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The personal experience of parenting a child with Juvenile Huntington's disease: perceptions across Europe

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Running title: Parenting and Juvenile Huntington's disease

Abstract

The study reported here presents a detailed description of what-it-is-like to parent a child with juvenile Huntington's disease in families across four European countries. Its primary aim is to develop and extend findings from a previous UK study. The study recruited parents from four European countries: Holland, Italy, Poland and Sweden,. A secondary aim is to see the extent to which the findings from the UK study were repeated across Europe and the degree of commonality or divergence across the different countries. Fourteen parents who were the primary care giver took part in a semi-structured interview. These were analysed using an established qualitative methodology, interpretative phenomenological analysis. Five analytic themes were derived from the analysis: the early signs of something wrong; parental understanding of juvenile Huntington's disease; living with the disease; other people's knowledge and understanding; need for support. These are discussed in light of the considerable convergence between the experiences of families in the UK and elsewhere in Europe.

Keywords: Juvenile Huntington's disease; parent; qualitative

Introduction

Huntington's disease (HD) is an inherited progressive neurodegenerative disorder and defining features include affective disturbance, cognitive dysfunction and movement impairment. There is a wide variation in the age of onset but it is often in mid life[1]. Onset occurring ≤ 20 years is classified arbitrarily as Juvenile onset HD (JHD) [2]. The youngest symptomatic child documented had an age of onset of 18 months[3] with other children developing symptoms in their teens, meaning that some individuals with JHD may now be in their twenties. HD is a life-limiting condition usually lasting 15-20 years from onset[1]. HD affects approximately 7-10 in every 100,000 of the population[4, 5, 6] but

may be higher[7]. A recent meta-analysis shows the proportion of cases with JHD in Northern Europe and North America is 4.81% (95% confidence interval 3.31% to 6.58%) meaning that that there are approximately 5 cases per million of the population[8]. Thus JHD is considered a rare condition.

Clinical presentation in children varies from that of adults with behavioural disturbance and failure at school predominating. Early motor abnormalities often include rigidity, clumsiness and unsteady gait with the choreic movements associated with adults being less common[9, 10]. The rare nature of JHD means that it is less well recognized than HD and as a consequence even less is known about the condition including the psychosocial effects on both the child with JHD and their family. A detailed understanding of what it is like to care for a child with JHD and its impact on individual and family functioning is important so that suitable psychosocial interventions can be developed. Given that there is currently no cure but only palliative treatment, research which focuses on the personal experience of families with a JHD child is especially relevant.

A recent qualitative UK study aimed to identify the health and social care needs of children affected by JHD and issues faced by the family. Key findings included: how families experienced isolation and a lack of understanding about JHD; how many parents were aware that something was wrong with their child; perceptions of helpful and unhelpful forms of social support from families, friends and healthcare professionals[11, 12, 13].

The study reported here aims to develop and extend existing understanding of the personal experience of families living with a child with JHD; how they think, feel and manage the challenges that face them. The study recruited parents from four European countries: Italy, Netherlands, Poland and Sweden. The aim was to see the extent to which the findings from the UK study were repeated across Europe and the degree of commonality or divergence across the different countries.

This study employed in-depth semi-structured interviews and interpretative phenomenological analysis[14] (IPA), as in the UK study. IPA is a well-established experiential approach in health and

clinical psychology, especially suitable for investigating comparatively novel areas of research such as JHD. Also IPA studies address aspects of life experienced as significant in some way: transformative or threatening events, decisions or conditions, and their emotional impact. IPA has been employed in a number studies examining psychosocial issues in genetics, for example[15, 16, 17] , in addition to the previous work on JHD[11, 12, 13].

Method

Participants

We aimed to include a diverse range of cultural groups. Hence we recruited from the following regions of Europe: North (Sweden), North West (Netherlands), South (Italy), (Central (Poland). Fourteen parents who were the primary care giver of a young person with JHD were invited to take part. Thirteen participants were mothers of affected children and one participant was a father. In each case the participant was the non-affected parent. For 12 of the 14 families, the affected parent was either deceased or did not live at home. Names and other identifying information have been changed and each participant given a pseudonym. See Table 1 for country and age of affected child.

