lake City, UT, USA

infancy to adulthood. The type and severity of manifestations are very variable, and the intra-familial variability is as noteworthy as inter-familial variability (McDonald et al. 2011).

Symptoms of HHT include recurrent nosebleeds (epistaxis), which can be severe enough to cause chronic anemia and require repeated medical attention. Telangiectases appear as small pink-red spots and occur in the nasal, oral, and intestinal mucosa, and skin of the hands and face. Approximately 50 % of patients with HHT experience disabling or life-threatening health complications such as stroke, brain abscess, or heart failure, from cerebral, pulmonary, and hepatic AVMs, respectively. Proper surveillance and medical management are effective in preventing most severe complications of HHT.

Despite HHT as a hereditary condition with significant health consequences, only a few studies have been published about how individuals cope with HHT on a daily basis. Some research assessed health-related quality of life (QoL) quantitatively among individuals with HHT with surveys such as the 36-Item Short Form Health Survey (SF-36) (Geirdal et al. 2012; Geisthoff et al. 2007). Compared to the general population in various countries, patients with HHT scored significantly lower on all domains of the SF-36 except for bodily pain (Geisthoff et al. 2007; Lennox et al. 2005; Pasculli et al. 2004; Geirdal et al. 2012). Within the HHT patient population, patients with severe epistaxis scored significantly lower than patients with mild epistaxis on all physical and mental dimensions of the SF-36 (Merlo et al. 2014). Increasing age and increasing number of years living with HHT are two other factors that have been shown to correlate with lower scores on one or more SF-36 scales for physical health and the mental health component summary (Geisthoff et al. 2007; Pasculli et al. 2004). Using a symptom-specific HHT questionnaire developed by Slotosch et al. (2006), 58 % of patients with HHT reported that the condition impacted their QoL (Geirdal et al. 2012). Participants reported that recurrent nosebleeds negatively affected their work and other social activities, and pain, fatigue, and time spent on nose care corresponded with decreased QoL (Geirdal et al. 2012). In response to an open-ended query, nosebleeds were reported by patients with HHT as the major reason for restricted participation in athletics, change in occupation, and psychological strain (Geisthoff et al. 2007).

One poorly understood aspect of living with a hereditary condition such as HHT is how individuals share information with social groups outside of the family. Communication within families is well studied, in part because of the emphasis on gathering and analyzing family medical history (Forrest et al. 2003; Gaff et al. 2007; Sobel and Brookes Cowan 2000). Understanding the patterns and significance of sharing a diagnosis outside of families can offer insight into the challenges of living with a hereditary condition. For example, sharing a diagnosis has a social function and can establish, strengthen, or dissolve relationships (Greene et al. 2006).

Sharing can also improve self-esteem and identity with the discloser (Greene et al. 2006).

Previous work explored the relationship between stigma and decisions to disclose a chronic health condition. Stigma involves an individual's social identity perceived as undesirable and different, a state that Goffman (1963) referred to as “discredited” (Ablon 2002). In chronic illness, the body's physical appearance can discredit an individual, while a “healthy” appearance can allow an individual with chronic illness to conceal their condition (Christian and D'Auria 1997; Joachim and Acorn 2000; Lowton 2004; Admi 1995). Individuals with cystic fibrosis (CF) reported hiding their condition during childhood in order to fit in socially (Christian and D'Auria 1997), while adults with severe signs and symptoms of CF were more likely to share their diagnosis than mildly affected adults who appeared “healthy” (Lowton 2004; Modi et al. 2010). The influence of physical appearance associated with illness has also been observed with conditions such as epilepsy and AIDS (Scambler and Hopkins 1986; Mansergh et al. 1995).

Research on how individuals self-disclose with regard to a hereditary disorder has been examined in several conditions including CF, hereditary breast and ovarian cancer (HBOC), Huntington disease (HD), and alpha 1 antitrypsin deficiency (AAT) (Lowton 2004; Modi et al. 2010; Klitzman and Sweeney 2011; Klitzman 2012; Hoskins et al. 2008; Keenan et al. 2013). A unique set of medical concerns, natural history, and treatment and management options for each hereditary condition may impact how individuals experience their condition on a daily basis, and this may alter an individual's motivation and need to share their diagnosis outside of family. Therefore, exploring how individuals experience their condition on a daily basis and how they share their diagnosis with various social groups is important.

