Parents' Experiences of Children's Healthcare for Hypermobile Ehlers-Danlos Syndrome and Hypermobility Spectrum Disorders

Lauren Bell[a] and Dr Gemma Pearce[a]

[a]School of Psychological, Social and Behavioural Sciences, Priory Street, Coventry University, CV1 5FB, United Kingdom.

Corresponding Author: Dr Gemma Pearce

Email: Gemma.Pearce@coventry.ac.uk

Authors Twitter accounts: @LMBell_ @GemmaSPearce @hEDStogether

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Abstract

Hypermobile Ehlers-Danlos syndrome (hEDS) and hypermobility spectrum disorders (HSD) are underdiagnosed hereditary connective tissue disorders requiring health care across specialties. Using mixed methods, we explored how parents have experienced children's health care for hEDS/HSD. Surveyed parents (N = 297) reported varying experiences, though professional understanding was negatively appraised by most parents. Themes identified from interviews (n = 13) were: (1) awareness and understanding are fundamental, (2) the importance of the therapeutic relationship, (3) limitations of health care systems, and (4) diagnostic labels are meaningful. Findings suggest that achieving person-centered care may broadly improve health care for families with hEDS/HSD.

Keywords: Ehlers-Danlos syndrome, Hypermobility Spectrum Disorders, Family-Centered Care, Health Care, Parent Professional Collaboration

The Ehlers-Danlos syndromes (EDS) are a group of hereditary connective tissue disorders thought to be related to a collagen synthesis defect (Malfait et al., 2017). Most types of EDS are rare and diagnosed using genetic testing (Malfait et al., 2017), however 80 to 90% of individuals with EDS have the remaining hypermobile type (hEDS), where any specific genetic etiology has not yet been identified (Tinkle et al., 2017). In 2017, three clinical criteria were proposed to indicate a hEDS diagnosis: generalized joint hypermobility (criterion 1), manifestations of generalized connective tissue disorder, family history of EDS, and/or musculoskeletal complications (criterion 2), and exclusion of alternative diagnoses (criterion 3). For individuals not meeting all criteria, a group of conditions named hypermobility spectrum disorders (HSD) was devised (Castori et al., 2017). HSD share symptoms and treatments with hEDS (Peterson et al., 2018) and both span equivalent levels of potential symptom severity (Smith, 2017). Before 2017, diagnoses in this spectrum were termed joint hypermobility syndrome (JHS), EDS Type III, and EDS hypermobility type, and reference to hEDS/HSD in this article encompasses these previous terms.

Reflective of hEDS/HSD as systemic diseases, individuals may experience various symptoms including joint hypermobility, chronic pain, recurrent dislocations, fatigue, gastrointestinal symptoms, anxiety, migraine, and frequently comorbid conditions including postural orthostatic tachycardia syndrome (POTS) and mast cell activation syndrome (Tinkle et al., 2017). Understanding as to the prevalence of hEDS/HSD is incomplete, though population estimations have increased from 1 in 5,000 and being considered rare, to more recent proposals that these conditions are more common (Demmler et al., 2019; Hakim & Grahame, 2014). Though diagnoses of hEDS/HSD have increased in recent decades, it has been estimated that 95% of individuals are undiagnosed (Grahame, 2008). Females were also found to be diagnosed on average 8.5 years later than males, and 72% of males and 41% of females were diagnosed in childhood (Demmler et al., 2019). Importantly, delayed diagnosis

can hinder symptom management and prevention (Demmler et al., 2019), and reported causes of delay have included insufficient awareness and understanding of hEDS/HSD, including among health care professionals (HCPs), and variable presentations of symptoms (Martin, 2019).

Moreover, there may be additional challenges for diagnosing children, who have increased hypermobility and subcutaneous fat than adults (De Baets et al., 2017; Malek et al., 2021), and limited guidelines exist for managing hEDS/HSD in pediatric populations (Engelbert et al., 2017). Children with hEDS/HSD have reported pain, fatigue, decreased physical activity, poorer sleep quality, reduced postural control, and reduced quality of life compared to peers (Mu et al., 2019; Muriello et al., 2018; Scheper et al., 2017) and hormone fluctuation during puberty can exacerbate symptoms (Hugon-Rodin et al., 2016). Diagnosis has been purported to facilitate access to health care, symptom management, and beneficial assistive devices for children (Engelbert et al., 2017; Palmer et al., 2016), however it has also been suggested that some HCPs are reluctant to diagnose hEDS/HSD in children (Grahame, 2017). In light of these unique considerations, it is crucial to understand how families experience, and can be best supported by, health care services.

Although some research has explored how adults with hEDS/HSD have experienced health care, less is known about how families have experienced children's health care. Adults have reported long journeys to diagnosis, dismissive attitudes from HCPs, misattribution of symptoms to psychological explanations, and inadequate professional awareness of hEDS/HSD which delayed care and impacted psychosocial wellbeing (Bennett et al., 2019a, 2019b; Berglund et al., 2010; Clark & Knight, 2017; Knight, 2015; Terry et al., 2015). Childbirth and maternity research has further evidenced that an informed and multidisciplinary approach to care can be essential for mothers and babies affected by hEDS/HSD, yet where understanding about these conditions among HCPs were indicated to

require improvement (Pezaro et al., 2018, 2020).

