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ORIGINAL ARTICLE

Role of older generations in the family's adjustment to Huntington disease

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Abstract

Genetic diseases are a family matter, requiring adjustment and management from the family system, particularly when the diagnosis is recent. Literature has evidenced the importance of the role of older relatives in families dealing with some genetic diseases; however, knowledge is scarce regarding rare incurable genetic disorders, such as Huntington disease. Therefore, this exploratory qualitative study aims at describing how adjustment to Huntington disease occurs, from a family perspective, considering the roles performed by older generations, in the Portuguese context. It adopts the critical incidents technique, administered based on semi-structured interviews, and comprises 10 participants, aged 28 to 72 years (8 females), from seven families. Participants reported 130 critical incidents. The interviews were audiotaped, transcribed, and submitted to thematic analysis. Findings portray participants and their families as "beginners" in understanding and incorporating Huntington disease in their lives, due to recent diagnosis. In addition, data suggest that older relatives play two relevant roles in the creation of family narratives: (1) "shaping awareness about HD" (68 critical incidents) and (2) "influencing HD management" (62 critical incidents). Genetic counseling and family-centered interventions aimed at supporting families with a history of hereditary genetic diseases, should consider a narrative approach involving older relatives, since they have a great influence in sustaining family stories.

Keywords Genetic disease · Genetic counseling · Family life cycle · Family counseling

Introduction

Illnesses may be considered a family matter, what gains a definite meaning with hereditary diseases. In fact, genetic information differs from other medical information because of its familial character. The diagnosis of a genetic disease or a pre-symptomatic test result may mean that relatives (including future offspring) are at-risk of developing the same disease. It may also mean that the disease existed in the family beforehand, although sometimes undiagnosed. Family adjustment to genetic diseases encompasses a transgenerational process of integrating the illness experience with genetic knowledge, which influences how the new information will be lived by the family (McDaniel 2005; Rolland and Williams 2005; Street and Soldan 1998; Werner-Lin and Gardner 2009).

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Huntington disease (HD) is a rare incurable, autosomal-dominant neurological disease (Bates et al. 2015). It is ultimately fatal and combines progressive motor (most typically involuntary movements), cognitive impairment, and behavioral changes (e.g., apathy and blunted affect) (Bates et al. 2015). Onset occurs usually in mid-life (average age at onset of about 41 years), although juvenile, infantile, and later onset cases exist (Hayden 1981; Myers et al. 1985). Without effective treatment or prevention, current management consists of multidisciplinary interventions to alleviate some symptoms (Ghosh and Tabrizi 2018). Pre-symptomatic testing (PST) is available for at-risk individuals from families with known history of HD. In Portugal, a national PST program was designed in 1996 to offer genetic counseling and psychosocial support in late-onset neurological diseases (Sequeiros 1996). The PST program is offered through the medical genetic services (Paneque et al. 2015). The profile of Portuguese consultands undertaking PST is similar to those reported in other international programs (Paneque et al. 2019). National psychosocial studies have documented individuals' representations of HD (Leite et al. 2016) and the psychological impact of PST (Lêdo et al. 2018). A recent study using a family-systems perspective showed how an HD diagnosis opened up a process of "making sense" about the disease in the family, contributing to improved awareness and activating a transgenerational process to understand it (Oliveira et al. 2020).

Although the molecular diagnosis and PST have been available for more than two decades, the diagnosis of HD is frequently delayed (Halpin 2011; Oliveira et al. 2020; Pascu et al. 2015). The process of obtaining a diagnosis is described as a long and often painful journey, due to low awareness about HD (from families and healthcare providers), often punctuated by a series of inadequate diagnoses (Halpin 2011; Oliveira et al. 2020; Pascu et al. 2015). Even if the diagnosis is not new, styles of communication about HD within the family vary and may evolve over family members' life-cycle (Brouwer-Dudokdewit et al. 2002). While some families may communicate openly, there are others for whom this proves difficult (Klitzman et al. 2007; Mendes et al. 2018; Etchegary 2006; Forrest et al. 2003). Studies have reported that HD was kept as a secret from some younger relatives, while others found out about it unexpectedly (Etchegary 2006; Sobel and Cowan 2000a; Forrest et al. 2003, 2009).

Families often reorganize their patterns of communication and roles after genetic testing (Sobel and Cowan 2000b). This experience may be most intense for support persons, often extending to their caregiving role (Williams et al. 2000). Different relatives may take up different roles, such as "the messenger of the news" or "the first utilizer" (Brouwer-Dudokdewit et al. 2002).

The role of older generations in families with genetic conditions has been studied mostly in hereditary cancers (Ashida et al. 2011; Koehly et al. 2009), familial amyloid polyneuropathy (Oliveira et al. 2017a, b), cardiovascular disease, and diabetes (Ashida et al. 2010). They are described as performing roles of "health leaders" (Pantaleao et al. 2019), who provide information, support, and encourage younger relatives' engagement in strategies for health promotion and surveillance behaviors (Ashida et al. 2011; Ashida and Schafer 2015; Ashida et al. 2010; Koehly et al. 2009; Oliveira et al. 2017a, b). Older family members play an important role in family's health management, since they act as "keepers of the family medical history," for their privileged access to health information and kinship (Koehly et al. 2009; Mendes 2012).