Procedure

The necessary ethical approval was obtained in each country. An interview schedule was developed in English and translated into the language for each country. Participants were recruited through clinical genetics or neurological services in each country. An interviewer with sufficient experience of qualitative research conducted interviews in the native language. These were translated into English and transcribed verbatim. Researchers proficient in both languages were responsible for translation and other members of the research team carried out a subsequent quality check. Interviewers were trained to offer appropriate and sensitive support in the event of a participant becoming distressed.

The transcripts were analysed using IPA. The first stage involved close readings of an individual transcript to obtain a holistic sense of the participant's story. This was followed by a line-by-line analysis of the participant's experiential concerns. These initial notes and observations were examined and developed into preliminary themes capturing the key characteristics of the participant's experience. Preliminary themes were reviewed and refined which involved combining, condensing and sometimes renaming them. These steps comprise an iterative process during which the researcher moves back and forth between the various analytic stages to ensure that the emerging thematic structure reflects the essence of the participant's experience. Related themes are then clustered. These analytic stages were followed for each transcript after which the themes for each individual were compared and a final table of shared themes was constructed. The results section provides a description and commentary of the themes evidenced with verbatim extracts from the interview material. For editorial succinctness, '…' indicates where some material has been removed. Points of clarification by the authors are contained in square brackets.

Results

Early signs of JHD: "I worried, wondered and suspected"

These parents had a strong sense that something was wrong with their children before help was sought and/or a diagnosis was made. Their words point to the often-overwhelming feeling of 'knowing-but-not-knowing':

> When I heard this [diagnosis], it merely confirmed my suspicions. So the news was no shock. It was just confirmation. I had been suspecting it for two to three years. It was hard going and I had no help during this time… I worried, wondered, and suspected all those years. It was mentally extremely trying. (Berta, Sweden)

Often, these suspicions were difficult to articulate initially:

> It was something in the way she looked at you, in her eyes, the way she moved. (Inge, Sweden)

> Weird, strange, sometimes completely absent and locked in her world, sometimes too vibrant, restless, very irritable. (Carla, Italy)

Once alerted, this initially hazy awareness that something is not quite right begins to crystallize. Due to bullying at school, Hanne spent more time with her daughter:

> Then you see more than I had noticed before. Well, just the way she moved and her clumsiness. It was just as if suddenly there was this magnifying glass in front of everything which made you recognize everything much better. (Hanne, Holland)

In addition, participants 'knew' because they compared their children to siblings and the children of other people:

> Ever since she was one-year-old and started to walk a bit, you could tell that she wasn't entirely like other kids. (Benjamin, Holland)

For Benjamin, in comparison to other children, his daughter was always "very late" in learning to do something whether it was not eating messily or riding a bike. Saskia compares her son with his siblings:

> He was a very difficult child from the moment he was born…compared to the other two; he was a completely different child. (Saskia, Holland)

Similarly, Bernadette referred to her son not being able to do things other children could with the added pressure that he was expected to in school:

> He did lag behind, he couldn't do certain things and then he started developing a certain tension too. He could no longer keep up that well but that was still required of him. So in the end, it all escalated. Like you really had a different child. (Bernadette, Holland)

Here, not only is the child with JHD different from other children, he is different from the child he was. Over time, the disease manifests itself in new ways which seemingly transforms the child in the eyes of the parent.

The participants' accounts are replete with words such as "funny" and "strange" to describe the physical changes they saw in their children. They expressed feelings of distress and worry as they saw them alter:

> He was not falling down but he started walking like an old man, giving the impression of being tired and this started disturbing me. (Joanna, Poland)

Joanna said her son had always been a "lively energetic child, always up to mischief" whereas now she describes him as an "old man". Other participants noted more psychological changes describing their children as "sad", "distracted", "depressed" and "careless". A sense of the children fading permeated the accounts:

> Before he was more energetic, he went to football school. Something like an internal slowness. (Rosa, Italy)

> I see my daughter, I mean day-by-day turning off. (Leonia, Italy)

Regardless of any knowledge they had about the disease and its presence in their families, these parents had a powerful awareness that something was wrong. In our view these parental concerns and observations are of importance in alerting doctors to a potential problem. Recognising that

parents are, in some sense, the 'experiential experts' of their children means that early signs can be attended to, despite the difficulties in establishing a firm diagnosis before these signs are unequivocal.