In sum, previous studies identified medical, psychological, and social factors that influenced disclosure. Medical factors included disease type, change in health status over time, and the possibility of concealing the condition as another less stigmatizing condition (Klitzman 2012; Klitzman and Sweeney 2011; Lowton 2004). Psychological factors encompassed negative outcomes such as fear of rejection or positive outcomes such as acquisition of emotional support (Lowton 2004; Klitzman and Sweeney 2011; Klitzman 2012). Regarding social relationships, the duration and quality of the relationship determined whom individuals told their condition about. Overall, most participants shared their diagnosis with those they considered emotionally close, such as close friends (Scambler and Hopkins 1986; Modi et al. 2010; Lowton 2004; Klitzman and Sweeney 2011; Klitzman 2012).

The objectives of this study were to improve understanding of the daily experiences of patients with diagnosed HHT and to investigate how they disclose their diagnosis to various social groups outside of their biological family. The findings

of this study may increase our knowledge of a rare genetic disorder with limited public recognition and distinct morbidity and mortality.

Materials and methods

Patient population

Institutional review board approval from the University of Utah was obtained prior to study initiation. The University of Utah HHT Clinic database was reviewed to identify 30–40 patients who met the following criteria: (1) were at least 18 years of age, (2) had a diagnosis of HHT confirmed according to consensus criteria, and (3) been diagnosed for at least 6 months. The minimum of 6 months from diagnosis was chosen to allow for a period of time in a patient's life since the diagnosis had been made. Eligible participants were selected from the patients seen most recently in the HHT Clinic and contacted in reverse chronological order.

Participant recruitment

Eligible participants were contacted by mail. The recruitment packet contained the following: (1) a recruitment letter describing the objectives and format of the study, (2) \$2 token incentive, (3) two copies of the consent form and one self-addressed stamped envelope for return of one signed copy, and (4) an opt out postcard.

A total of 40 eligible participants were contacted. Of the packets mailed, 4 returned the opt-out postcard and 23 returned the signed informed consent form (response rate for opt-in was 58 %). Three participants consented but were unreachable by telephone for the interview, and one participant was not contacted because data collection was completed before receipt of the consent form. Informed consent was obtained from all individual participants included in the study. When participants returned the signed consent form, participants provided their phone number, age, age at diagnosis, ethnicity, and marital status. Participants completed a 30–45-min phone interview.

Data collection

A qualitative descriptive approach was deliberately chosen as the best tool to capture the experiences, thoughts, and emotions of participants living with HHT and sharing their diagnosis. Paired with qualitative content analysis, qualitative description provides meaningful answers to research questions by focusing on the facts presented and the use of participants' own words, without requiring reliance on pre-existing frameworks and theories to formulate an answer (Sandelowski 2000, 2010).

Semi structured telephone interviews were conducted based on questions used by Klitzman and colleagues (Klitzman 2012; Klitzman and Sweeney 2011). The interview guide was piloted over the telephone by the first author with another author (J. McDonald) and a genetic counseling student prior to participant interviews to improve question clarity. Interview questions were open ended and focused on the participant's experiences with HHT and the process of sharing their diagnosis with four different social groups (friends, dating partners, employers, and coworkers). These four groups were chosen because they were the groups most extensively discussed in previous publications (Klitzman 2012; Klitzman and Sweeney 2011). Three questions are provided below to illustrate the types of questions asked:

1. What are your understandings of what it means to have HHT?
 - (a) For your physical health?
 - (b) For your emotional health?
2. Has HHT affected you in any way, and if so, how?
3. How did you make these decisions (to share a diagnosis)?

The interview guide is available upon request.

Data analysis

The interviews were audio recorded, transcribed, and verified for accuracy. A qualitative content analysis was conducted on the transcripts. The initial two transcripts were read through once carefully to identify codes that emerged from the data and the interview questions (Rothwell et al. 2012). Codes (sections of text expressing a similar concept) were generated from the topics of the interview guide and the transcripts. Next, codes were applied to transcripts, and additional codes were added. Throughout the data analysis process, team meetings were conducted to review the coding and assure accuracy of the coding template. Data saturation was reached by the 11th interview with no new codes emerging from the analysis. However, data analysis included all of the 19 interviews. Once coding was finished, codes were grouped into larger categories based on similarity of the data.