Roles of older family members may also include withholding and controlling information, mainly through secrecy, about the family disease and non-disclosure of testing results (Delliaux et al. 2008; Forrest et al. 2009; Holt 2006; Oliveira et al. 2017a; Sobel and Cowan 2000a). Other studies reported that some older relatives may discourage younger members to undertake PST (Oliveira et al. 2017a; Klitzman et al. 2007).

Literature is scarce, however, regarding older members' roles in families affected by incurable, highly incapacitating, late-onset genetic diseases, such as HD, particularly when the diagnosis is recent in the family. Therefore, this exploratory qualitative study aims at describing how adjustment to HD occurs, from a family perspective, considering the roles performed by older generations, in the Portuguese context.

Methods

This paper draws on empirical data from a qualitative study exploring inter- and intra-generational management of genetic diseases, within Portuguese families, including HD (Oliveira et al., 2020; Oliveira et al. 2017a, b). It describes the roles performed by older family members in relation to the process of adjustment to the family disease. Data were collected using the critical incidents technique (CIT), which allows for gathering meaningful events from the participants (key informants who experienced critical incidents) (Flanagan 1954; Kemppainen 2000). An incident is an observable major human activity that typically includes a detailed description of a situation, key players, and outcomes. This technique has been used in health contexts (Sousa et al. 2011; Sousa and Ribeiro 2013) and family studies (Sousa et al. 2010).

Procedure

This study was presented to the national HD patients' association who then circulated it among its members. Eligible participants included members from HD families, 18 years or over, and able to consent. Those interested contacted the main researcher (CRO), by telephone or email, who explained the

study in detail and required collaboration. After agreement, informed consent was obtained and interviews were scheduled according to participants' convenience (including how those would be conducted, i.e., in person, via telephone, or video call). This process was complemented by snowball sampling (Silverman 2000). Data collection occurred between August and November 2018. Institutional ethical approval was obtained (UICISA: E-Ref. AD P595-5/2019).

Instrument

Semi-structured interviews were conducted with 10 participants via telephone (8), Skype (1), or in person (1). Average duration was 41.5 min [20–67]. During the interviews, participants were invited to recall situations (CI) involving an older family member that were significant and meaningful, and that had influenced how the family adjusted, managed, and lived with the HD. Participants were asked to think about events they remembered clearly and to describe them in detail (e.g., what impressed them most, who participated, who did what, how did it end). The following data was then collected from participants and from older and younger protagonists: social and demographic (age, gender, and education), disease status (affected, at risk, non-carrier, pre-symptomatic carrier, non-biological family member), and kinship. Participants reported 130 CI (range 3–27).

Participants

The study included 10 participants (narrators), from seven families. Eight were females aged 28 to 60 years (mean age 40), and two were male aged 67 and 72. Two participants had 5–9, three had 10–12 years of schooling, and five had a university degree. Two were non-carriers, two were pre-symptomatic-carriers, and six were non-biological family members (spouse, in-law, or aunt/uncle).

Data analysis

Interviews were audiotaped, transcribed, and submitted to thematic analysis. Our analysis was informed by the family systems theory (Galvin and Young 2010; McDaniel et al. 2006), which describes the family as an interactive social system, influenced by the ongoing development of family members and their evolving relationships with each other. We drew particular attention to the interdependency of family members' roles in how they interpret and manage interactions about HD (Rolland and Williams 2005). Data analysis consisted of a process of successive refinement, involving two independent judges/authors (LS + CRO) and, when needed, a third one (AM), and was centered on roles played by older relatives towards younger ones. The process was as follows: (i) LS and CRO read all the CI, in order to remove non-related material (one incident was eliminated); (ii) afterwards, they independently created a system of categories and subcategories; (iii) then, they met and discussed these systems, and doubts arising were discussed with AM; (iv) the process was repeated until an agreement was reached; and lastly, (v) all CI were classified into the categorization system.

Results

Older family members involved in CI are mainly a parent, grandparent, or uncle/aunt. Participants reported 130 CI (Table 1) and tended to be the younger (83) or older protagonist (36), or an independent witness (11 CI). Data analysis showed two main roles (categories) (Table 1): "shaping awareness about HD" and "influencing HD management."

Shaping awareness about HD

This role (68 CI out of 130) is framed by the little awareness in these families about HD, due to a recent diagnosis in a family member (usually obtained in the last 5–15 years). Members of older generations performed this role either by promoting (56 CI) or hampering (12 CI) awareness about HD in the family.

Promoting awareness is performed by being an "awareness trigger," "living transmitter of the disease," allowing a "retrieved testimony," or "provide information about HD."