Parents are an integral part of their children's health care, and occupy roles in decision-making, communicating with health professionals, delivering care, and supporting self-management (Aarthun & Akerjordet, 2014). Family-centered care also recognizes and integrates the important role of families in pediatric health care, ensuring parent-professional collaboration, information sharing, and family-support (Kokorelias et al., 2019). In the United Kingdom (UK), research has found that families of children with an undiagnosed condition have described health as requiring improvements to care-coordination, awareness and advocacy, and that parents are emotionally impacted while managing uncertainty (Aldiss et al., 2021; Oulton et al., 2021). Similarly, research globally has identified that access to health services and information provision could be improved (Brannon et al., 2021; Kiernan et al., 2020). Understanding the meaning and impact of health care experiences is therefore crucial to improve family-centered care. It is further recognized that different health conditions are associated with unique health care considerations, as outlined above, and this study considered families living with hEDS/HSD.

While research has identified common factors important to children's health care globally, health care structures and systems also differ across countries, and this research focused on families in the UK. The core health system, the National Health Service (NHS), provides free at-the-point-of-access health care to UK residents. Unless requiring urgent or emergency care, most patients will first access primary care (e.g., General Practitioners [GPs]), who can then refer, as gatekeepers, into secondary care (e.g., for tests or consultations with specialist professionals such as rheumatologists). Patients can also pay for private health care at primary and secondary level, with an estimated 3% of GP consultations conducted privately (The King's Fund, 2014).

Aims

To explore families' health care experiences, this study included parents or primary caregivers whose children had been diagnosed with hEDS/HSD or whose children were currently suspected to have hEDS/HSD. This decision meant that families with meaningful health care experiences were not excluded, given that these families were still accessing children's health care and that hEDS/HSD are underdiagnosed (Demmler et al., 2019). Parents were also recruited to this study rather than children, as they were suitably placed to recall and evaluate children's health care in the context of family life and parents are an integral part of their children's health care (Kokorelias et al., 2019).

This mixed-methods study therefore aimed to explore how parents in the UK have experienced health care for their children with suspected or diagnosed hEDS/HSD, including previous terms of JHS, EDS Type III, and EDS hypermobile type. An online survey aimed to evaluate participants' experiences towards diagnosis and their appraisals of health care and different types of health professionals. Purposefully sampled follow-up interviews, with a maximum variation approach, then aimed to explore the depth, nature, context, and impact of these health care experiences on families.

Method

Participants

Eligibility criteria were that participants were the parent or primary caregiver of at least one child, aged 0 to 16 years at the time of participation, with suspected or diagnosed hEDS/HSD. Diagnosis was self-reported by participants, and was not medically verified, as this study aimed to explore how families had experienced health care where hEDS/HSD was an existing or potential diagnosis, with both circumstances likely to contribute meaningful experiences. A hEDS/HSD could therefore be ‘‘suspected’’ by parents and/or health care

professionals. Participants were over 18 years old, living in the UK, and recruited online via social media and hEDS/HSD-related charities and online support groups. Ethical approval was received from a UK University ethics board (P79018) and participants gave written online informed consent prior to the survey and again before any interview. No compensation was provided for participation.

Design

In this mixed-methods study, an online survey was completed by a larger sample of participants, some of whom participated in a follow-up interview. Participants completed the online survey between February and May 2019, and purposeful sampling was used to invite some interested parents to be interviewed to identify shared patterns across broad experiences (see Palinkas et al, 2015). This sampling process aimed for maximum variation, rather than solely extreme cases. The process was managed with, but not determined by, statistical software, and the following variables informed the process: health care experiences (positive, negative and neutral or mixed appraisals), diagnosis, UK region, child age, and relationship to the child. Appraisals of health care experiences and diagnosis were weighted more heavily in sampling, and this was balanced with aiming to recruit from all UK regions, and with a range of family characteristics.

Interviewed participants chose between telephone, online (video) call, online text-only messenger, and face to face interviews conducted between March and June 2019. This choice increased accessibility and participants could engage how they felt most comfortable, an ethical consideration which can also improve the validity of collected data (Pearce et al., 2014). While mixed-methods were adopted to explore the research aims, the qualitative methodology was recognized as contributing more substantially to the research aims to explore depth in families' accounts. In this mixed-methods approach, the quantitative results

were integrated complementarily to understand how experiences are reported among a larger sample of families (Creswell & Plano Clark, 2018).

Materials

Online Survey

Participants reported demographics (age, ethnicity, UK region, relationship to the child, and child age) and their child's diagnosis or suspected diagnosis. Parents of children suspected to have hEDS/HSD reported the child's age when symptoms began, selected reasons hEDS/ HSD was suspected, and reported any reasons a diagnosis had not yet occurred. If applicable, parents reported their child's age when health care was first accessed and which HCPs they had accessed. Parents of diagnosed children stated their child's age at diagnosis, the length of time between first accessing health care and diagnosis, and factors which led to diagnosis. These parents rated the diagnostic process from 1 (*very negative*) to 10 (*very positive*). Parents were asked, ''which are the key factors which have influenced your healthcare experiences as positive (*negative*)? Tick all that apply,'' with eight listed factors (e.g., ''*access to diagnostic procedures*'' and ''*none/not applicable''*), and then appraised their experiences with different professionals (e.g., ''*General Practitioners''*) from 1 (*very negative*) to 10 (*very positive*). Parents repeated the questions for any additional children and provided contact details if they wished to be interviewed.

Follow-up interviews

The first author conducted semi-structured interviews which were audio-recorded, transcribed verbatim, and anonymised. Parents were asked about symptoms which affected their child(ren) and their experiences with health care, including experiences towards and following diagnosis, and what families wanted from future health care. Parents were also asked what support had been or would be beneficial, and what they would advise to other

parents and HCPs. The semi-structured schedule is included in the Supplementary material, alongside examples of follow-up questions asked to gain clarity or explore further details about, or the impact of, experiences.