Results

Participant demographics

Participant demographic information is presented in Table 1. Four categories emerged during the analyses. These included the emotional impact of HHT, the social impact of living with

Table 1 Demographic information for participants

	N=19 (%)
Gender	
Women	12 (63.1 %)
Men	7 (36.8 %)
Age (years)	
Range	19–82
Mean	48.5
Median	47
Age at diagnosis	
Range	5–70 years
0–18 years	7 (36.8 %)
≥18 years	12 (63.1 %)
HHT type	
HHT type 1	5 (26.3 %)
HHT type 2	14 (73.7 %)
Marital status	
Single	2 (10.5 %)
Married	13 (68.4 %)
Divorced/widowed	4 (21.1 %)
Ethnicity	
White	100 %

HHT, concerns for current and future health implications of HHT, and the social context of symptoms drives disclosure.

Emotional impact of HHT

When participants were asked how HHT affected them, most responses focused on the emotional impact of HHT. The most common experiences reported were annoyance, frustration, and embarrassment. These emotional experiences were attributed to frequent and unexpected nosebleeds, the most common symptom of HHT. Besides the challenge of unexpected onset, nosebleeds were also unpredictable in nature: bleeding could be mild or severe, vary in duration, and easy or difficult to stop. Depending on these factors, participants reported that a nosebleed had the potential to disrupt whatever activity they were engaged in at the time. Participants felt having a nosebleed and the reaction of others who did not know about their HHT caused embarrassment. Positive emotions reported included acceptance and adjustment to HHT as part of daily life. One participant stated “I’m not ashamed or embarrassed about it. And I’m not freaked out by it, either. It’s just a part of me and a part of my daily life.” (female, participant 5, age 21). Some representative quotes included

Emotionally, this actually takes a pretty heavy toll. It’s very, very frustrating, especially when you’re trying to work or doing things, playing with your kids, or things

like that, to have a nosebleed or to have to stop because you’ve had so many that week.

Male, participant 9, age 27

I was on the golf course one day addressing a ball and right there in front of everybody, I mean literally the blood just starting dripping out of my nose and everybody thought I was going to die or that I needed to go the hospital so at times it can be embarrassing.

Male, participant 16, age 62

Social impact of living with HHT

Participants reported that living with HHT often restricted and limited engagement in various social activities. Most of these experiences were described through examples in leisure and work contexts. For example, one participant stated “sometimes for no reason, which is the usual MO [modus operandi] of HHT, you’ll find yourself bleeding. You don’t want to go out in public.” (male, participant 16, age 62). Although many expressed that HHT did not hold them back, most of the participants stated that there was always a risk for nosebleeds, and this risk was taken into consideration before deciding to take part in an activity. One participant stated

Sometimes it can be very depressing. It interferes with your, I don’t want to say my public life, but being out with people sometimes is very limiting. I’ve left a number of friends in restaurants to pay the bill while I had to head back home.

Male, participant 16, age 62

Another participant stated

we get nosebleeds sometimes every day a couple of times a day and that does affect things we go do with people sometimes because you don’t want to get a nosebleed. Other than that, you always want to sit on an outside edge to get out of the room fast if you need to but other than that it’s living life.

Female, participant 3, age 25

The other common context where nosebleeds were a significant problem was work settings. Several participants reported that nosebleeds and other health complications impacted their ability to work. One participant stated “I’m a hairstylist, and a lot of times I have to stop what I’m doing and get it to stop bleeding because I refuse to work with kleenex in my nose.” (female, participant 6, age 36). Another stated “I used to teach, and that was a real problem because I would have all these kids, so it is impossible leave the room just on the spur of the moment.” (female, participant 17, age 50). Finally, one

participant stated she chose a profession that allowed her to work at home because of her frequent nosebleeds.

Concerns for current and future health implications of having HHT

Participants expressed concerns regarding their own current health and the health of affected family members. For themselves, participants reported varied impact of HHT on physical health, depending on their symptoms. At one end of the spectrum, participants reported minimal impact from nosebleeds, as expressed by one participant who said “No, it’s nothing big. To me, I get bloody noses every day. It’s just part of my daily routine.” (female, participant 5, age 21). However, the most common concern voiced by participants was blood loss and resultant anemia. For example, one participant said “if I bleed too much, then the whole iron issue comes in. You know, the being lethargic and having low iron.” (female, participant 6, age 36). This concern was also stated in terms of taking health-related precautions. One participant stated “I have to be a lot more careful about making sure to keep my body healthy so I don’t wear my heart out, bleed to death.” (male, participant 9, age 27).