The older generations acted as an "awareness trigger" (6 CI) when an older undiagnosed affected relative began showing odd signs or behaviors (involuntary movements, aggressiveness). This led younger members to look for medical help to obtain a diagnosis, which came along with awareness of hereditary risk and the possibility of other relatives being affected ("*a big shock*"). Older relatives are "living transmitters of the disease" (23 CI) when younger relatives witness onset and progression of symptoms in older affected family members. This includes involuntary movements, cognitive (e.g., not recognizing daily objects), and behavioral changes (e.g., eating excessively, cursing, aggressiveness); or deterioration and suffering ("*became a bag of bones*," "*screaming and moaning*," "*choking*"). The "retrieved testimony" (13 CI) occurs due to a recent diagnosis and the need to understand the disease in the family. It occurs in two ways: (i) younger members go back to their memories of older relatives that they now think had HD but were not diagnosed; or (ii) older members narrate cases about deceased relatives they now believe had HD. This process allowed members to identify the affected family side, and to recognize and learn about HD symptoms ("*someone who was always speeding up*," "*often disoriented and falling constantly*," "*dropping things*," "*choked to death*"). In addition, participants may reframe their relatives' behavior that they could not have understood until the

Table 1 Roles performed by the older relatives in families with HD

Categories and subcategories (CI) *N*=130	Interviews excerpts
1. Shaping awareness about HD (68)	
1.1 Promoting awareness (56)	
Awareness trigger (6)	*My father-in-law used to come home with those choreic movements and people started saying he had been drinking. Then I tried to see what he was doing and in fact (…) there was nothing that justified those movements. Then the symptoms got worse, and Dr.* [name of doctor] *went to see him and he immediately diagnosed Huntington disease.* (Man, 72, non-biological family member)
Living transmitters of the disease (23)	*The image of him* [father-in-law] *that remained in all of us was that image of skin, bones and suffering.* (Woman, 44, non-biological family member)
Retrieved testimony (13)	*My great grandmother at the time, at least from stories in the family, was said to be a witch, because of the involuntary movements.* (Woman, 37, pre-symptomatic carrier)
Provide information about HD (14)	*My mother* [spouse of an affected carrier] *searched for all the information she could (…). She went to the patient's association, tried to talk to the best doctors, to know all the national and international research available.* (Woman, 29, non-carrier)
1.2 Hampering awareness (12)	
Denial (7)	*We were leaving the hospital* [after seeing the neurologist], *I turned to him and said: "let's go schedule an appointment", to which he replies: "No, I am not sick, the doctor is, I have nothing". My father never assumed he had an illness.* (Woman, 37, pre-symptomatic carrier)
Silence (5)	*She* [affected mother] *knows she has her father's disease but she never opened up a lot. She never mentioned it, she never talked about it, never, ever.* (Woman, 33, pre-symptomatic carrier)
2. Influencing HD management (62)	
2.1 Supporting or not (37)	
Providing and receiving support (29)	*I am now the oldest in the family (…). I am the one who has been supporting the family, not only concerning HD but in all other domains* (Man, 72, non-biological family member, son-in-law of affected carrier, uncle of at-risk members)
Not providing support (2)	*My family just doesn't care, in fact, they pass by the door of my house and don't even call or ring the bell to find out if she* [affected mother] *is alive.* (Woman, 33, pre-symptomatic carrier)
Discouraging PST (3)	*It's on him* [at risk son] *to decide if he wants to undertake PST or not, but our opinion, which we shared with him, is that before he decides to have children…then yes,* […] *but he is now too young to carry this burden right? He's 23 now, so I think he still has a lot of time (…)*(Woman, 43, non-biological family member, spouse of affected carrier)
Encouraging/supporting any decision about PST (3)	*I told him* [nephew] *when you want to perform the test just say it, if you don't want to do it you don't have to.* (Woman, 44, non-biological family member)
2.2 Modeling health-related behaviors (25)	
Normalize (10)	*I always kept things as normal as possible. I tell my son* [at risk] *"Your friends know that your father is sick so we will not stop doing things"* (Woman, 60, non-biological family members, spouse of affected carrier)
Disrupt (9)	*Going to appointments with my mother and she admitting that she could put an end to her life (…) I will never forget that she had the courage to say that in front of my sister and me.* (Woman, 28, non-carrier)
Advocacy (6)	*I tried as a self-taught person to learn everything everywhere I could, and I feel bad for not being able to share this. So, I had in mind going to the Hospital of* [name of town] *and talk to social workers there, and find out if they know of some patients, and share some information on how to help these patients (…). Also, to share my testimony with nursing students.* (Man, 67, non-biological family member, spouse of affected carrier)

diagnosis of HD was established (e.g., detachment, apathy, aggression, instances where affected relatives were named as "*weird*," "*bipolar*," "*possessed by witchcraft*"). Older relatives "provide information about HD" (14 CI) when they carry out efforts to obtain knowledge about the disease, by contacting the patients' association, health professionals, reading recent research, and/or attending conferences. They share this information with younger relatives by instructing them on symptoms, genetic risk and reproductive options, and updating them on the latest research: "*My mother went to the patients' association, tried to talk to the best doctors, to get access to all the research on HD*."

