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## Qualitative Research

# Living with joint hypermobility syndrome: patient experiences of diagnosis, referral and self-care

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### Abstract

**Background.** Musculoskeletal problems are common reasons for seeking primary health care. It has been suggested that many people with ‘everyday’ non-inflammatory musculoskeletal problems may have undiagnosed joint hypermobility syndrome (JHS), a complex multi-systemic condition. JHS is characterized by joint laxity, pain, fatigue and a wide range of other symptoms. Physiotherapy is usually the preferred treatment option for JHS, although diagnosis can be difficult. The lived experience of those with JHS requires investigation.

**Objective.** The aim of the study was to examine patients’ lived experience of JHS, their views and experiences of JHS diagnosis and management.

**Methods.** Focus groups in four locations in the UK were convened, involving 25 participants with a prior diagnosis of JHS. The focus groups were audio recorded, fully transcribed and analysed using the constant comparative method to inductively derive a thematic account of the data.

**Results.** Pain, fatigue, proprioception difficulties and repeated cycles of injury were among the most challenging features of living with JHS. Participants perceived a lack of awareness of JHS from health professionals and more widely in society and described how diagnosis and access to appropriate health-care services was often slow and convoluted. Education for patients and health professionals was considered to be essential.

**Conclusions.** Timely diagnosis, raising awareness and access to health professionals who understand JHS may be particularly instrumental in helping to ameliorate symptoms and help patients to self-manage their condition. Physiotherapists and other health professionals should receive training to provide biopsychosocial support for people with this condition.

**Key words:** Benign hypermobility syndrome, diagnosis, Ehlers–Danlos syndrome, hypermobility type, life experiences, referral, self-management.

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## Introduction

Musculoskeletal problems are common reasons for seeking primary health care (1). Joint hypermobility syndrome (JHS) is a hereditary connective tissue disorder, characterized by musculoskeletal pain and an excessive range of motion in joints (2). As there are no laboratory tests to indicate JHS (3), it is usually subjectively assessed using the Brighton criteria (4), which include the Beighton score (5) for joint hypermobility. It has been reported that symptomatic joint hypermobility affects around 5% of women and 0.6% of men (6). The prevalence of JHS amongst those attending rheumatology and physiotherapy clinics has been estimated to be between 30% and 60% and is higher in non-Caucasian populations (7,8). However, the diverse and fluctuating symptoms associated with JHS may easily be attributed to other causes and the true prevalence of JHS may be much higher than previously estimated. It has been suggested that many patients presenting with painful non-inflammatory musculoskeletal problems may have unrecognized JHS (9).

Although most individuals exhibiting joint hypermobility do not experience problems, a diagnosis of JHS may be given when symptoms such as arthralgia, proprioception difficulties, fatigue, soft tissue injury and joint instability are observed in the absence of genetic markers to indicate disorders such as osteogenesis imperfecta or Marfan syndrome (10). JHS, osteogenesis imperfecta, Marfan syndrome and Ehlers–Danlos syndrome share many symptoms and many experts now consider JHS to be indistinguishable from Ehlers–Danlos syndrome, hypermobility type (11). In this article, the term JHS will be used.

Primary-care practitioners are usually the patients' first point of contact on entering the health-care system. They can do much to assist individuals to effectively self-manage their condition (12) and refer patients for appropriate secondary care such as physiotherapy, although the effectiveness of physiotherapy has yet to be established due to the lack of high-quality research in this area (13). The lived experience of JHS from patients' perspectives has received little attention. The aim of the current investigation is to examine patients' lived experience of JHS, their views and experiences of JHS diagnosis and management.

## Methods

Four focus groups were conducted between January and February 2013 in four locations in the UK. Participants were recruited via physiotherapy services at two National Health Service (NHS) trusts, local members of the Hypermobility Syndromes Association (HMSA) and patients who had previously expressed an interest in assisting with research activity at two University locations. Eligible participants were aged 18 years or over, had previously received a diagnosis of JHS (including Ehlers–Danlos syndrome, hypermobility type), had attended physiotherapy within the preceding 12 months and were able to speak English.