Parental understanding of JHD: "It is a sentence"

Parents describe the immense impact of caring for a child with JHD. They often use striking images to convey the significance and emotional impact of the disease:

> HD is a devastating disease, is a monster because it affects everything that makes the human person – mind, emotions, feelings, everything in short, the loss of personal autonomy and in this moment of my life I do not see a disease worse than this. The image that comes to mind is an octopus who takes all…it sucks in everything. (Teodora, Italy)

> I am lost for words, feel helpless. It's a terrible disease, the worst being that there is no cure or possibility of arresting the progress of the disease. For me and my child it is a sentence. (Irena, Poland)

JHD is devastating for many parents because the disease does not let their children be children in the short life they have:

> She does not have friends with whom she can be in her leisure time, close friends so she is alone, an outsider. (Berta, Sweden)

> The most painful for me is his loneliness, he is deprived of his friends and meeting friends, he is just isolated. (Kazia, Poland)

It is painful for any parent to see their child lonely and describing her daughter as an ‘outsider’ emphasizes how Berta perceives the disease to have denied her daughter the opportunity to be a ‘normal’ child in the time she has.

JHD was described as a latent disease, letting parents believe their child was healthy:

> It would have been easier if we would have known this from the beginning, that it had been visible since birth…I simply feel deceived that she was like a normal healthy child, healthy and growing, just like any normal healthy child. (Inge, Sweden)

Inge suggested that a ‘visible’ illness clearly identified at birth is more easily faced and managed because expectations for the future are, from the outset, revised.

Many of these parents reflected upon how the disease had derailed their lives leaving them feeling hopeless and powerless:

> I have lost my hope [crying]. This disease deprived me of joy of life; there is nothing that could make me really happy. (Kazia, Poland)

One participant described how JHD inures one from other people’s misfortune:

> The mothers too get twisted minds in the long run; it has an enormous effect on them. A lot of things just don’t influence your life anymore…the lady next door has cancer, bowel cancer, that’s very serious. Well, I guess so but there’s still hope at least. (Hanne, Holland)

The enormity of caring for a child with JHD means that there is no room to extend care and concern to others. The phrase “twisted minds” hints at a skewed perspective of life which is perceived to be a direct consequence of the disease.

Living with JHD: "Everything changes"

Most of the participants emphasized how JHD means that life has become painstaking and repetitive with little or no room for spontaneity:

> Everything changes. You always have to take account of it. When we go somewhere I have to prepare her for it, tell her we're going to do this or that today or tomorrow and stick to it. (Benjamin, Holland)

For some participants, demanding routines leave them feeling they have no life:

> Sometimes the days with him are particularly intense since the morning I wash, dress, prepare his food and feed him. I have to keep up with him 24 hours to 24. I cannot think of me anymore, it's impossible (Adriana, Italy)

> I cancelled my life for her. (Carla, Italy)

> It's very tightly scheduled…Everything is built on routines. Life is only about professional appointments. (Inge, Sweden)

The deadening effect of a life which has become limited to caring routines appears to have severe consequences for parents. They feel lonely, isolated and unable to cope. In Hanne's case, the move to an adapted house meant a loss of support from friends:

> I've lost all my friends too because they used to drop by on their bikes during school hours. And I'm not going to cycle 14 kilometers for a cup of coffee, that's too far. So that contact gets weaker because I can no longer leave the house so I don't go there anymore and they don't come to me so that has had a huge impact on my life…I've never been this lonely. (Hanne, Holland)

For a small number of participants, the sense of isolation is enforced because the disease is not talked about or even remains a secret:

> I must honestly admit that I'm not coping with the situation, have no one with whom to discuss my problems and in all honesty I wouldn't like to tell anyone about it. No one in my family knows about my son's illness. (Irena, Poland)

In addition, feelings of guilt and anxiety were not uncommon. For example:

> There is my youngest son who just cannot accept her; he avoids the daily routine of our family, isolates himself from us. It makes me feel guilty and inadequate I could not raise my children by finding the right tools to live with this disease (Carla, Italy)

Carla believes she has failed to find the "right tools" to live daily life with a child with JHD. Isolation is experienced both within the family and from the outside world.