Hampering awareness involves denial or silence. Some older family members "deny" the existence of HD in the

family (7 CI). Participants reported two situations: despite having symptoms, affected family members refuse to accept that they are sick and reject help; or older non-biological relatives refuse to accept that "*someone in the family is guilty*" for transmitting the disease. "Silence" (5 CI) involved not speaking or sharing limited information about the family's past or (supposedly) affected members from former generations. Younger family members tend to consider silence as reflecting fear of shame and stigma attached to the disease: "*my father's family was very afraid of what others could think or say*."

Influencing HD management

This role (62 CI out of 130) suggests that family members from older generations influence younger members on how to manage HD. They performed their role by supporting them or not (37 CI) and by modeling health-related behaviors (25).

"Providing and receiving support" (29 CI) was a frequent role that takes on an emotional and a practical dimension. Emotional support is directed mostly towards relatives at risk or affected and non-affected family members who care for an affected relative. It was accomplished by being available to talk or accepting the younger relative's silence in relation to HD, by making them feel accepted and loved, or by creating opportunities for family members to spend time together. The practical dimension is played towards affected dependent relatives; in that case, the role of older affected family members was typically to "receive support," usually from their children. This had a major impact in the younger person's life and professional options ("*I have no life. I've got my mum* [with HD]*, that's it! This is my life*"). "Not providing support" was rare (2 CI); however, two participants described situations where older family members did not support their affected relatives (one participant mentioned her father-in-law abandoned his severely sick wife, when his son was very young). Current younger generations were usually the first to undertake PST, since previous generations were unaware of the disease. Some older relatives explicitly "discouraged PST" (3 CI), as they believed younger relatives were not ready to deal with an unfavorable result. Others "encourage or support any decision" (3 CI), usually when younger members have not yet decided whether to uptake PST or not. An older family member plainly encouraged her daughter to uptake PST, referring that she "*has to do the test*" to avoid transmitting the disease to future offspring.

"Modelling health related behaviours" (25 CI) comprises events in which the older persons influence the younger ones by their own example. Some older family members tended to "normalize" their experience with the disease within the family (10 CI), through two main circumstances: (a) older affected members trying to remain optimistic, adapting and keeping a *normal* life despite being ill (e.g., going to parties, to the beach); (b) older non-biological family members keeping on with their lives as normally as possible (e.g., not showing fear or being scared by HD, not being ashamed of their relatives' symptoms), and including the younger members in the care of affected ones: "*she* [mother, spouse of an affected carrier] *was always committed to not giving up, not getting depressed*." To *disrupt* (9 CI) the HD, experience occurred when older affected members showed signs of *giving up* or hopelessness. Some isolated or withdrew from social activities (they tend to think that they always "*mess things up*" and that others perceive them as "*crazy*"). Sometimes, they blame themselves for not having been able to identify the disease in deceased family members, despite their symptoms, and having had children at risk for HD. Some older (usually non-biological) relatives became "advocates" (6 CI), supporting awareness about HD outside the family (e.g., being active in the patients' association, and sharing their experience among students, health professionals, and the community).

Discussion

This study aimed at describing adjustment to HD in the family, considering the roles performed by members of older generations towards younger ones. These roles need to be interpreted considering that most families in our study had a recent diagnosis of HD or reported limited knowledge about the disease (when compared to other family's experiences' or other genetic conditions which have been known in the family over generations).

Although this was not an inclusion criterion, accounts from most participants report a recent diagnosis of the HD in the family, as previously reported (Oliveira et al. 2020).

The roles emerging in this study, in comparison with data available for other genetic conditions, such as FAP (Oliveira et al. 2017a) and hereditary cancers (Ashida et al. 2011), portray participant families as "beginners" in understanding and incorporating HD in their lives. Therefore, findings convey the roles performed by older relatives in the process of adjustment to HD in the family, which can be considered an unexpected, although required, task in the family life cycle. Literature on family life cycle (Carter and McGoldrick 1995, 1999; Patrão and Sousa 2010) highlights that a task in family development involves the following: precipitant events; emotional experience with meanings; challenges regarding management; and resolution and integration. Roles performed by older generations seem to follow and support this process (Table 2).

The *precipitant event* here was the diagnosis of HD in an affected member (the *proband*) of the family. Older affected relatives sometimes act as the *awareness trigger*, since the younger ones witness their symptoms and launch an *odyssey* to obtain a diagnosis (Oliveira et al. 2020). With the diagnosis of HD comes knowledge of the hereditary nature and absence of treatment or cure, which shapes new *meanings*. One relevant meaning is that the disease is not new, but (in most instances) has been