Individuals with other known musculoskeletal pathology causing pain, particularly osteoarthritis and inflammatory musculoskeletal disease such as rheumatoid arthritis, were excluded. The purposive sampling strategy aimed for diversity with regard to age, gender, socio-economic situation and geographical location to capture maximum variation in views and experiences. Ethical approval was obtained from the North East NHS Research Ethics Committee (12/NE/0307) and all participants gave written consent.

## Procedure

Data reported in this article were collected from focus groups with individuals with JHS, aimed at developing a physiotherapy

intervention for JHS management. Topic guides used to facilitate discussions covered issues of living with JHS, day-to-day self-management and provision of support for symptom management. In line with an inductive approach, topic guides were revised in the light of emerging findings. The focus groups were conducted in non-clinical settings, facilitated by two researchers (STP and JPH), and open-ended questioning techniques were used to elicit participants' own experiences and views. The focus groups lasted between 71 and 100 minutes.

## Data analysis

All focus groups were audio-recorded, fully transcribed, anonymized, checked for accuracy and imported into a qualitative software package (NVivo 10) to aid data analysis. Thematic analysis, using the constant comparison technique (14), was used to scrutinize the data to identify and analyse patterns across the dataset and the data were scrutinized for negative cases. Transcripts were examined on a line-by-line basis, with codes being assigned to segments of the data and an initial coding frame developed. An inductive approach was used to identify participants' perceptions of their experiences. RHT and JPH independently coded transcripts and any discrepancies were discussed to achieve a coding consensus and maximize rigour. The emergent themes were discussed by the multi-disciplinary research team to ensure credibility and confirmability. Scrutiny of the data showed that data saturation had been reached at the end of the analysis, such that no new themes were arising from the data (15).

## Results

Twenty-five individuals with JHS participated in the focus groups, aged 19–66 years (mean: 38.2 years), 22 were female and 23 were of 'White' ethnicity (Table 1). The analysis led to the development of four key interrelated themes: 'the impact of JHS', 'JHS as a poorly understood condition', 'receiving a diagnosis' and 'JHS management and self-care'. Verbatim extracts are provided to illustrate the findings.

### The impact of JHS

Participants described in detail the impact of JHS, which included fatigue, pain and proprioception problems:

...day in day out you're managing your pain and it's a lot of pain, it's a dull ache and it makes you sleepy and it makes you tired and you're exhausted (Female G, age 30, FG1).

...it's on your mind the whole time because I'm constantly thinking about where my hands and feet are (Female G, age 48, FG2).

Recurring joint dislocation and 'cycles' of injury and recovery were common and participants frequently talked about the need to modify or restrict behaviours and activities.

...it's just difficult to know how much to push yourself because then you are worried about injuring and then you're setting yourself back, it's a vicious cycle really (Female B, age 27, FG3).

However, participants acknowledged that the impact and consequences of these varied symptoms were different for each patient, with one participant noting, 'all of us are probably so different yet we're categorised as the same' (Female D, age 21, FG1). Thus, some participants found living with JHS symptoms to be 'very debilitating', (Male E, age 36, FG3), whilst others were determined to persevere with their chosen activities, in spite of their symptoms:

**Table 1.** Participants' characteristics and demographic information relating to the participants of the four focus groups, carried out between January and February 2013 (total  $n = 25$ )

Characteristic	Subcategory	$n$ (except where specified)
Age (years)	18–29	8
	30–39	7
	40–49	6
	50–59	2
	>60	3
	–	Mean 38.2, median 36 years
Gender	Female	22
	Male	3
Ethnicity	White	23
	Other	2 (both 'British White and Chinese')
Socio-economic status (SES) <sup>a</sup>	1 (least deprived)	8
	2	8
	3	4
	4	3
	5 (most deprived)	1
Education	Left school	3
	<16 years/schooling to 16 years	
	College diploma or equivalent	6
	University degree or equivalent	10
Employment	Postgraduate degree	6
	Employed full time	7
	Employed part time	8
	Student full time	4
	No paid job	5
	Retired	1

<sup>a</sup>SES was measured as Index of Multiple Deprivation (IMD) quintile from home postcode.