Another participant stated concerns about undetectable bleeding:

As far as physical health, I just have that little kind of thing of what if it happened in my digestive tract, the swallowing thing and you were bleeding quite a bit and not aware of it. When it’s in your head, when it’s in your nose, you know it’s there and you know how much blood you’re losing and that but I wonder just a little about the other thing.

Male, participant 12, age 79

Several participants reported that they worried about the health of family members with HHT. One participant stated

I’m surrounded by family members who struggle with needing blood transfusions. My own dad has to have blood transfusions on a regular basis because he has issues with kidneys and gastrointestinal and things because of HHT.

Female, participant 18, age 43

Participants reported witnessing the progressive nature of the disease in older relatives. Many referenced other family members with HHT who struggled as they aged: “There’s an emotional toll that goes with seeing where it could go.” (female, participant 18, age 43). Another quote included

As I get older and watch my mom with struggle with it, it’s really sad to know that that’s probably going to be

what kills her one day so it’s just kind of hard to see but at the same time nothing is guaranteed.

Female, participant 3, age 25

Experiences with family members who have HHT influenced attitudes regarding their own future health. Participants expressed worry and uncertainty regarding the future because of the possibility of experiencing additional or more severe symptoms of HHT as they aged: “I worry about the future and what it’s going to do to my later years.” (female, participant 18, age 43). A participant who accompanies her affected mother to her doctor’s appointments observed “I can kind of see where my situation might go also in the next 24 years.” (female, participant 7, age 60). One representative quote that captured this concern included

I’m at risk for things that can happen, I can have more pulmonary AVMs, I could have GI bleeds, I could have liver problems, I could have heart trouble as I get older. It’s been a little scary the last few years.

Female, participant 7, age 60

Social context drives disclosure of HHT

Participants described two factors that contributed to sharing a diagnosis of HHT: experiencing HHT symptoms such as a nosebleed and the nature and importance of their relationship with the recipient. Neither factor alone, but their interplay, prompted sharing a HHT diagnosis on a need-to-know basis. For example, participants who experienced a nosebleed in a public setting such as a restaurant would tell their friends who were present that they had HHT, but telling others present was not necessary and unlikely to occur. Representative quotes included

I think it’s usually if I’ve had a nose bleed. I don’t just bring it in to conversation willy-nilly but if I should get a little nose bleed

Male, participant 12, age 79

if there are people that I’m meeting intermittently or I don’t have a bleeding problem around or it’s just a short-term duration of whatever, unless it is impacting them I don’t feel a need to say I’ve got this and I’m going to have a problem. So there are some people that I just choose not to bring it to the forefront and tell them unless it’s necessary

Male, participant 9, age 27

Participants disclosed HHT to others with whom they had emotionally close relationships, such as friends and serious dating partners. Friends were likely to observe a nosebleed

because of the amount of time spent together. One participant stated

my friends used to freak out a lot when I got bloody noses every day. And they started to get a little concerned. So I had to explain to them that I have HHT, and it's just part of the symptoms that I get bloody noses.
Female, participant 5, age 21

Participants fell into two major categories for dating relationships. Some participants were diagnosed in adulthood, after they had established committed romantic relationships. These participants often reported that partners were involved and informed about HHT at the same time as them so disclosure was not necessary. Other participants knew they had HHT while dating. Participants chose to tell once they felt the relationship had the potential to become meaningful. One participant stated “I have told my dating partners about it. Obviously not right away, because it's not a big deal to me. But if it [the relationship] was serious, then yes.” (female, participant 5, age 21).