Table 2 Family adjustment to HD: a task in the family life cycle

Older relatives' roles		
Family life cycle: task components	Categories	Subcategories
Precipitant events	Shaping awareness	Promoting awareness: awareness trigger
Meanings		Promoting awareness: living transmitters of the disease; retrieved testimony; provide information about HD Hampering awareness: denial; silence
Management	Influencing HD management	Not/supporting: providing and receiving support; not providing support Encourage, support any decision about PST; discourage PST
Towards resolution and integration		Modeling health-related behaviors: normalize; disrupt; advocacy

present in the family for a long time; therefore, what is new is the awareness about it and its implications. Families may now build on a new (biomedical) understanding about the disease (symptoms, progression) and the family (from which side it came, who had it, who has it now, or might develop it in the future). It also allows reframing the family history around HD (e.g., stories of persons with odd behaviors) and some family relationships (e.g., a detached or aggressive parent). Older relatives contribute to meaning-making by *shaping awareness* about the disease, mostly by *promoting* it through sharing their disease experience in multiple ways: living transmitters of the disease, retrieved testimony, and providing information about HD. Distinctively, when older family members hamper awareness about HD, understanding of the disease and the family history might be delayed and/or blocked among younger relatives. This also limits younger members' options with whom to discuss risk and disease management options within the family. However, this context may perhaps prompt younger relatives to find alternative communication channels that suit their needs.

Still, when older family members facilitate awareness, new meanings arise (e.g., from "a family of mad people," to "a family of ill persons"). Acknowledgment of HD allows some answers and raises the challenge of incorporating genetic risk in families' lives (Klitzman 2009; Sobel and Cowan 2000b). This encompasses the need to reprocess the family history and alternative illness explanations running in the family, in order to incorporate new information, with probable impact on family identity (Ferring 2017; Klitzman 2009; Patterson and Garwick 1994; Phipps and Lazzarini 1987; Walter and Emery 2005). Awareness of its hereditary nature comes with new concerns and uncertainty about the present and the future (for themselves and other family members, including future generations). This includes the need to deal with medical interventions (e.g., PST, assisted reproductive technologies), but also with anticipated loss and fatalism (Halpin 2018; Rolland 2006; Werner-Lin and Gardner 2009). Such challenges require the emergence of specific roles for families to manage HD. This study showed that some management roles were already being performed by older relatives, namely providing and receiving support, although now challenged by the new meanings and foreseen implications of HD. Families must deal with the coexistence of members with different disease status; therefore, providing support assumes an extended scope for the family system. Some older relatives provide emotional and practical support to younger members who take care of an affected parent; others emotionally support relatives who are still in the process of making decisions about PST. Dealing with the possibilities associated with the genetic risk (namely PST) is a new management demand. The roles related to PST, *encourage and support any decision and discourage*, elicited a low number of CI, probably because the challenges of HD are still being incorporated into family's functioning, but also because some families communicate less openly about genetic testing decisions (Forrest et al. 2003; Sobel and Cowan 2000a, b).

Older relatives seem to be contributing towards a process of *resolution and integration*, by modeling health-related behaviors, which comprise *normalize*, *disrupt*, and *advocacy*. To *normalize* involves accepting the disease and to go on with life, as normal as possible. To *disrupt* is portrayed by difficulties in coping with HD, such as isolation, withdrawal, or despair. This can contribute to inhibit better coping strategies among younger members, causing difficulties in facing challenges and decisions (namely concerning PST, and other life planning issues, in general) and perpetuating stigma across generations.

Similar roles have been assigned to older family members with FAP, in which *normalizing* the illness experience seemed to help younger members making the disease compatible with other family, social, or professional functions, while *dramatizing* seemed to be associated with fear and hopelessness (Oliveira et al. 2017a, b).

Some older relatives assume the role of *advocates*, aiming at disseminating awareness about the disease in the society; this has been suggested as evidencing loyalty and protection towards the family, while also highlighting its normalcy (Hays and Colaner 2016). This can possibly be seen as a sign of change onto more open communication styles, from some family members. However, it does not eliminate the coexistence of different-lived experiences among the younger generations (including, e.g., limited awareness, misinformation, and isolation).

Adjustment to HD in the family is a transition moment that implies coexistence with other normative tasks of the individual and family life cycle. For example, families are confronted with the need to make decisions regarding communication of disease-related information, while transitioning between developmental stages (e.g., from being a "single young adult" into forming a "new couple" or "having children"). HD has been established as a paradigmatic example of a progressive, incurable, and fatal condition to inform the study of adjustment to genetic illnesses in the context of the family's life cycle (Brouwer-Dudokdewit et al. 2002). Families receiving a diagnosis of a genetic condition for the first time will probably undergo this task at some point in their adjustment; in particular, it is likely that this task becomes more salient when diagnosis is recent or knowledge about the disease in the family is generally scarce. This can be particularly relevant in a context with more disorders being recognized as having an inherited component and individuals and families increasingly facing the possibility of broad-based genetic testing (Bailey et al. 2014).