Analytical Plan

Survey data informed interview sampling and were analyzed descriptively using SPSS.

Interview data were analyzed using inductive, reflexive thematic analysis, as described by Braun and Clarke (2006, 2020), to analyze patterns in how parents had experienced children's health care. A critical realist perspective informed analysis, and parents were assumed to hold meaningful experiences and perspectives which could be influenced by the wider context and perceptions of others' experiences (Wiltshire, 2018). For reflexivity, the first author had limited knowledge of these conditions prior to this research. The second and corresponding author is a Chartered Psychologist who leads research in hEDS/HSD and related comorbidities. Both authors observed active online communities and read patient's experiences with hEDS/HSD. It was recognized prior to analysis that this awareness could have biased analysis. Developing familiarity with the collected data was an essential part of the analysis, and alternative interpretations of the data were considered to reduce bias.

In the first stage of analysis, the first author developed familiarization with the dataset through conducting, transcribing and re-reading interviews. Inductive semantic codes were generated for data related to the research question, and comparable codes across interviews were collated and assigned an overall code. These broader codes were grouped around common underlying concepts, and groups of codes were checked and modified to ensure consistency and relevance. Themes were then developed around distinct organizing group

features which represented the dataset. Data and development of these themes were discussed, deliberated, and revisited with the second author, and final themes were named and defined. Thematic analysis engaged a collaborative approach between authors, where candidate themes were discussed and revised throughout theme development and written analysis. Independent coding or inter-coder reliability was not relevant for this reflexive approach (Braun & Clarke, 2020). Quotes are presented to illustrate findings and have been modified to aid readability, such as removing hesitations.

Results

Participants

In total, 297 parents completed the online survey in *M* = 17 minutes and reported on 321 children. Of the survey respondents, 201 (67.7%) were interested in an interview, and 28 (13.9%) were invited for interviews. Thirteen participants (46.4%) responded and chose interviews via telephone (*n* = 8; *M* = 57 minutes), online video call (*n* = 3; *M* = 52 minutes), online call without video (*n* = 1; 47 minutes) and face to face (*n* = 1; 70 minutes). The participant characteristics of surveyed and interviewed parents, and their reported children, are shown in Table 1.

[Table 1 near here]

Online Survey

Accessing Health Care and Diagnosis

Children diagnosed with hEDS/HSD (*n* = 222) were aged *M* = 7.8 years (*SD* = 4.0 years, range 0 - 16 years) at the time of diagnosis. From when health care was first accessed, diagnosis was most often made within 6 months to 2 years (32.9%), and otherwise within 0 to 6 months (21.2%), 2 to 4 years (18.5%), 4 to 6 years (13.5%), 6 to 10 years (9.0%), or over

10 years (5.0%). Parents reported mixed experiences with diagnosis (*M* = 4.1, *SD* = 2.8), though 27.0% of parents scored 1 (*very negative*) and 5.4% scored 10 (*very positive*).

Parents reported that children with suspected hEDS/HSD (*n* = 99) displayed symptoms from aged *M* = 3.4 years (*SD* = 3.7 years, range 0 - 15 years), and initially accessed health care for these symptoms at age *M* = 5.6 years (*SD* = 4.4 years, range 0 - 16 years). To date children had been accessing health care for suspected hEDS/HSD for *M* = 4.3 years (*SD* = 3.5 years, range 0 - 14 years). Most often, hEDS/HSD was suspected due to relevant symptoms (*n* = 90), family history (*n* = 87), and comorbidities thought to be associated with hEDS/HSD (*n* = 21) such as POTS (*n* = 7) and autism (*n* = 4). HCPs who had discussed hEDS (*n* = 37) or HSD (*n* = 23) as possible diagnoses were also reported.

Health Care Appraisals

Parents of children with diagnosed and suspected hEDS/HSD reported positive and negative health care experiences (range 1 - 10) with all types of HCPs, except professionals accessed privately (*n* = 34) who were rated positively (range 7 – 10, *M* = 9.21, *SD* = 1.57). Where *n* > 30 participants reported about a type of professional, mean scores indicated experiences were neither positive nor negative for GPs (*n* = 263, *M* = 5.16, *SD* = 3.26), pediatricians (*n* = 127, *M* = 4.83, *SD* = 3.31), physiotherapists (*n* = 187, *M* = 6.02 , *SD* = 3.11), rheumatologists *(n* = 79, *M* = 4.41, *SD* = 3.20), orthopedic specialists (*n* = 39, *M* = 5.13 , *SD* = 3.40), and nurses (*n* = 35, *M* = 4.57, *SD* = 3.91), though occupational therapists were rated more positively (*n* = 62 , *M* = 7.77, *SD* = 2.42). As shown in Table 2, most parents appraised professional understanding as contributing negatively to their health care experiences (i.e. professional understanding was perceived to be inadequate). Similarly, more participants reported that their experiences with healthcare professionals were negative in terms of communication, ease of access to healthcare, and symptom management.

[Table 2 near here]

Qualitative Findings

As displayed in Table 3, findings were organized into four interrelated main themes, along with subthemes, which indicated elements which were fundamental to families' experiences of children's health care. While parents described varying experiences owing to the purposeful sampling approach, common patterns were identified relating to which aspects of health care were important to parents' experiences. Names of participants were changed for privacy and confidentiality reasons. Quotes are accompanied by pseudonyms (e.g., ''Beth'') and whether this parent was referring to a child with diagnosed or suspected hEDS/HSD. To illustrate nuances, data supporting each theme are included in the supplementary material.