Unlike friends and committed romantic partners, participants had reasons for not telling employers and coworkers about HHT. Motivations for not telling their employers about HHT included no need to know and the desire to avoid any negative connotations regarding work competency and professionalism. As one participant remarked, his goal is “to keep it professional.” (male, participant 9, age 27). Another participant noted that “Not very many corporate environments are very supportive of people who can't just be slaves to their job.” (female, participant 18, age 43). The most common motivation for sharing was explaining the reason for possible disruption to work activities, since nosebleeds could require a participant to step away from their duties suddenly. One quote that represented this situation included

my immediate boss knows so if I have to step out and I am in the restroom and it takes me a while to come back out or if I have to leave a meeting, he knows the reason, just to kind of make him aware of it.
Female, participant 15, age 47

Of note, participants reported that coworkers who became friends were told about HHT. One participant stated about why she disclosed HHT to some but not all coworkers: “It just depended on how close I was to them.” (female, participant 1, age 75).

Discussion

We identified four categories that describe the daily experience of adults living with HHT, including the experience of

sharing their diagnosis outside of their family. The first category, the emotional impact of HHT, encompassed both adjustment and acceptance of HHT as well as annoyance, frustration, and embarrassment about experiencing nosebleeds. These negative feelings towards nosebleeds confirm previous patient reports of nosebleeds as the primary cause of psychological strain (Geisthoff et al. 2007). The coexistence of acceptance and adjustment contrasts with the negative impact of HHT on professional and social life. However, this finding is consistent with previous studies of patients with HHT in Norway and Germany. First, 58 % reported that HHT symptoms negatively impacted their QoL (Geirdal et al. 2012). Second, this study confirmed work as the primary area of everyday life affected by HHT (Geirdal et al. 2012). A subset of German patients with HHT reported that experiencing nosebleeds led to a career change (Geisthoff et al. 2007). Third, Geirdal et al. (2013) identified acceptance as a coping mechanism in patients with HHT.

For social activities, the disruptive nature of nosebleeds reported here fits with the correlation between lower SF-36 mental and physical component scores and a decreased number of social contacts due to HHT. Together, these data emphasize the importance of regular assessment of a patient's emotional health and coping strategies in concert with physical health. Patients who are experiencing restrictions because of HHT on their professional and social lives may benefit from discussion of additional support resources as well as medical interventions. Given the variability in personal medical history, clinical findings, family history, social factors, and coping styles, discussion and selection of resources and medical interventions should be tailored to best meet each patient's needs. For example, a patient whose nosebleeds have increased in severity may benefit from discussion of coagulation therapies, while a patient who is dealing with a new and unexpected diagnosis may benefit from being connected with a local support group.

Concerns about current and future health emerged as a third category and illustrated how participants with HHT perceived their health similar to the health of affected family members. Previously, Geisthoff et al. (2007) demonstrated a correlation between increased “worries about having HHT” with lower SF-36 physical and mental component scores. We identified the experience of affected family members as an influence on how participants with HHT anticipated future changes in their health. Participants expressed uncertainty and worry about the future physical impact of HHT and resultant lifestyle changes. These data highlight that eliciting a patient's experience of HHT in their family may offer a valuable perspective for health care providers. A patient who is the first person to be diagnosed with HHT in their family may have a distinct set of concerns compared to a patient who has multiple immediate family members also affected with HHT. This finding is consistent with the identification of family history of a genetic

condition as a key component of an individual's subjective perception of genetic risk and illness, and subsequent health behaviors. A family's experience with illness is a dynamic construct that may incorporate personal attitudes, beliefs, and meanings of illness over time. For instance, 25 % of the 22 female participants at risk for HBOC reported that physical or personality similarities with affected relatives influenced their cancer susceptibility (Werner-Lin 2007). Bakos et al. (2008) found that women from HBOC families with multiple affected family members continued to perceive their breast cancer risk as elevated above the general population, even after receiving a negative genetic test for their familial mutation. Heightened risk perception can also alter health behaviors: Babb et al. (2002) identified an association between personal and/or family history of cancer and uptake of prophylactic oophorectomy instead of ovarian cancer screening in women at risk for HBOC.