Implications for practice

Families who receive a diagnosis of HD go through a development task of adjustment, in which older relatives may play a relevant role. This process entails the need to reframe or retell a coherent narrative about the family, with probable impact in family identity (Armstrong et al. 1998; Kellas 2005; Petersen 2006). Within this process, older relatives are determinant to understand the family history and support younger generations through this task. Genetic counseling and interventions aimed at supporting families should considerer a narrative approach specifically involving older relatives, since they have a great influence in sustaining family stories (Mendes 2012; Merrill and Fivush 2016). A narrative-based approach has shown promising results in HD families (Stopford et al. 2020). Also, acknowledgement of how older generations cope with illness demands is important to understand the family's style of adjustment, and to identify their strengths and vulnerabilities in this process (Carter and McGoldrick 1999; Rolland and Williams 2005).

Limitations and perspectives of research

The main limitation of this study is its reduced sample size and its limited diversity (namely without affected or at-risk participants and composed mostly by non-biological family members). HD is a rare disease and larger and more diverse samples are difficult to assemble. Also, although not explicitly stated by the participants, we sensed that they avoided involving some relatives in the study, perhaps to protect them (anticipating they would not feel comfortable or fearing any negative impact to them). Nevertheless, further research should attempt to include a larger sample, more heterogeneous considering gender, age, education, and disease status, what may allow a wider perspective on the older generations' roles regarding adjustment of the family to HD. Also, it would allow to clarify some emerging aspects in our participants' accounts, namely if the adjustment to a "new diagnosis" is the most common experience for HD families in the Portuguese context or merely reflects our sample experience. Future research could also explore (e.g., by including it in the interview schedule) what did not impress the participants in the events they recalled (CI). Additionally, considering that families might be experiencing a transition moment, a longitudinal approach should be envisaged in order to analyze how older generations' roles evolve in the individual and family life cycles.

The data collection method, mostly performed via telephone, has been used by the authors in previous studies, with good results. This process might allow participants to feel more comfortable to share sensitive information (Novick 2008). We acknowledge, however, that this option could limit access to contextual and some nonverbal data that might have interfered with the results.

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Declarations

Institutional ethical approval was obtained (UICISA: E-Ref. AD P595-5/2019). All procedures were in accordance with the national ethical standards and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all participants included in the study.

Conflict of interest The authors declare no competing interests.

References

Armstrong D, Michie S, Marteau T (1998) Revealed identity: a study of the process of genetic counselling. Soc Sci Med 47:1653–1658. https://doi.org/10.1016/s0277-9536(98)00241-x

Ashida S, Hadley DW, Goergen AF, Skapinsky KF, Devlin HC, Koehly LM (2011) The importance of older family members in providing social resources and promoting cancer screening in families with a