[Table 3 near here]

1) Awareness and Understanding are Fundamental.

This theme presents how parents explained the importance of HCPs being aware and understanding of hEDS/HSD to avoid inappropriate, delayed, or unsupportive health care. The identified subthemes illustrate the variety and nature of understanding about the conditions across professionals and parents.

''Deeply variable'' professional understanding. Parents had experienced and perceived a range of awareness and understanding of hEDS/HSD among HCPs. One parent summarized professional understanding as the following, ''deeply variable. From 'haven't heard of it, that's not a thing', to 'yes, I've got that'.'' (Beth, child suspected to have hEDS). While some parents reported knowledgeable HCPs, including HCPs who lived with hEDS/HSD

themselves, this experience was often contextualized as being uncommon. These parents felt they were lucky with their experiences of children's health care, perceptions based on their own experiences of health care, or what they had heard from other families.

> We've talked about moving up north and the reason we're not is that her current medical team are so good…we're not going to risk taking her away from a team that work together, they understand each other, they understand EDS. (Kelly, child diagnosed with EDS type III/hypermobility type).

Specific misunderstandings among some HCPs were about the diagnostic criteria, failing to consider hEDS/HSD as systemic conditions, and underestimating the impact of hEDS/HSD on children. Some parents considered that the health care system was responsible for inadequate or inaccurate knowledge about hEDS/HSD, and identified that education about the range of symptoms and comorbidities would improve care, ''they're not being taught about it in medical school, you know, in more detail, because obviously it's connective tissue and it affects not just your joints. I think people think it affects just the joints and it doesn't'' (Mel, child diagnosed with hEDS).

The need for clear care guidelines. Parents wanted information and clear guidelines and treatment pathways to be available to HCPs and parents managing hEDS/HSD. Though parents recognized that further research was required to consolidate understanding, there were concerns that incorrect advice from HCPs or other sources could be detrimental to their child's health. One parent expressed wanting trusted sources of information, ''somewhere where it's openly available and it's a medically checked one, because the trouble is if you just rely on things like Facebook groups…it could really go wrong'' (Kelly, child diagnosed with EDS type III/hypermobile type).

A lack of clear care pathways meant some children experienced frequent delays and

referrals, particularly when HCPs wrongly perceived that hEDS/HSD was a diagnosis which could only be given by specific professionals, ‘‘the same pediatric rheumatologist [said] that he couldn’t diagnose Ehlers-Danlos and that she’d need to see a geneticist…so we got then put on a waiting list’’ (Hannah, child diagnosed with hEDS).

Honest and proactive health care professionals are valued. Some parents noted that HCPs could not be experts in all conditions, and it was important that HCPs were prepared to develop their professional understanding. Parents preferred HCPs who were honest about the limitations of their knowledge, particularly given the variable symptoms of hEDS/HSD, and felt let down by HCPs who did not aim to develop professionally. Parents also indicated frustrations when HCPs could, but did not, develop their knowledge, because that is what families had done.

> Whenever you go and see sort of a new GP it’s always, ‘oh I’ve not got that much understanding, I’ll look into it’. So, I’ll make a point of going back to the same GP next time, only to find they actually haven’t looked into it and they still don’t know. (Fran, child diagnosed with EDS Type III/hypermobility type).

***Parents become experts and researchers*.** When professional understanding was inadequate, parents described needing to become an expert in their child’s condition. As a result, parents described feeling alone and unsupported by health care and undertook their own research online or through engaging with other parents. Because of this, parents often perceived knowledge to be held in the hEDS/HSD community, who had developed self-management advice through lived experience, more so than among HCPs, ‘‘it’s almost that EDS is like this big secret and it’s left to the patients to figure it all out by themselves’’ (Alice, child suspected to have HSD).

Many parents were frustrated by the need to play the role of the health professional,

which sometimes conflicted with their parental role. Parents described that research could be tiring and time-consuming, and, as illustrated below, the challenges of researching hEDS/HSD could impact on work and family life.

> It's unhealthy as a parent to be researching constantly. I think I have become kind of obsessed with it, but I've had to, because there hasn't been anyone else to do it. I think it affects my relationship with my daughter. (Hannah, child diagnosed with hEDS).

These experiences further exemplified that parents felt unsupported by some HCPs, and that experiences could be improved with greater awareness and professional understanding of hEDS/HSD.

2) The Importance of the Therapeutic Relationship

The nature of the therapeutic relationship between families and HCPs contributed substantially to parents' evaluations of their health care experiences. The four organizing subthemes identified the importance and variable nature of therapeutic relationships, and how these relationships can impact upon future physical and social engagement with health care.

***Respect and collaboration*.** Central to how parents appraised health care was the extent of respect and collaboration in the therapeutic relationship. Many parents had perceived disrespect or disinterest from HCPs, and described frustrations that HCPs failed to collaborate with families who held the lived experience of these conditions, ''I usually end up having arguments with them, because I know more up to date stuff than they do, so I'll correct them, and they don't like that'' (Alice, child suspected to have HSD).

As inferred above, some parents perceived HCPs to be ''defensive'' (Hannah and Leah) rather than collaborative in their child's health care. However, one parent outlined a positive example of collaborative practice, where the unique experiences and contributions of the parent, child, and professional were each respected and consolidated to strengthen the

child's care plan.

> That physio was brilliant, she listened not only to me but to [my child] as well. And so, the three of us like a team came up with things that could work, and we figured it out… each of us used our knowledge. [My child] used her knowledge of her body, I used my knowledge… as a person with EDS and a mum, and she used her physiotherapy and mum knowledge. (Grace, child diagnosed with hEDS).

