Physiotherapy Pain Association

Exploring the experiences of diagnosis and physiotherapy management in patients with hypermobility spectrum disorders and shoulder instability: a focus group report.

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Abstract

Objective

To explore patients experiences of the diagnosis and management of hypermobility spectrum disorders (HSD)and shoulder instability.

Design

An exploratory qualitative design. A focus group was organised, transcribed, and analysed. The collected data was analysed using a thematic approach.

Setting

A single NHS Trust within the UK

Participants

Four people with HSD and shoulder instability

Results

All participants identified difficulties and delays in receiving a diagnosis for their HSD. They reported a lack of awareness amongst health care professionals, including physiotherapists. The participants described mixed experiences of physiotherapy treatment and alongside a perceived lack of awareness of HSD, they reported increased levels of anxiety prior to commencing treatment. Only one patient had received specialist shoulder physiotherapy for their shoulder instability resulting in a positive experience.

Conclusions

Physiotherapy is perceived by patients as an important component in the management of HSD and shoulder instability and should include appropriate exercise therapy and advice on joint management and on shoulder relocation. However, patients continue to report variable experiences of physiotherapy. Improving awareness and education amongst physiotherapists is essential for improving the experiences and long-term management of this patient group. Future work should explore what educational strategies would be optimal for health care professionals and which treatment strategies patients find the most beneficial.

Contributions of the paper

- Patients often experience difficulties in receiving a diagnosis for their HSD, resulting in delayed access to appropriate advice and treatment.
- Patient experiences improve once they are treated by health care professionals with knowledge of HSD and shoulder instability, further work is required to develop services.
- Reduced awareness and knowledge of HSD and shoulder instability amongst health care professionals' results in increased levels anxiety amongst patients when referred to physiotherapy.

Keywords

Hypermobility Spectrum Disorder, Ehlers Danlos Syndrome, physiotherapy, focus group, shoulder instability

Introduction

Hypermobility spectrum disorder (HSD) previously known as Joint Hypermobility Syndrome is a benign heritable connective tissue disorder affecting collagen production. This can result in excessive range of motion in joints, increased tissue laxity and fragile connective tissue. This can be present in the general population with no clinical symptoms [1]. Patients often seek medical advice when they develop musculoskeletal symptoms such as joint pain, repetitive joint subluxations and dislocations which can impact on

quality of life [2]. Further characteristics of HSD include non-musculoskeletal symptoms such as fatigue, chronic pain, and proprioceptive deficits [3,4]. Ehlers – Danlos Syndrome (EDS) is a spectrum of heritable connective tissue disorders. Ehlers-Danlos hypermobility type (hEDS) is diagnosed on a clinical basis with generalised joint hypermobility being the most prevalent clinical symptom [4]. As hEDS shares many similar characteristics to HSD, many experts now agree that they are indistinguishable from each other [5,6].

The prevalence of hypermobility spectrum disorder is estimated to be around 3% in the UK population. However due to a lack of understanding and difficulties with diagnosis the true prevalence is expected to be much higher [7]. Diagnosis of HSD is often one of exclusion and is based on clinical presentation. The Brighton criteria are often used alongside laboratory testing to rule out a systemic cause to symptoms [4]. The Beighton Score is calculated as a measure of general joint laxity and a score of four or more is considered indicative of increased joint laxity [8]. Patient's report seeing several healthcare professionals prior to receiving a diagnosis, resulting in feelings of frustration, and of being dismissed [9]. A survey by the European Organization for Rare Diseases showed that patients with EDS had the longest delay in diagnosis. Of the respondents with EDS 58% reported consulting with five physicians and 20% had seen over 20 before receiving a correct diagnosis. Of the respondents with EDS 50% took an average of 15 years to receive a diagnosis and a further 25% took 28 years. This is compared to other conditions such as cystic fibrosis and Duchennes Muscular Dystrophy. Fragile X syndrome was the next longest taking 50% of respondents 2.8 years to receive a diagnosis [10]. These reported difficulties and delays in diagnosis are further supported in literature exploring patients with HSD/hEDS experiences of their condition. This lack of awareness and misdiagnosis can result in psychological distress and patients receiving inadequate treatment or information [11-13].

Patients with HSD often report shoulder pain which is highly associated with shoulder instability. Shoulder instability can be defined as symptomatic abnormal motion of the shoulder. It can include a wide variety of symptoms including pain, apprehension, subluxations, and dislocations of the shoulder [14]. Instability can develop without a history of trauma and is associated with generalised soft tissue laxity [15]. Symptoms of shoulder instability can affect a patient's participation in hobbies and exercise as well as impacting on a person's ability to perform work-based tasks [1]. Previous literature has demonstrated a link between shoulder instability and hEDS and one study demonstrated that shoulder dislocation was often the first stage of the condition especially in those with hEDS [16]. Rombaut et al [4] found that 85.2% of hEDS individuals reported shoulder pain and 63% had a history of shoulder dislocations. Individuals with hypermobility were 2.5 times more likely to report a history of shoulder instability than those who did not [17]. Current literature has explored patient's experiences in the diagnosis and management of their HSD and experiences with physiotherapy. However, there is a lack of research exploring the experiences of diagnosis and physiotherapy management in patients with HSD who have shoulder instability [10,11].