For Berta the burden is constant:

> The anxiety I constantly feel never subsides, it's firmly rooted. When I am with her the anxiety goes away but I am constantly reminded of the disease. But as soon as we are separated, as soon as she is in school and I am at work, the anxiety is unrelenting. (Berta, Sweden)

She also finds that her emotions and moods fluctuate and this affects her family:

> I get annoyed and sad and angry and I try not to be but I can't help it sometimes. It really is up and down the whole time. There are times when you sleep poorly and just think too much. There are times when everything is going well. (Berta, Sweden)

Some participants practiced suppression to help them manage daily life whilst others were more stoical:

I just think you've got two halves in your brain; you hide everything away in one and carry on thinking with the other (Bernadette, Holland)

> It could be better for sure but I try to cope with difficulties by myself. I try not to complain about anything. We try to live normally with his disease like normal people. (Joanna, Poland)

Most participants described how their lives are lived in the present and how the future is not contemplated:

> We don't have any future plans so we live in the present. And then in a rather typical way I think that life around us ceases to a great extent. Many people withdraw. There are not so many friends and all that. (Inge, Sweden)

Not being able to make future plans means one is placed outside the social world in which families and friends share hopes and aspirations.

Other peoples knowledge and understanding: "It feels like no one wants to touch us"

Participants expressed concern and frustration at a lack of knowledge and understanding on the part of many healthcare and other professionals:

> She [doctor] didn't even see one person with HD. So when there is a case why do they pretend to be smart? I know it's me who's expert here. That's what I think, she doesn't know anything. (Kazia, Poland)

> I was saying but what is it? Something has to be there and they aren't able to understand this illness. (Adriana, Italy)

Often, parents concluded that they knew more than doctors who have to "consult their books". Kazia hints that some doctors are reluctant to admit they do not know what is wrong, a feeling which is echoed by Berta:

> No one knew anything about Huntington's at the Department of Pediatric Neurology. They had never heard of it and had never encountered any child with the illness. So going there and saying you suspected your child had it made you extremely suspect. They may have believed it was all my imagination. (Berta, Sweden)

In addition to a lack of knowledge, participants talked about poor communication and exchange of ideas between the various professionals involved:

> No one really knows what anyone else had done or not done. Everybody in the hospital is given something to do but they all work separately. (Bernadette, Holland)

Benjamin's frustration with those involved in supporting his child is evident when he talks of the difficulties getting financial support to make changes to their house:

> Again that ignorance, like do you know what that disease means? The child will become handicapped. She'll become completely demented, she won't be able to walk, she won't be able to eat, she'll turn into a vegetable. Do they expect me to carry her upstairs, such a big girl? (Benjamin, Holland)

Lack of knowledge and awareness about what the disease entails from involved professionals increases the burden on families.

Furthermore, participants drew attention to the lack of awareness and understanding from families, friends as well as wider society. For example, Benjamin described the need to justify his actions to other parents:

> Over and over again, I came to hate that in the end. I was like, if you all know better, you can borrow the child for a week. Then you'll know what you're dealing with. Oh, I got so angry towards the end, yes I might go crazy. You just become so crabby because you have to justify your actions towards others while she's our child. (Benjamin, Holland)

In contrast to other parents and friends 'knowing best', Bernadette noted how people have pulled away from her family which she explains as fear, which is likely to be a consequence of not knowing anything about JHD:

> Everyone starts pulling away from you too because they're also afraid. So at some stage you're not alone but you've created an island. And people row away from it because they don't want to hear it either…so you get more and more isolated obviously. (Bernadette, Holland)

Inge captures powerfully the isolation and aloneness these participants feel:

> It is so misunderstood in society, yes, not only in society, also within the health care system. It feels like no one wants to touch us really. (Inge, Sweden)

In sum, participants often believed they knew more than the healthcare professionals they were seeking help from. Moreover, they felt a lack of understanding from families and friends consolidating their sense of being alone.