Accusations and dismissal of symptoms. Some parents had experienced accusations of ''doctor shopping'', fabricating and inducing illness, or referrals to children's social care after attending health care for their children's symptoms or requesting information or medical tests. Families experienced lasting emotional trauma beyond when families were cleared of these accusations, ''they put us under child protection…I was crushed'' (Mel, child diagnosed with hEDS). Some mothers had also experienced HCPs who dismissed their concerns as psychiatric, with underlying gender bias, ''they treat you like you're mentally ill or neurotic, especially if you're a woman'' (Isobel, child suspected to have HSD).

One father also noted an unusual reaction to him suggesting connections between his son's symptoms, where the father's research into symptoms had been dismissed.

> I started reading about the association between GI disorders and hypermobility and I kind of posited this to them and they were very dismissive of it. Sort of accused me of being a bit fixated on it, even though I'd mentioned it once or twice. (Edd, child diagnosed with HSD).

It was further perceived by several parents that some HCPs held negative biases or assumptions about hEDS/HSD, which delayed diagnosis and appropriate care. One perception was that some HCPs could be dismissive towards even the existence of hEDS/HSD, ''the worst one is when they just don't seem to believe it is a thing'' (Beth, child

suspected to have hEDS). As exemplified here, disbelief as to the existence of hEDS/HSD was a substantial and confusing frustration among parents who perceived a lack of support from HCPs.

Parents are advocates. In response to dismissive professional attitudes, parents often described health care as a ‘‘battle’’, a word used by five participants, and needing to push for appropriate care, ‘‘if you’ve got a difficult condition, then unless your parent acts as your really, really strong advocate, I don’t think you’ll ever actually get treatment’’ (Josie, child diagnosed with hEDS). As indicated here, parents often developed negative expectations about their children’s health care following negative experiences. Some parents also developed negative expectations from their own experiences living with hEDS/HSD, ‘‘it’s probably made me why I fight so much, ‘cause I know what I missed *[sic]*’’ (Fran, child diagnosed with EDS type III/hypermobility type).

Several parents were determined to avoid their children experiencing health care as negatively as they had, and approached health care determined to challenge any disbelief or lack of understanding for their children.

Disengaging from health care. Because of damaged therapeutic relationships and a lack of trust, several participants described disengaging with health care due to fear of further accusations. Josie, whose child is diagnosed with hEDS, described that health care could be a ‘‘waste of time’’ due to perceptions of a lack of understanding and proactivity among HCPs. Some parents identified that their children had also developed negative attitudes towards health care, as illustrated here, ‘‘she refused to go to casualty because she doesn’t want to see any more doctors’’(Hannah, child diagnosed with hEDS). Hence, therapeutic relationships which lacked trust, collaboration, and respect meant some families chose to avoid engaging with health care for symptom management.

3) *Limitations of Health Care Systems*

The structures, processes, and accessibility of the health care system impacted how effectively children received diagnosis, treatment, and access to specialists. Organized around three aspects of the system, this theme describes the importance of connected health care, restrictions within systems, and the comparisons with private care.

***Connective tissues need connected systems*.** Owing to hEDS/HSD being systemic conditions where professional understanding can also be inadequate, children often experienced frequent referrals across services. Many parents experienced this process to be disconnected and parents often coordinated their child's health care where information sharing across specialties was inadequate.

> I think there are assumptions made across the system about who knows what and who will share what with who. And we haven't got that right. And it is very frustrating from a family point of view when you think of people who are charged with aspects of your child's care and support, who don't talk to each other and don't share information with each other. (Debby, child suspected to have hEDS).

A lack of holistic oversight for children's health care could also negatively impact on schooling and family life, ''last week we had five appointments for all at the same hospital, all on different days. They could try and tie things up or realize what other interventions are taking place, then it could be a much smoother service'' (Fran, child diagnosed with EDS Type III/Hypermobility type). As illustrated here, ineffective collaboration across specialisms was often perceived to be particularly inadequate for systemic conditions like hEDS/HSD, with a broad range of involved specialties.

Restrictions on health care. Several parents described insufficient availability of knowledgeable specialists and ongoing access to treatments like pain relief, hydrotherapy,

and physiotherapy in public health care. One issue was a lack of children's services, together with frustrations that HCPs who could support with symptoms were inaccessible due to the separation of HCPs in adult and child services, ''there's all these children not being able to be treated [for bladder issues] just because of their age'' (Josie, child diagnosed with hEDS).

The health care structure also meant that children could not access prompt care to alleviate symptoms and were repeatedly delayed by the referral process, ''you can go on for ages and ages and ages and then all of a sudden something can happen that means you need physio immediately. But then you have to go through the whole referral process'' (Beth, child suspected to have hEDS). In this way, parents felt that hEDS/HSD were not treated as long-term conditions requiring ongoing health care, and where symptoms can arise unexpectedly. Though some parents identified these issues coming ''down to money'' (Fran, child diagnosed with EDS type III/hypermobility type) it was disappointing that beneficial treatments were available for a limited number of sessions and could not be accessed consistently without repeated referral.

Going private. Issues in the public health care system resulted in many parents employing private health care to ''move things quicker'' (Kelly, child diagnosed with EDS type III/hypermobility type) including towards diagnosis. Some parents recruited private health care due to delays, unavailability, or a lack of continuity with publicly-available treatment, ''it's hard to access the right treatment, except if you go privately'' (Charlie, child diagnosed with hEDS).