Taking a step back, the almost universal experience of nosebleeds reported by our participants informs both the emotional impact and current and future health concerns related to HHT. One plausible reason for this is the fact that about 95 % of individuals affected with HHT experience nosebleeds over their lifetime, and nosebleeds (along with telangiectases) are often the presenting symptom (McDonald et al. 2011). With an average age of onset of epistaxis of 12 years (McDonald et al. 2011), it is likely that nosebleeds are the HHT symptom that participants have coped with for the longest amount of time. However, it is worth noting that multiple participants discussed additional severe symptoms such as liver, spine, and brain AVMs and gastrointestinal bleeding (unpublished data). Since the goal of qualitative content analysis is to identify common experiences across a participant sample, nosebleeds instead of other severe complications emerged as having a primary impact on the daily life of patients with HHT. Bernhardt et al. (2011) found that individuals with HHT erroneously perceived epistaxis as the key diagnostic feature of HHT, leading to misunderstanding about which family members were at risk of HHT. This finding suggested that individuals with HHT may have limited knowledge of additional severe complications. In our study, multiple participants described that their understanding of the health implications of HHT increased with time (unpublished data). While we cannot conclusively eliminate the possibility of knowledge bias towards nosebleeds as a factor in our data, the natural history of HHT supports the reported impact of epistaxis.

The final category, social context drives disclosure of HHT, identified two factors that influence participants to share their diagnosis. The majority of participants reported that sharing their diagnosis occurred with others who were important to them (friends and dating partners), most often after having a nosebleed in their presence. The motivation to share often stemmed from the feeling that friends and committed partners deserved to know because of the nature of the

relationship, as well as a desire to reduce any concern or worry they may feel.

Study findings were similar to the themes identified for disclosing genetic information in dating relationships proposed by Klitzman and Sweeney (2011) and others (Lowton 2004). Participants with HHT reported taking into account the fact that HHT symptoms were present (represented twice under the “factors” and “disclosure decisions” themes) similar to those experiencing visible symptoms of Huntington disease or CF (Klitzman and Sweeney 2011; Lowton 2004). A second consideration described by many participants with HHT was the nature and degree of relationship with a person before sharing a diagnosis; this finding parallels “length of relationship” and “emotional closeness” identified within the “factors” theme in Klitzman and Sweeney's study (2011). Finally, participants with HHT chose to tell when they felt the relationship had become “serious,” which had been identified as part of the “disclosure decisions” theme. The overlap with the “factors” and “disclosure decision” themes highlights potential core elements of the decision to share a diagnosis of any chronic or hereditary condition, as previously explored for CF and epilepsy (Lowton 2004; Scambler and Hopkins 1986).

Participants with HHT had comments about their motivation for sharing and not sharing a diagnosis similar to participants in previous studies (Klitzman 2012; Klitzman and Sweeney 2011). Many participants with HHT voiced that HHT was not something to hide or be embarrassed about, and telling others was an expression of their self-identity. However, the desire to avoid negative consequences in the workplace led participants with HHT to avoid sharing their diagnosis with coworkers and employers, despite the fact that regular, long-term social interaction may characterize these workplace relationships. As Klitzman (2012) aptly put it, coworkers occupy an unusual social space as “work friends.” If sharing a HHT diagnosis was relevant to a particular social exchange with coworkers or employers, participants were more likely to share their diagnosis.

Together, the disclosure behavior of participants with HHT towards dating partners, employers, and coworkers is reminiscent of Lowton's classification of these relationships as “high-risk” for individuals with CF (Lowton 2004). The risk level is based on relationship qualities such as duration and interaction extent, resulting in an increased chance of discovery despite efforts at disorder concealment (Lowton 2004).

Differences were identified between the HHT patient population, in comparison to the participants interviewed by Klitzman and Sweeney (Klitzman 2012; Klitzman and Sweeney 2011). The most striking difference was the ease with which participants with HHT appeared to engage in disclosing HHT. When participants with HHT were queried about how they made the decision to tell someone, participants often described *why* they decided to disclose instead of *how* they reached their decision. Participants may not have

made conscious decisions about sharing information. This finding differs from the process described by the group interviewed by Klitzman and Sweeney (Klitzman 2012; Klitzman and Sweeney 2011) where the decision to share is characterized as a “series of dilemmas” or “quandaries” faced by individuals who are “confronting” hereditary disease (Klitzman and Sweeney 2011). One possible explanation for this difference is the visibility and frequency of nosebleeds in HHT, compared to other conditions. While certainly there can be physical manifestations resulting from other hereditary diseases, such as mastectomy and infusion port placement for AAT enzyme replacement, it may be argued that public perception is qualitatively different for a severe nosebleed. A few participants described perceived responses of others who are unaware about HHT to blood and its associations with transmissible infections such as HIV/AIDS and Ebola (unpublished data).