I teach like rock-climbing, surfing, body boarding and all of that stuff, like, and I'm not going to stop doing it because I'm in pain like you can't live your whole life with pain dictating what you can and can't do (Female G, age 45, FG3).

Although JHS is characterized by pain, participants alluded to the complexity of their pain experience, for example, describing difficulties in distinguishing between chronic and acute pain, and in recognizing how—or 'if'—injuries had occurred:

how do we know whether we've injured something? Because we've got pain all the time (Female C, age 40, FG1).

Participants also described how prior experiences of repeated injuries led to heightened levels of anxiety and catastrophizing about future injuries: 'I feel like I'm in a constant state of anxiety, waiting for the next injury and trying to pre-empt anything that's going to cause it' (Female G, age 48, FG2). Others extrapolated their current or prior experiences to an imagined future: 'Oh my god is this going to be like this for the next 60 years of my life?' (Female B, age 27, FG3).

#### 'JHS—a poorly understood condition'

Participants perceived JHS to be a condition that was poorly understood by health professionals, and within society more widely.

I think I was described as a biomechanical conundrum by one of the physiotherapists I saw [...] and this is what I found repeated over and over again, that hypermobility shouldn't be causing pain, it's just the way you are' (Female C, age 53, FG2).

Participants described feeling stigmatized and fraudulent:

...I'm really struggling, but because of people's expectations and their perceptions and you don't want to ruin that, you don't want people to start thinking "oh well, you know, [unclear 1.02.47], we don't employ people with disabilities because this is what happens" (Female C, age 40, FG1).

They think you're trying to cheat out of doing something ... (Female D, age 21, FG1).

and felt they had been 'blamed' for their symptoms:

when I was at school I just had to sit at the side while they were doing all the games, they sort of almost, I felt they were blaming it on me ... (Female D, age 32, FG3).

Participants reported how they felt they were 'the odd one out' (Female B, age 34, FG2) and tried to hide their experiences and appear 'normal':

'... it's so exhausting mentally and physically to try and appear to be normal and do normal things throughout the day with everybody and pretend it's alright' (Female G, age 48, FG2).

Participants felt that the unpredictable, diverse, evolving and fluctuating nature of their symptoms exacerbated others' misunderstanding of the nature of JHS and contributed to a lack of social support:

if you're inconsistent as well, they sort of go, 'she was alright with that last week, why is it this week she's saying that, you know, that's going to be difficult for her today' (Female C, age 53, FG2).

#### Receiving a diagnosis

Many participants reported lengthy diagnosis trajectories, and being treated for individual symptoms (e.g. pain) rather than JHS:

we've all been passed from pillar to post where people don't recognise it or they just attribute a pain to something else, when a snap kind of diagnosis just comes out of the air and you know, you progress from there (Female G, age 45, FG3).

Often, obtaining a correct diagnosis was a coincidental occurrence:

it was only because a locum happened to be in the day I went in because my GP was off sick, and he just started saying, well, to start bending everything... but if hadn't been for him I wouldn't have been put on the right track, ... because otherwise what other route do you really have if it's not through your GP? (Female B, age 27, FG3).

Receiving a diagnosis was considered necessary in order to access appropriate care pathways, for example, referral for physiotherapy for JHS rather than for an acute single joint problem:

I was originally seen by a physio who hadn't diagnosed with the hypermobility and then went back to a musculo-skeletal specialist who then put me forward to specialist hypermobility physiotherapist and since then it's been amazing; I feel like it's been worthwhile and it felt like the right thing to do and I've been really enjoying it (Female B, age 27, FG3).