However, some parents reported that HCPs refuted private diagnoses, perceiving HCPs considered private diagnoses more attainable and therefore less valid, ''I've also seen obviously a lot of the controversy with a lot of the people in the NHS not accepting his [a private] diagnosis of EDS'' (Josie, child diagnosed with hEDS). Further frustrations related

to the costs of private care. Some parents perceived inequalities in access to treatment due to the reliance on private care, indicating the need for available and ongoing treatment and knowledgeable specialists in public health care, ‘‘privately you get a much better standard of care. But it’s rather, how do I put it, unfortunate, that it’s only because I can afford to get that access and most people can’t. It’s not really great is it’’ (Charlie, child diagnosed with hEDS).

4) Diagnostic Labels are Meaningful

The processes towards and outcomes of a diagnosis were important parts of most parents’ experiences. This theme describes three core features including families’ challenging journeys to diagnosis, that diagnosis is a starting point, and that frustrations and limitations can arise following a diagnosis.

Challenging journeys to diagnosis. Many parents described that their child’s diagnosis was delayed due to aversive attitudes among some HCPs about labelling children, or more specifically towards the diagnosis of hEDS/HSD. Some parents experienced HCPs who refused to diagnose hEDS/HSD. One parent expressed frustrations about the resistance her family had experienced towards diagnosing hEDS, ‘‘if a child has got diabetes or cancer or a leg amputated, you don’t not label it…The consultant said, ‘first things first, we don’t give diagnoses of Ehlers-Danlos’.’’ (Grace, child diagnosed with hEDS). These frustrations aligned with the perception among some parents that HCPs were reluctant to diagnose hEDS/HSD as they perceived them to be less valid diagnoses compared to conditions diagnosed through laboratory tests. As a result, reluctance to diagnose contributed to perceptions that hEDS/HSD were dismissed and disbelieved as conditions.

Moreover, some parents had experienced their children’s symptoms being labelled as psychological or psychiatric.

> All his physical symptoms got bundled up as a psychiatric problem…He [child] said, 'I'm not stressed, I'm not, I just have this pain', and they didn't believe him…That's just been such a blight on him getting the correct treatment ever since. (Edd, child diagnosed with HSD).

The implications of HCPs assigning psychological associations were that children were delayed access to diagnosis and treatments which could reduce non-psychological symptoms. Though parents recognized that children could experience comorbid anxiety and depression, it was important that HCPs understood that the physical symptoms of hEDS/HSD could then impact upon mental health.

Diagnosis provides an answer and a starting point. Several parents described relief and other psychological benefits to a diagnosis which provided an answer. As exemplified below, diagnosis could support a more reassuring interpretation of symptoms, and some families felt comforted that one diagnosis explained diverse symptoms. As a result, parents felt more informed about their child's symptoms and optimistic about future health care, particularly when families had previously experienced dismissal, ''does a name really help? No, but it does change attitudes. And I think from a medical point of view having a diagnosis of certain conditions gives them a firm foundation to build upon'' (Leah, child suspected to have hEDS). Similarly, some parents of children suspected to have hEDS/HSD expected diagnosis to facilitate access to health care, ''we want to get him diagnosed and get his care sorted so there's less chance that he's gonna develop a more significant illness like I experience'' (Isobel, child suspected to have HSD). Based on their own experiences, this parent appraised health care as beneficial to prevent worsening of symptoms, and perceived that health care was inaccessible without a diagnosis.

Frustrations following diagnosis. Though some parents of diagnosed children agreed

that diagnosis facilitated easier access to treatment, others identified that the benefits were limited due to insufficient access to treatment outside of private care. Some parents also identified disadvantages to diagnosis. One disadvantage related to the specific label given and related assumptions, including perceptions that diagnoses of HSD were subject to a lower hierarchy than diagnoses of hEDS, ‘‘this whole renaming could have been handled in a way that didn’t leave those of us who didn’t fit the hEDS criteria fully feeling like second class patients’’ (Isobel, child suspected to have HSD). In identifying unintended consequences of the reclassification, some parents perceived that HSD was less supported than hEDS, due to erroneous assumptions that HSD reflects a less severe diagnosis, less requiring of treatment. Though parents recognized benefits in distinguishing patients who fulfil different criteria for research purposes, frustrations resulted from the specific labels chosen, and a lack of education about what these labels signified.

Discussion

This study aimed to explore how families in the UK have experienced children’s health care for hEDS/HSD, and quantitative and qualitative findings reported the meaning of diverse appraisals of health care. Important factors which were important to health care across parents were professional awareness and understanding of hEDS/HSD, relationships with HCPs, the connectedness and accessibility of health care systems, and the processes and impact of diagnosis. These factors reflect existing guidance for person-centered care, where optimal health care involves compassion, respect, co-ordination, shared-decision making, and is user-focused at individual and systemic levels to benefit patients and health services (De Silva, 2014). Taking these issues together, parents in this study advocated for improving person-centered and family-centered children’s health care for hEDS/HSD.