In summary, this study explored the experiences of adult participants living with HHT and how they shared their diagnosis with friends, dating partners, employers, and coworkers. Participants reported that experiencing HHT symptoms, primarily recurrent nosebleeds, impact their emotional health, social lives, and perceptions of their current and future health. Participants reported that both HHT symptoms and the nature of social relationships contributed to the timing and appropriateness of disclosing HHT to different social groups. These data suggest that sharing a HHT diagnosis occurs most often with friends and committed romantic partners because of emotional closeness. Relationships with employers and coworkers are less likely to be emotionally close, and disclosing may but does not always occur. These social boundaries may be fluid, as several participants spoke of sharing a diagnosis with a coworker once they became a friend. Ultimately, these data suggest that inquiring specifically about the impact of HHT on these four categories may enhance psychosocial assessment of patients with HHT by health care providers.

Study limitations

This study was conducted with a small sample size ($n=19$), and all participants were patients who had been seen at a HHT center of excellence. Therefore, this patient population may not be representative of the larger population of patients with HHT.

Acknowledgments The authors would like to thank the study participants, the University of Utah HHT Clinic, and the University of Utah Graduate Program in Genetic Counseling for their support; Kali Roy for piloting the interview guide; and Connie Larrinaga-Pope for assistance with disbursement of study funds.

Compliance with ethics guidelines 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.

Conflict of interest The authors declare that they have no competing interests.

References

- Ablon J (2002) The nature of stigma and medical conditions. *Epilepsy Behav* S2–S9
- Admi H (1995) “Nothing to hide and nothing to advertise”: managing disease-related information. *West J Nurs Res* 17(5):484–501
- Babb SA, Swisher EM, Heller HN, Whelan AJ, Mutch DG, Herzog TG and Rader, JS (2002) Qualitative evaluation of medical information processing needs of 60 women choosing ovarian cancer surveillance or prophylactic oophorectomy. *J Gen Counsel* 81–96
- Bakos AD, Hutson SP, Loud JT, Peters JA, Giusti RM, Greene MH (2008) BRCA mutation-negative women from hereditary breast and ovarian cancer families: a qualitative study of the BRCA-negative experience. *Health Expect* 11(3):220–231
- Bernhardt BA, Zayac C, Pyeritz RE (2011) Why is genetic screening for autosomal dominant disorders underused in families? The case of hereditary hemorrhagic telangiectasia. *Genet Med* 13(9):812–820
- Christian BJ, D’Auria JP (1997) The child’s eye: memories of growing up with cystic fibrosis. *J Pediatr Nurs* 12(1):3–12
- Forrest K, Simpson SA, Wilson BJ, van Teijlingen ER, McKee L, Haites N, Matthews E (2003) To tell or not to tell: barriers and facilitators in communication of genetic risk. *Clin Genet* 64(4):317–326
- Gaff CL, Clarke AJ, Atkinson P, Sivell S, Elywn G, Iredale R, Thornton H, Dundon J, Shaw C, Edwards A (2007) Process and outcome in communication of genetic information within families: a systematic review. *Eur J Human Genet* 999–1011
- Geirdal AO, Dheyauldeen S, Bachmann-Harildstad G, Heimdal K (2012) Quality of life in patients with hereditary hemorrhagic telangiectasia in Norway: a population based study. *Am J Med Genet A* 1269–1278. doi:10.1002/ajmg.a.35309
- Geirdal AO, Dheyauldeen S, Bachmann-Harildstad G and Heimdal K (2013) Living with hereditary haemorrhagic telangiectasia: coping and psychological distress—a cross-sectional study. *Disabil Rehabil* 206–213. doi: 10.3109/09638288.2012.690500
- Geisthoff UW, Heckmann K, D’Amelio R, Grünewald S, Knöbber D, Falkai P, König J (2007) Health-related quality of life in hereditary hemorrhagic telangiectasia. *Otolaryngol Head Neck Surg* 136:726–733
- Goffman E (1963) Stigma: notes on the management of spoiled identity. Englewood Cliffs, Prentice Hall
- Greene K, Derlega VJ, Mathews A (2006) Self-disclosure in personal relationships. In: Vangelisti AL, Perlman D (eds) *The Cambridge handbook of personal relationships*. Cambridge University Press, New York, pp 409–427
- Hoskins LM, Roy K, Peters JA, Loud JT and Greene MH (2008) Disclosure of positive -mutation status in young couples: the journey from uncertainty to bonding through partner support. *Fam Syst Health* 296–316. doi:10.1037/a0012914
- Joachim G, Acorn S (2000) Stigma of visible and invisible chronic conditions. *J Adv Nurs* 32(1):243–248
- Keenan KF, Simpson SA, Miedzybrodzka Z, Alexander DA, Semper J (2013) How do partners find out about the risk of Huntington’s disease in couple relationships? *J Genet Couns* 22(3):336–344
- Klitzman RL (2012) *Am I my genes? Confronting fate and family secrets in the age of genetic testing*. Oxford University Press, USA, Oxford