hereditary cancer syndrome. Gerontologist 51:833–842. https://doi.org/10.1093/geront/gnr049
Ashida S, Schafer EJ (2015) Family health information sharing among older adults: reaching more family members. J Commun Genet 6:17–27. https://doi.org/10.1007/s12687-014-0197-x
Ashida S, Wilkinson AV, Koehly LM (2010) Motivation for health screening: evaluation of social influence among Mexican-American adults. Am J Prev Med 38:396–402. https://doi.org/10.1016/j.amepre.2009.12.028
Bailey DB, Lewis MA, Roche M, Powell CM (2014) Family relations in the genomic era: communicating about intergenerational transmission of risk for disability. Fam Relat 63:85–100. https://doi.org/10.1111/fare.12054
Bates GP, Dorsey R, Gusella JF, Hayden MR, Kay C, Leavitt BR, Nance M, Ross CA, Scahill RI, Wetzel R, Wild J, Tabrizi SJ (2015) Huntington disease. Nat Rev Dis Primers 1:15005. https://doi.org/10.1038/nrdp.2015.5
Brouwer-Dudokdewit AC, Savenije A, Zoeteweij MW, Maat-Kievit A, Tibben A (2002) A hereditary disorder in the family and the family life cycle: Huntington disease as a paradigm. Fam Process 41:677–692. https://doi.org/10.1111/j.1545-5300.2002.00677.x
Carter B, McGoldrick M (1995) As mudanças no ciclo de vida familiar: uma estrutura para a terapia familiar, Artmed
Carter B, McGoldrick M (1999) The expanded family life cycle. In: Individual, Family and Social Perspectives, 3rd edn. Allyn and Bacon
Delliaux M, Delval A, Krystkowiak P, Destee A, Defebvre L, Dujardin K (2008) About Huntington's disease: role of families and health professionals in information transmission. Rev Neurol (Paris) 164:148–155. https://doi.org/10.1016/j.neurol.2007.08.002
Etchegary H (2006) Discovering the family history of Huntington disease (HD). J Genet Couns 15:105–117. https://doi.org/10.1007/s10897-006-9018-7
Ferring D (2017) The family in us: family history, family identity and self-reproductive adaptive behavior. Integr Psychol Behav Sci 51:195–204. https://doi.org/10.1007/s12124-017-9383-9
Flanagan JC (1954) The critical incident technique. Psychol Bull 51:327–358. https://doi.org/10.1037/h0061470
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 family communication about genetic risk. Clin Genet 64:317–326. https://doi.org/10.1034/j.1399-0004.2003.00142.x
Forrest K, van Tejlingen E, McKee L, Miedzybrodzka Z, Simpson S (2009) How young people find out about their family history of Huntington's disease. Soc Sci Med 68:1892–1900. https://doi.org/10.1016/j.socscimed.2009.02.049
Galvin K, Young M (2010) Family systems theory. In: Gaff C, Bylund C (eds) Family Communication about Genetics: Theory and Practice. Oxford University Press, pp 102–119
Ghosh R, Tabrizi SJ (2018) Clinical features of Huntington's disease. In: Nóbrega C, Pereira de Almeida L (eds) Polyglutamine Disorders. Advances in Experimental Medicine and Biology. Springer, pp 1–28
Halpin M (2011) Diagnosis, psychiatry and neurology: the case of Huntington Disease. Soc Sci Med 73:858–865. https://doi.org/10.1016/j.socscimed.2011.03.034
Halpin M (2018) Science and suffering: genetics and the lived experience of illness. Soc Probl 65:360–376. https://doi.org/10.1093/socpro/spw057
Hayden MR (1981) Huntington's Chorea. Springer-Verlag, London
Hays A, Colaner C (2016) Discursively constructing a family identity after an autism diagnosis: trials, tribulations, and triumphs. J Fam Commun 16:143–159. https://doi.org/10.1080/15267431.2016.1146722
Holt K (2006) What do we tell the children? Contrasting the disclosure choices of two HD families regarding risk status and predictive genetic testing. J Genet Couns 15:253–265. https://doi.org/10.1007/s10897-006-9021-z
Kellas JK (2005) Family ties: communicating identity through jointly told family stories. Commun Monogr 72:365–389. https://doi.org/10.1080/03637750500322453
Kemppainen JK (2000) The critical incident technique and nursing care quality research. J Adv Nurs 32:1264–1271. https://doi.org/10.1046/j.1365-2648.2000.01597.x
Klitzman R, Thorne D, Williamson J, Marder K (2007) The roles of family members, health care workers, and others in decision-making processes about genetic testing among individuals at risk for Huntington disease. Genet Med 9:358–371. https://doi.org/10.1097/GIM.0b013e3180653c5a
Klitzman R (2009) "Am I my genes?": questions of identity among individuals confronting genetic disease. Genet Med 11:880–889. https://doi.org/10.1097/GIM.0b013e3181bfd212
Koehly LM, Peters JA, Kenen R, Hoskins LM, Ersig AL, Kuhn NR, Loud JT, Greene MH (2009) Characteristics of health information gatherers, disseminators, and blockers within families at risk of hereditary cancer: implications for family health communication interventions. Am J Public Health 99:2203–2209. https://doi.org/10.2105/AJPH.2008.154096
Lêdo S, Ramires A, Leite A, Dinis MA, Sequeiros J (2018) Long-term predictors for psychological outcome of pre-symptomatic testing for late-onset neurological diseases. Eur J Med Genet 61:575–580. https://doi.org/10.1016/j.ejmg.2018.03.010
Leite A, Dinis MA, Sequeiros J, Paúl C (2016) Subjects at-risk for genetic diseases in portugal: illness representations. J Genet Couns 25:79–89. https://doi.org/10.1007/s10897-015-9846-4
McDaniel SH (2005) The psychotherapy of genetics. Fam Process 44:25–44. https://doi.org/10.1111/j.1545-5300.2005.00040.x
McDaniel S, Rolland J, Rubin L, Miller S (2006) It runs in the family: family systems concepts and genetically linked disorders. In: Miller S, McDaniel S, Rolland J, Feethen S (eds) Individuals, families and the new era of genetics: Biopsychosocial perspectives. Norton, New York, pp 118–318
Mendes Á (2012) Doenças hereditárias, aconselhamento genético e redes familiares e sociais: da ética intergeracional ao papel dos mais velhos. Revista Kairós. Gerontologia 15:199–216. https://doi.org/10.23925/2176-901X.2012v15iEspecial11p199-216
Mendes A, Metcalfe A, Paneque M, Sousa L, Clarke A, Sequeiros J (2018) Communication of information about genetic risks: putting families at the center. Fam Process 57:836–846. https://doi.org/10.1111/famp.12306
Merrill N, Fivush R (2016) Intergenerational narratives and identity across development. Dev Rev 40:72–93. https://doi.org/10.1016/j.dr.2016.03.001
Myers RH, Sax DS, Schoenfeld M, Bird ED, Wolf PA, Vonsattel JP, White RF, Martin JB (1985) Late onset of Huntington's disease. J Neurol Neurosurg Psychiatry 48:530–534. https://doi.org/10.1136/jnnp.48.6.530
Novick G (2008) Is there a bias against telephone interviews in qualitative research? Res Nurs Health 31:391–398. https://doi.org/10.1002/nur.20259
Oliveira CR, Mendes Á, Sequeiros J, Sousa L (2020) Management of information within Portuguese families with Huntington disease: a transgenerational process for putting the puzzle together. Eur J Hum Genet 28:1210–1217. https://doi.org/10.1038/s41431-020-0630-z
Oliveira CR, Mendes A, Sousa L (2017a) Health promotion in families with paramyloidosis: the role of elders with younger family members. Cad Saude Publica 33:e00185515. https://doi.org/10.1590/0102-311x00185515
Oliveira CR, Mendes A, Sousa L (2017b) From older to younger: intergenerational promotion of health behaviours in Portuguese families affected by familial amyloid polyneuropathy. Eur J Hum Genet 25:687–693. https://doi.org/10.1038/ejhg.2017.40