The aims of this focus group were to explore patient's experiences of receiving a diagnosis for their HSD and the care management they have received. This included exploring their experiences around physiotherapy management for both their HSD and shoulder instability.

Methods

An exploratory qualitative study design using a focus group method was used to explore the diagnosis and treatment experiences of patients with a history of shoulder instability and HSD.

Participants

Inclusion criteria

Male and female patients over the age of 18, with a diagnosis of HSD and a history of shoulder pain and instability were eligible to participate in the focus group. Participants were required to have previously attended physiotherapy within secondary care and consented to taking part in the local patient advisory group.

Participant Selection

Patients who met the inclusion criteria and were currently receiving physiotherapy at one NHS site were invited to participate in the group. Members of the physiotherapy team identified suitable participants and gained permission to share their contact details with a member of the research team. The designated member of the research team contacted each patient to discuss the details of the focus group and took verbal consent to send further information; including a patient information sheet inviting them to participate. Six participants were invited to take part, of which five agreed to attend. One of these participants failed to attend the focus group resulting in four participants. As this focus group was conducted as part of South Tees NHS Foundation Trusts' Research and Development Departments approved Patient and Public Involvement and Engagement activity, No further ethical approval was required which was confirmed using the HRA decision tool.

Sampling

A purposive sample method was used to identify patients

with experience of HSD and shoulder instability who had attended for physiotherapy at the NHS hospital site.

Data collection

The focus group took place in the research and development department of South Tees NHS Foundation Trusts' in February 2020. An introduction was provided by the researchers to discuss the background and aims of the study and the participants were given the opportunity to ask any questions. All participants verbally consented to participating in the group. They were advised that their responses would be anonymised and there would be no patient identifiable information included in the research report. Open questions were utilised during the focus group to provide patients with the opportunity to discuss their experiences. A patient experience questionnaire was developed by the research team and was used to guide the direction of the discussion. This discussion was facilitated by a physiotherapist (study lead), patient and public information lead, and an orthopaedic surgeon. Topics explored within the group included: participants' experience of receiving a diagnosis for their HSD and the physiotherapy management they had experienced for their HSD and shoulder instability. Further topics explored included the participants' experience of the awareness of HSD and shoulder instability amongst health care professionals. All participants were assigned a letter as a pseudonym. Transcripts of the focus group were taken by an independent member and were reviewed by the lead researcher and patient and public lead. The analysis was performed by the lead researcher and the thematic analysis was reviewed by the orthopaedic consultant and patient and public lead who attended the group.

Results

Four individuals took part in the focus group. The group included three female and one male participant. All patients had a diagnosis of HSD/hEDS and had a history of shoulder pain/instability and previously attended physiotherapy.

Key Themes from Focus group

Five major themes were identified following analysis of the focus group.

Theme 1: Delays and difficulties in diagnosis

All patients described difficulties in receiving a diagnosis with onset of symptoms in childhood. They were treated for individual joint problems and two patients underwent hip and knee stabilisation surgery. Patient A described having problems with hips from birth and was treated as a 'lax, clumsy baby'. They suffered multiple joint dislocations before being diagnosed by their GP.

Patients often described having their symptoms dismissed or not taken seriously prior to receiving a diagnosis. Patient B was dismissed as suffering from 'growing pains' despite having patella dislocations from the age of 10 and felt the lack of diagnosis affected the management he received. 'I was advised not to exercise in childhood, but I now realise that this was not the best advice'

Patient D: 'it took 19 years for me to be diagnosed. I was classed as a problem teenager.'

Two patients received a diagnosis privately after seeing a number of health professionals. Patient C had symptoms from the age of 12 and despite being under a rheumatologist did not receive a diagnosis 'I paid privately to see a specialist in London to receive a diagnosis.'

Theme 2: Variable experiences of physiotherapy.

All patients had undertaken physiotherapy and had mixed experiences in terms of the treatment they had received, and the awareness therapists had of their condition.

Patients agreed that they had better experiences when treated by specialist physiotherapists for both their HSD and shoulder instability. Patient B described that 'the specialist physiotherapist would tailor the exercises to my individual needs, yet other physios would, even though they could see you were struggling, still expect all the exercises to go ahead'

Three out of four patients had received hydrotherapy as part of their physiotherapy management. Patient D felt that specialist hydrotherapy was now available and more beneficial.