Most surveyed parents reported that insufficient understanding of hEDS/HSD

contributed negatively to their health care experiences, consistent with existing research which has reported insufficient understanding of these conditions in adult health care (Bennett et al., 2019b; Palmer et al., 2016). Though some parents had experienced knowledgeable HCPs, insufficient understanding also delayed diagnosis and treatment access for some children. Approximately half of parents with diagnosed children reported a diagnosis within 2 years of first accessing health care, shorter than previous reports of a medium time of 16 years for women and 4 years for men (Kole & Faurisson, 2009). Though this difference may reflect improvements in the diagnostic processes, or that diagnoses are quicker for children, additional considerations may mean that the results of this study underestimate diagnostic times. First, some families had accessed private health care, and diagnostic times may not represent the process in public health care. Second, by definition this time excluded children not yet diagnosed, who had so far accessed health care for an average of 4 years. Third, parents unaware about the possibility of hEDS/HSD causing their child's symptoms would not have participated in this study. It should also be recognized that currently there are insufficient pediatric guidelines for these conditions, related to insufficient evidence about managing symptoms in children (British Society for Rheumatology, 2020). These challenges, and related professional tensions, may therefore have also contributed to parents' different experiences with health care and diagnosis.

Many interviewed parents were diagnosed themselves and had advocated for diagnosis. Parents drew on their own experiences and research to teach professionals about hEDS/HSD and share resources such as the EDS Toolkit (Reinhold et al., 2019). Many interviewed parents also reported that a diagnosis of hEDS provided validation and relief, with a unifying explanation for diverse symptoms. Though diagnosis can provide explanation, legitimization, and a social identity (Jutel, 2010), diagnosis must categorize patients without stigma or harm (Clarke & Iphofen, 2005). Some parents in this study

perceived a hierarchy of diagnosis, specifically where HSD could be misunderstood by others as less severe than hEDS. This finding further signifies the need for education around hEDS/HSD and consideration for the psychosocial impact of diagnostic labels.

While some parents had experienced interested and respectful HCPs, many parents reported fractured relationships with health care. Consistent with adults who felt stigmatized and discredited (Berglund et al., 2010; Clark & Knight, 2017), some parents had experienced HCPs who believed hEDS/HSD were not valid diagnoses. When HCPs hold limited knowledge of a condition, as has been reported for hEDS/HSD, doubt about the etiology, diagnosis, or treatment of a condition can lead to medical uncertainty (Han et al., 2011). Though diagnostic uncertainties are inevitable in medicine, HCPs can respond to uncertainty in numerous ways (Alam et al., 2017), and one possibility is that some parents in this study experienced HCPs who had responded to medical uncertainty with psychiatric explanations or disbelief, rather than compassionate suspension of judgement (Jutel, 2010; Kennedy, 2013). Illness stigma and medical knowledge can be socially constructed (Conrad & Barker, 2010), and assumptions about hEDS/HSD from HCPs may have also restricted collaboration with parents. Furthermore, parents who experienced dismissive health care often developed negative expectations towards future health care, and subsequently approached health care assertively in order to support their child. In turn, it is possible that HCPs responded with dismissal or even accusations, reflecting how individual experiences construct the social health care context (Lian & Robson, 2019).

This study further highlighted that health care systems must support patients and professionals through high quality information and coordination of health care across time and specialties. The ease of accessing care was more often experienced negatively among surveyed parents, and some parents reported that treatments and care pathways were unclear and inadequate for the systemic and chronic nature of hEDS/HSD, consistent with reports by

adults (Bennett et al., 2019b; Palmer et al., 2016). The impact for some children included challenges accessing appropriate specialists, diagnosis, and symptom management, and some families consequently disengaged from public health care. These findings further support the importance of person-centered health care, in line with health care guidance (Health and Social Care Act, 2012; National Health Service England, 2017) and other research which has indicated the importance of accessible, coordinated, and family-centered children's health care (Kiernan et al., 2020).

An additional finding of this study was that many interviewed parents perceived private health care to be necessary for diagnosis or ongoing treatment. More surveyed parents also reported that financial factors contributed negatively towards their experiences rather than positively. These findings were surprising in a UK context, where much health care is free at the point of access and may indicate that some families recruited private health care for their child's health, rather than this being an existing and affordable aspect of family life. Taken together, these findings further suggested that public health care could benefit from access to diagnosis, knowledgeable HCPs, and appropriate treatments for these children, to reduce any potential implication for social inequalities in access to health care.

Potential Strengths and Limitations

Though findings of this study were not assumed to reflect the experiences of all families with hEDS/HSD, limitations in the recruitment approach are recognized. The purposeful sampling approach intended to identify the meaning of a broad range of experiences to understand how positive and negative experiences can inform recommendations for practice. However, these qualitative findings should not be extrapolated to represent the distribution of experiences among the broader population of families with hEDS/HSD. Moreover, due to time limitations, interviews were not conducted after closing

the survey, where the widest breadth of experiences could have been identified. However, parents with diverse experiences reported common factors which were important towards family-centered healthcare. The qualitatively-weighted mixed-methods approach was also advantageous to richly explore parents' health care experiences (Shneerson & Gale, 2015), and may guide future research priorities and practice developments.

Among a larger sample, the survey findings indicated how different factors were appraised as positively or negatively contributing towards their health care experiences, including factors such as understanding among health professionals. Participants were not obliged to answer about each factor, however, and so there are limitations for interpreting these findings. It is not clear how far participants who did not report a factor as positive or negative considered this factor to be neutral, irrelevant, or simply did not report about the factor. The findings indicate that further research examining these variables in more depth is warranted to develop this understanding.

Parents were also recruited through social media and hEDS/HSD charities, and it is possible that participants were more likely to engage in these online communities following unsupportive health care experiences. It is noted that parents who appraised health care positively often deemed their experiences to be unique among families with hEDS/HSD. These perceptions of others' experiences may have been gained from online communities or engaging with other families, and while conclusions cannot be generalized to all families, these findings do suggest that children's health care for hEDS/HSD warrants improvements. On the other hand, it is also recognized that parents' negative appraisals of health care in this research may have been strengthened by an awareness of others' experiences and may reflect collective frustration towards health care.