- Klitzman RL, Sweeney MM (2011) In sickness and in health? Disclosures of genetic risks in dating. *J Genet Couns* 20(1):98–112. doi:[10.1007/s10897-010-9331-z](https://doi.org/10.1007/s10897-010-9331-z)
- Lennox PA, Hitchings AE, Lund VJ, Howard DJ (2005) The SF-36 health status questionnaire in assessing patients with epistaxis secondary to hereditary hemorrhagic telangiectasia. *Am J Rhinol* 19(1):71–74
- Lowton K (2004) Only when I cough? Adults' disclosure of cystic fibrosis. *Qual Health Res* 14(2):167–186
- Mansergh G, Marks G, Simoni JM (1995) Self-disclosure of HIV infection among men who vary in time since seropositive diagnosis and symptomatic status. *AIDS* 9:639–644
- McDonald J, Bayrak-Toydemir P, Pyeritz RE (2011) Hereditary hemorrhagic telangiectasia: an overview of diagnosis, management, and pathogenesis. *Genet Med Off J Am Coll Med Genet* 13:607–616. doi:[10.1097/GIM.0b013e3182136d32](https://doi.org/10.1097/GIM.0b013e3182136d32)
- Merlo CA, Yin LX, Hoag JB, Mitchell SE, Reh DD (2014) The effects of epistaxis on health-related quality of life in patients with hereditary hemorrhagic telangiectasia. *Int Forum Allergy Rhinol* 4(11):921–925. doi:[10.1002/alr.21374](https://doi.org/10.1002/alr.21374)
- Modi AC, Quittner AL and Boyle MP (2010) Assessing disease disclosure in adults with cystic fibrosis: the Adult Data for Understanding Lifestyle and Transitions (ADULT) survey disclosure of disease in adults with cystic fibrosis. *BMC Pulm Med* 46–54
- Pasculli G, Resta F, Guastamacchia E, Di Gennaro L, Suppressa P, Sabbà C (2004) Health-related quality of life in a rare disease: hereditary hemorrhagic telangiectasia (HHT) or Rendu-Osler-Weber disease. *Qual Life Res Int J Qual Life Asp Treat Care Rehab* 13(10):1715–1723. doi:[10.1007/s11136-004-7865-y](https://doi.org/10.1007/s11136-004-7865-y)
- Rothwell E, Ellington L, Planalp S, Crouch B (2012) Exploring challenges to telehealth communication by specialists in poison information. *Qual Health Res* 12(1):67–75. doi:[10.1177/1049732311420446](https://doi.org/10.1177/1049732311420446)
- Sandelowski M (2000) Whatever happened to qualitative description? *Res Nurs Health* 23:334–340
- Sandelowski M (2010) What's in a name? Qualitative description revisited. *Res Nurs Health* 33:77–84
- Scambler G, Hopkins A (1986) Being epileptic: coming to terms with stigma. *Sociol Health Illn* 8:26–43
- Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJ, Kjeldsen AD, Plauchu H (2000) Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). *Am J Med Genet* 91:66–67
- Slotosch D, Koller M, Werner JA, Folz BJ (2006) Recurrent nosebleeds in patients with hereditary hemorrhagic telangiectasia. *Dtsch Med Wochenschr* 131(11):535–539. doi:[10.1055/s-2006-933691](https://doi.org/10.1055/s-2006-933691)
- Sobel S, Brookes Cowan D (2000) Impact of genetic testing for Huntington disease on the family system. *Am J Med Genet* 90(1):49–59
- Werner-Lin AV (2007) Danger zones: risk perceptions of young women from families with hereditary breast and ovarian cancer. *Fam Process* 46(3):335–349