Need for support: "Someone who knows, who really knows about it"

Participants talked about how much onus is on parents to find out what is available and what sort of help the family can expect:

> It has been incredibly hard…you need to find out so much by yourself…Everything takes such a long time. (Inge, Sweden)

Similarly, Benjamin stressed the need for support not to take so long to get in place:

> The child is going to die, she is going to die. So give her that space (house extension) and the opportunity to enjoy it while she's still got her wits about her…not take so much time, come on. (Benjamin, Holland)

The daily routines of care, the hospital appointments and so on consume so much time but more has to be 'found' by parents' themselves.

Most participants were clear about what would help them: a more integrated approach with good communication channels between the professionals and themselves:

> That there was someone who was coordinating everyone. There are so many things, Huntington's is a complicated story and progressive, that there was someone who had an overview all the time. There are so many strings to pull, so many people involved. (Inge, Sweden)

Hanne echoed the need for coordinated support with a contact person who would inform them about support available. She described how it was not clear how financial support could be used:

> So now we've hired a friend through the personal budget, he comes over two afternoons a week. The child has the afternoon of her life. Then you think, we should have thought of that, years ago, that things like that are possible. (Hanne, Holland)

An important priority is respite care. Parents want respite care for themselves and their families but need to know that the child with JHD is being cared for properly.

> A camp or something like that with professionals who understand her disease and whom she can feel safe. (Berta, Sweden)

And some parents wanted their child to be offered activities to reduce their isolation:

> Support of a psychological nature for my son or to propose activities for him rather than spend the whole day doing nothing. (Teodora, Italy)

> Some organizations that could arrange and run some occupation therapies during the day. (Kazia, Poland)

Parents wished to be able to talk to someone who understood what their life was like:

> A buddy system. I myself would like to have someone coming around regularly. Someone who knows about HD, who really knows about it. And who talks to you about what happens, who listens to you and who also has practical solutions. Because you search until you're blue in the face…[Someone] that you can hand over care for a while, you see? Like going out shopping by yourself for an afternoon. And that there's someone then who knows what it's like and who can deal with it and take care of him. (Saskia, Holland)

Thus, we see the overwhelming sense of isolation experienced by parents who feel that there is much room for improvement in terms of the support they are offered.

Discussion

In this discussion we include a comparison of these findings with those from the UK study and we emphasize the striking convergence between the various countries.

Almost all the participants in the current study described knowing something was wrong with their child, what we have called a felt sense[18], and which increased over time. Initially, awareness was hazy and difficult to articulate and parents sometimes felt doctors thought they were imagining things. This growing awareness is clear in the UK study as well as in qualitative studies of other childhood illnesses[19,20]. Parents observed changes in their children, most notably motor disorder, declining performance at school and shifts in cognitive and affective functioning. These observations and descriptions support and add flesh to the taxonomy of features developed by Nance and the US Huntington Disease Genetic Testing Group[21].

Receiving a diagnosis of JHD disrupts parents' expectations and hopes for their affected child and for the family as a whole. Often parents in this study used dramatic metaphors to convey how their lives had been derailed and changed irrevocably. Having a child with JHD meant that life was lived with no hope because there is no cure. Moreover, life became increasingly isolated for both child and carer. The UK study proposed that isolation is a dynamic and two-way process which arises out of particular forms of social interaction between the child/family and other people around them such as family and friends. In this study, parents perceived their children becoming outsiders, increasingly unable to interact with their peers.

Lack of knowledge and understanding was one of the most striking convergences across the countries including the UK. Parents believed this added to the challenges they faced and they frequently felt angry and frustrated. Parents also believed there is poor communication between the various professionals involved with their child and family and were frustrated by how long it took to get various forms of support in place.