Implications for Research and Practice

Future research may consider exploring the nature and construction of health professional's understanding and attitudes towards hEDS/HSD, which could identify areas for professional training or support. In addition, parents often became knowledgeable of hEDS/HSD through their own research and lived experience, and situated their child's health in the context of family life. Further research may greater explore how parents being diagnosed with hEDS/HSD themselves impacts upon diagnosis and health care interactions within children's health care. Developments in clinical practice, such as self-management guidelines or changes to diagnostic procedures, may therefore benefit from involvement of families and professionals (Filipe et al., 2017). This process, termed co-production or co-creation, recognizes that HCPs, patients, and families have their own expertise, and that collaboration can support cost-effective, ethical, practical, and beneficial health care developments (Filipe et al., 2017). Similarly, future beneficial research may directly examine how children with hEDS/HSD experience and appraise their health care to further understand how family and person-centered care can be achieved. The survey results also indicated that more detailed understanding among a large sample of families is warranted and may usefully examine certain key factors in more detail. For example, as more parents with diagnosed hEDS/HSD reported about finances, it could be hypothesized that private/financial factors were more salient in the experiences of these parents. Further research may usefully examine the role of obtaining private healthcare to diagnose hEDS/HSD in the UK and possible associations with health inequalities. Though parents recognized that some issues require further medical research, systemic changes, or financial investment, other areas such as the therapeutic relationship and awareness and understanding of hEDS/HSD were perceived to be issues which could be improved through education, training, and person-centered care.

Conclusion

This study reports the meaning of diverse experiences of health care for suspected and

diagnosed hEDS/HSD in the UK. Overall, however, findings indicated that children's health care requires improvements via increased awareness and understanding of hEDS/HSD, positive therapeutic relationships, a more connected and accessible health care system, and diagnoses associated with health and psychosocial benefits. Building on this study, further research could explore the nature and construction of health professional's knowledge and attitudes towards hEDS/HSD, the experiences of children with these conditions, and understand how families and professionals can be better supported. Clinical practice which utilizes the knowledge and lived experience of families could also be beneficial to strengthen person-centered and family-centered health care for children with hEDS/HSD.

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Declaration of Interest

The authors declare no conflict of interest.

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Tables

Table 1

Characteristics of surveyed and interviewed parents and their children

Characteristic	Surveys		Interviews	
	N	%	*N*	%
Parents				
Female	290	97.7	11	84.6
Male	7	2.4	2	15.4
Age				
20-29 years	18	6.1	0	0.0
30-39 years	86	30.0	0	0.0
40-49 years	154	51.9	10	76.9
50-59 years	39	13.1	3	23.1
Ethnicity				
White	289	97.3	12	92.3
Ethnic Minority	7	2.4	1	7.7
Not reported	1	0.3	0	0.0
Region				
North of England	57	19.2	2	15.4
Midlands	69	23.2	4	30.8
South of England	142	47.8	5	38.5
Scotland	16	5.4	1	7.7
Wales	9	3.0	1	7.7
Northern Ireland	4	1.3	0	0

Children				
Age				
Under 5 years	25	8.4	1	7.7
5 – 11 years	149	50.2	3	23.1
12 – 16 years	147	49.5	9	69.2
Diagnosis				
hEDS	60	18.7	5	38.5
HSD	31	9.7	1	7.7
JHS	71	22.1	0	0[a]
EDS Type III or hypermobility type	60	18.7	2	15.4
Suspected condition				
hEDS	79	21.8	3	23.1
HSD	29	9.0	2	15.4

Note. hEDS = hypermobile Ehlers-Danlos syndrome; HSD = hypermobility spectrum disorder; JHS = joint hypermobility syndrome; EDS = Ehlers-Danlos syndrome

Table 2

Percentage of Parents Who Reported Each Variable as Positive or Negative to Their Child's Health Care

Variable	Suspected hEDS/HSD		Diagnosed hEDS/HSD	
	Positive (%)	Negative (%)	Positive (%)	Negative (%)
Ease of accessing health care	13.10	33.33	20.72	46.85
[a]Access to diagnostic procedures/ [b]Length of diagnosis	6.06	48.48	23.42	57.66
Communication with health professionals	10.10	43.43	27.48	66.22
Treatment or symptom management	8.08	44.44	15.32	72.07
Financial	5.05	7.07	13.06	24.32
Location of health care services	13.13	14.14	21.62	34.20
Understanding of EDS/HSD among health professionals	3.03	68.69	13.96	77.48
None / Not Applicable	42.42	2.02	38.29	5.41

Note. As parents did not have to respond about each variable, percentages do not total 100. hEDS = hypermobile Ehlers-Danlos syndrome; HSD = hypermobility spectrum disorder.

[a] Question to parents of children suspected to have hEDS/HSD. [b] Question to parents of children diagnosed with hEDS/HSD.

Table 3

List of Themes and Subthemes

Theme	Subtheme
Awareness and understanding are fundamental	
	‘‘Deeply variable’’ professional understanding
	The need for clear care guidelines
	Honest and proactive health care professionals are valued
	Parents become experts and researchers
The importance of the therapeutic relationship	
	Respect and collaboration
	Accusations and dismissal of symptoms
	Parents are advocates

	Disengaging from health care
Limitations of health care systems	
	Connective tissues need connected systems
	Restrictions on health care
	Going private
Diagnostic labels are meaningful	
	Challenging journeys to diagnosis
	Diagnosis provides an answer and a starting point
	Frustrations following diagnosis