In addition, a diagnosis helped to validate participants' experiences and was psychologically helpful:

Getting a diagnosis on paper, this is what's wrong with me? I mean that helped me hugely psychologically (Female A, age 60, FG2).

A diagnosis in itself, however, still did not guarantee beneficial treatment:

When I was first diagnosed, I wasn't, I really felt I wasn't given that much information about the condition [...] and it just seemed to be all exercises they'd given me at the time seemed to make me worse (Female B, age 27, FG3).

Similarly, a diagnosis did not necessarily lead to greater understanding and support from others in the participants' social networks:

There are people who don't feel it's a genuine diagnosis, that it's something psychological, that you just need to be a bit braver (Female A, age 21, FG2).

### 'JHS management and self-care'

#### Education

Education for health professionals was a key issue for participants, to facilitate timely diagnosis of, and referral for, JHS. Thus, education for health professionals was a prerequisite for diagnosis, and diagnosis was a prerequisite for participants to access education. Participants recognized that individuals with JHS also need education, in order to find ways to self-manage their condition and to understand and engage with prescribed treatments.

I suppose it's where someone who doesn't really know about it, they've got to learn about it first because you can't tell someone to do it [ie engage with a particular treatment] if they don't understand it (Female D, age 21, FG1).

Participants also felt that they could provide valuable information about the nature of JHS to clinicians:

I think actually it's the health professionals that need educating [...] there's lots of things I still need to know about hypermobility, but on the flip side I do think it's the health professionals that need to know more (Female G, age 42, FG3).

Participants described learning and understanding about JHS as a two-way process:

So I think I get a huge amount of enlightenment from her [the physiotherapist], and I lend her books and she lends me books about hypermobility and all that helps (Female D, age 54, FG2).

#### Identifying specific self-care activities

Participants accepted that treatment should aim to manage symptoms rather than provide a cure: 'it's all about them helping you to manage your pain, rather than cure it' (Female, age 44, FG1). Participants recognized the importance of self-care activities such as appropriate exercise, once a diagnosis had been received and a clear understanding of JHS had been developed. Participants also recognised that activity pacing 'is really key' to managing JHS symptoms (Female B, aged 32): 'I have this balancing act, if I do too much it all hurts, don't do enough, it all hurts, do it just right, I'm okay' (Male A, age 50, FG1).

Participants sought to deepen their understanding of JHS and access to health professionals who understood the nature of JHS in order to take a 'whole-body' approach to self-care over a life course:

the more you see the physio who knows what they're talking about ... the more you understand how the body works and the more you can apply your own sort of thinking to what you're doing in your exercises (Female E, age 44, FG1).

Yeah, it's got to be holistic, it really has to be (Female D, age 21, FG1).

Participants also felt that education about JHS should be provided at an early age, and therefore, early diagnosis was necessary:

I'm only 19 now but if you'd have said that when I was 16, I might not be in as much pain as I am now [...] so if like if they'd have told me more about how to treat it back then it probably wouldn't be as bad as it is now (Female F, age 19, FG3).

### Discussion and conclusions

Participants provided many insights into the lived experience of JHS. Pain, fatigue and repeated episodes of injury were particularly challenging features of the wide-ranging symptoms of JHS. Bury (16) describes how chronic illness brings about 'biographical disruption', and that the illness or disease requires the individual to make certain changes to their lives (17,18). In our study, participants' lives were disrupted repeatedly by fluctuating symptoms and 'cycles' of injury and recovery. Participants described the complex and individual nature of their pain experience and their responses to unpredictable symptoms. For some, previous episodes of pain or injury led to heightened levels of anxiety and catastrophizing about the future and their symptoms required them to modify or restrict their activities, while others refused to be 'dictated to' by their symptoms.