Paneque M, Mendes A, Saraiva J, Sequeiros J (2015) Genetic counseling in Portugal: education, practice and a developing profession. J Genet Couns 24:548–552. https://doi.org/10.1007/s10897-015-9827-7

Paneque M, Félix J, Mendes Á, Lemos C, Lêdo S, Silva J, Sequeiros J (2019) Twenty years of a pre-symptomatic testing protocol for late-onset neurological diseases in Portugal. Acta Medica Port 32:295–304. https://doi.org/10.20344/amp.10526

Pantaleao A, Young JL, Epstein NB, Carlson M, Bremer RC, Khincha PP, Peters JA, Greene MH, Roy K, Achatz MI, Savage SA, Werner-Lin A (2019) Family health leaders: lessons on living with Li-Fraumeni syndrome across generations. Fam Process 10:1–16. https://doi.org/10.1111/famp.12497

Pascu AM, Ifteni P, Teodorescu A, Burtea V, Correll CU (2015) Delayed identification and diagnosis of Huntington's disease due to psychiatric symptoms. Int J Ment Heal Syst 9:33. https://doi.org/10.1186/s13033-015-0026-6

Patrão M, Sousa L (2010) Transmissão da herança material: uma tarefa normativa das famílias envelhecidas. Psychologica 1:371–393. https://doi.org/10.14195/1647-8606_52-1_18

Patterson J, Garwick A (1994) The impact of chronic illness on families: a family systems perspective. Ann Behav Med 16:131–142

Petersen A (2006) The best experts: the narratives of those who have a genetic condition. Soc Sci Med 63:32–42

Phipps EJ, Lazzarini A (1987) Fighting dragons: the construction of explanatory systems in genetic disease. Fam Syst Med 5:304–312. https://doi.org/10.1037/h0089723

Rolland JS (2006) Living with anticipatory loss in the new era of genetics: a life cycle perspective. In: Miller SM, SH MD, Rolland JS, Feetham SL (eds) Individuals, families, and the new era of genetics: Biopsychosocial perspectives. W W Norton & Co, pp 139–172

Rolland JS, Williams JK (2005) Toward a biopsychosocial model for 21st-century genetics. Fam Process 44:3–24. https://doi.org/10.1111/j.1545-5300.2005.00039.x

Sequeiros J (1996) O teste preditivo da doença de Machado-Joseph. Instituto De Biologia Molecular e Celular, Universidade do Porto, Porto

Silverman D (2000) Doing qualitative research: A practical handbook. SAGE

Sobel S, Cowan DB (2000) Impact of genetic testing for Huntington disease on the family system. Am J Med Genet 90:49–59

Sobel S, Cowan DB (2000b) The process of family reconstruction after DNA testing for Huntington disease. J Genet Couns 9:237–251. https://doi.org/10.1023/A:1009416021896

Sousa L, Almeida A, Simões CJ (2011) Emergency room: the role of the accompanying person. Saúde e Soc 20:195–206. https://doi.org/10.1590/S0104-12902011000100021

Sousa L, Ribeiro AP (2013) Nursing care for elderly people: experiences and impacts. Saúde e Soc 22:866–877. https://doi.org/10.1590/S0104-12902013000300019

Sousa L, Silva AR, Santos L, Patrão M (2010) The family inheritance process: motivations and patterns of interaction. Eur J Ageing 7:5–15. https://doi.org/10.1007/s10433-010-0139-3

Stopford C, Ferrer-Duch M, Moldovan R, MacLeod R (2020) Improving follow up after predictive testing in Huntington's disease: evaluating a genetic counselling narrative group session. J Commun Genet 11:47–58. https://doi.org/10.1007/s12687-019-00416-9

Street E, Soldan J (1998) A conceptual framework for the psychosocial issues faced by families with genetic conditions. Fam Syst Health 16:217–232. https://doi.org/10.1037/h0089851

Walter FM, Emery J (2005) "Coming down the line"- patients' understanding of their family history of common chronic disease. Ann Fam Med 3:405–414. https://doi.org/10.1370/afm.368

Werner-Lin A, Gardner DS (2009) Family illness narratives of inherited cancer risk: continuity and transformation. Fam Syst Health 27:201–212. https://doi.org/10.1037/a0016983

Williams JK, Schutte DL, Holkup PA, Evers C, Muilenburg A (2000) Psychosocial impact of predictive testing for Huntington disease on support persons. Am J Med Genet 96:353–359