In addition to good communication and a greater knowledge and awareness of JHD, parents were clear that respite care and contact with families in similar situations was a priority. This appears important for these participants because there was little evidence of informal support from family and friends in their accounts. They attributed this to fear as well as their own withdrawal. We believe participants wanted to normalize their experiences and have regular contact with other JHD families. Saskia's idea of a buddy system in which mutually beneficial support structures are established between families seems a good one. It is likely to be easier to implement in regions where population density is greater. Online support avoids this obstacle and some participants mentioned how having a computer for this purpose would enhance their lives, especially if it was supplemented with occasional "live" meetings. Understandably, parents seem to want a "one-stop-shop" where they can find out what resources and forms of support are available to them, ask for advice and so on.

Our findings extend the current literature on family coping with chronic or life-threatening childhood disease, with implications for diagnosis and longer-term adjustment. Parents are often the first to be aware of signs and symptoms in the child including changes in behaviour before a diagnosis is achieved[22] suggesting that delays in diagnosis of rare conditions can be minimized where doctors acknowledge parents' unique insights into their child's behaviour.

As reported previously, chronic disease restricts activities and participation in social activities[23]. This can be aggravated where treatment results in compromised immune function as in cancer where restrictions result from fear of infection[24] as well as in JHD where mothers discussed the demanding nature of caring for the child and associated feelings of isolation. Other family members are also affected including healthy siblings. Typically, parents become experts in the child's condition. They come to know more than medical staff about this rare disease and have to advocate on their child's behalf[22] Finally as in other conditions, families emphasise the need for coordinated

services perhaps led by a keyworker with overall picture of the child's needs and services involved[25]. Parents also talked about the need for respite care. Camps for children with chronic conditions have been reported and appear to be associated with some success in terms o therapeutic outcomes[26].

A set of first-hand accounts of the families' perspectives on JHD has been published[27, 28], but these accounts have not been subjected to thematic analysis. Nonetheless, some of the themes reported here can be discerned in those accounts together with aspects which gave hope and/or fulfillment despite the losses. Attempts at offering support to families could draw on these accounts of positive coping strategies.

The apparent gender bias in this study can be explained on the basis that the majority (approximately 75%) of cases of JHD the transmitting parent is the father[29] In future research it would be useful to try to recruit families with good support networks as again this may help offer suggestions for helpful interventions. It would also be interesting though challenging to design a study to examine systemic issues in the broader family context where members of different generation are each suffering from a form of HD. While it is useful to consider future research plans it is also important to note that JHD is a rare condition and we were fortunate to be able to interview the number of parents we did.

At present, there is no evidence base for specific interventions in JHD but HD research is being organized on an international basis[30, 31] which will facilitate opportunities to increase the sample size for both qualitative and quantitative work and generate discussion of good practice.[32] There is now greater awareness of JHD; for example, the patients' organization in England and Wales employs a care adviser with a specific remit for JHD within that role. A weekend for young people with JHD and their families is organized on an annual basis in the UK which provides families with a chance to get peer support and to help normalize their experiences. It may be possible

that the experience gained here can subsequently be disseminated through the European Huntington's Disease Network (and in particular the EHDN JHD working group) to HD organizations in other countries.

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Conflict of Interest Statement

No author has a conflict of interest.

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Table 1: Participants in the study

No	Participant Pseudonym	Country	Affected child
1	Mother (Berta)	Sweden	Daughter Aged 15
2	Mother (Inge)	Sweden	Daughter Aged 17
3	Father (Benjamin)	Holland	Daughter Aged 12
4	Mother (Saskia)	Holland	Son Aged 24
5	Mother (Bernadette)	Holland	Son Aged 9
6	Mother (Hanne)	Holland	Daughter Aged 17
7	Mother (Adriana)	Italy	Son Aged 13
8	Mother (Leonia)	Italy	Daughter Aged 17
9	Mother Rosa)	Italy	Son Aged 17
10	Mother (Teodora)	Italy	Son Aged 20
11	Mother (Carla)	Italy	Daughter Aged 20
12	Mother (Irena)	Poland	Son Aged 16
13	Mother (Kazia)	Poland	Son Aged 22
14	Mother (Joanna)	Poland	Sons Aged 9& 10