Participants also explained the psychosocial consequences of living with JHS. Patients perceived JHS to be often poorly understood by health professionals and those in their wider social environment and reported feeling fraudulent and blamed for their symptoms. Participants felt stigmatized, 'marked out as different (19)' and 'alien' to others in society and found it exhausting to try to 'appear normal'. Stigma can have wide-ranging negative biopsychosocial consequences including reduced participation in activities and an exacerbation of disability and disease, for example, through delayed diagnosis and treatment (20).

A lack of awareness of JHS amongst health professionals meant that obtaining a diagnosis of JHS was often difficult. Participants had often been misdiagnosed or treated for symptoms (e.g. pain) rather than the condition itself and 'passed from pillar to post' often until a serendipitous or coincidental diagnosis of JHS was made. Previous studies have reported similar findings (9) and others have emphasized that being understood and believed by health professionals and significant others, along with social support, is instrumental in long-term pain management by facilitating (or inhibiting) pain acceptance (21,22). Participants highlighted the importance of a correct diagnosis in facilitating access to appropriate health care, support and education and helping to validate participants' experiences. Having lived with problematic symptoms of JHS sometimes for long periods of time, it is possible that the receipt of a diagnosis represented the beginning of a process during which they were able to understand and make sense of their symptoms, obtain appropriate treatment and subsequently find ways to self-manage the condition. Participants' experiences resonate with Williams (23) who uses the term 'narrative re-construction' to describe how individuals with chronic illness re-establish order and meaning in their lives.

Primary-care practitioners play an important role in helping patients to understand and self-manage long-term health conditions. A prerequisite of being able to provide support for patients is that primary-care practitioners are able to recognize and diagnose JHS and to refer patients to JHS-trained specialists. Without a correct diagnosis, unsuitable treatments or information may be given, which may exacerbate symptoms (24). Currently, primary-care practitioners and other health professionals such as physiotherapists do not routinely receive training related to JHS (9) and the validity of diagnostic criteria (such as the Beighton score) has recently been questioned (25).

Education and access to information was important for participants to allow them to make informed health-care choices.

Participants described the value of collaborating with health professionals and reciprocal learning between practitioners and patients in order to develop effective self-care strategies, and for holistic long-term management. Recognizing that JHS would not be cured, participants felt that health professionals and those with JHS could potentially develop a deeper understanding of JHS management by learning from each other as a 'teachable dyad (26, p. 682)'.

### Strengths and limitations

To ensure validity, participants were recruited from four different geographical locations in the UK and had the experience of different health-care services. Participants provided rich personal narratives of the day-to-day experiences of living with and managing JHS from patients' perspectives and data analysis demonstrated general consensus and shared experiences. A limitation of this research is that participants were recruited through JHS support groups or via physiotherapy services for JHS and may therefore have been more 'active' or 'aware' of their condition than, for example, newly diagnosed individuals. The focus groups formed one phase of a study to develop a physiotherapy intervention to manage JHS. It was clear that the lived experience of those with JHS that emerged from the data analysis was an important 'story', which to date has received little attention. The issues raised in these focus groups highlight the need for more in-depth research in this area. Future research could conduct interviews to provide a more detailed investigation of personal accounts of living with JHS, in particular those newly diagnosed with JHS.

### Implications for practice

Individuals with JHS experience diverse, fluctuating and often debilitating symptoms and diagnosis is often slow. Without a correct diagnosis, treatment may exacerbate symptoms. Increased awareness of JHS in primary care could help improve the diagnosis and referral processes. Following diagnosis, access to JHS-trained health professionals could help patients to effectively manage their condition over a life course and receive psychological support when needed. Patients and practitioners may be able to learn from one another and so assist in developing a deeper understanding of a currently poorly understood condition.

### Declaration

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Ethical approval: this study received ethical approval from the North East NHS Research Ethics Committee (12/NE/0307).

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