Patient C also attended hydrotherapy. This was a generic group for musculoskeletal patients, the patient reported that the exercises were not tailored to their capabilities, and they were unable to complete many of the exercise 'I was expected to complete all exercises even though they were painful'

Patient A found a generic hydrotherapy group difficult 'Patients are expected to do repetitive exercises and are unable to comply with that request'

With regards to treatment for the shoulder one patient had treatment from a shoulder specialist with knowledge of HSD which he found very beneficial. A key component of this was 'the physio showed my wife how to put the joint (shoulder) back in place which made a big personal difference'

Patient C had 'physiotherapy for her shoulder which was learned from Youtube' after negative experiences in physiotherapy.

Patient C had a good experience when referred to occupational therapy. This included splints and advice on fatigue and pain management, which she felt was an often-overlooked part of the condition.

Theme 3 Anxiety and physiotherapy.

Patients reported that shoulder instability can cause anxiety, particularly if the shoulder was to come out of joint in certain situations. Patient D suffered a dislocation on a plane and had to travel to get home before being able to reduce the shoulder. All patients reported experiencing feelings of anxiety or depression related to their HSD

Due to previous experiences within physiotherapy and a perceived lack of awareness of their condition amongst physiotherapists, patients reported feelings of anxiety and apprehension when referred to commence a new course of physiotherapy.

One patient felt pushed by their previous physiotherapists and would not complete the exercises if they felt they would cause exacerbations of pain and not attend further appointments. If their physiotherapist had knowledge of hEDS then participants agreed they would feel more comfortable discussing the difficulties, they were experiencing with their exercises (Patient C).

Patient A went to new physiotherapy appointments with 'low expectations' due to an expected lack of awareness of their condition amongst therapists.

Theme 4 Lack of education and awareness

Although patients felt that awareness of HSD/hEDS has improved, they agreed more training should be provided to health care professionals, medical students and junior doctors. Patient D felt more education for A&E staff would be beneficial for when attending with dislocations and felt accessing specialist centres was difficult. Patient B explained that 'GPs needed a good awareness of what services were available for patients with hEDS.'

Patient C felt that there needed to be a greater understanding of the condition and that it is not only joints that are affected 'you wouldn't think that it affects internal organs but it does'. Patients also highlighted the importance of the social implications of HSD. This included the impact of pain, fatigue and the challenges this presents to daily activities and social interactions. They felt that this should form part of the treatment package to assist with long term self-management.

Theme 5 Care pathways

All participants felt that improving services and care pathways for patients with HSD was required and further research was essential. Patient B felt that 'it would be useful for them to have a direct contact for when they experience difficulties, which will assist with improving the services that patients receive.' The other participants agreed that ongoing access to support is not always available and there are delays accessing services, they highlighted the need for support towards self-management.

Discussion

The findings of this focus group support those of previous studies that suggest patients with HSD often experience delays in receiving a diagnosis for their condition. They are often seen by a number of health care professionals prior to receiving a diagnosis and symptoms frequently present in childhood (16). This can lead to feelings of frustration and anxiety, which may impact on a patient's quality of life. The five major themes that emerged from our patient focus group may help identify areas for quality improvement in the care pathways for patients with HSD and future research. Patients within the group found their experiences of treatment improved for both their HSD and shoulder instability, once they were seen by health care professionals with an increased understanding of their condition. Patients also identified a lack of long-term support for managing their HSD.

Physiotherapy including exercise is the primary treatment for patients with HSD however, patients' experiences in the physiotherapy management they have received can vary [18]. This is supported by the findings of this patient group. Patients within the group reported increased anxiety when initially attending physiotherapy, as the perceived level of clinicians' knowledge and awareness of HSD/hEDS was variable. They often felt that treatment programmes were not individualised to their abilities. Patients admitted that they found it more difficult to discuss any challenges or difficulties they were experiencing with their treatment programme if they felt their physiotherapist did not have sufficient knowledge and understanding of HSD. This had a negative impact on the therapeutic relationship often resulting in reduced compliance or failure to complete treatment. Effective communication and shared decision making between patients and therapists assists with developing an effective therapeutic relationship, this has been shown to improve clinical outcomes and enable patients to manage their long-term musculoskeletal pain more effectively [19,20].

At present, there is a lack of evidence for what treatment strategies are the most effective for patients with HSD [21]. A survey of physiotherapy practice found that the most common interventions included advice, education, self-management techniques, pacing and goal setting [22]. Patients within this group identified long term management as a key component of their care which has been highlighted as important to patients in previous research [3]. Reduced knowledge and understanding of HSD amongst health care professionals and delays in diagnosing HSD, can affect both the quality of the treatment received and patient's experiences of physiotherapy [3,21,22].

Previous study findings that patients' experiences improve once they work with physiotherapists who have specialised in treating HSD [3] are also supported in this study. A study exploring physiotherapists' knowledge of HSD showed that 51% of physiotherapists had received no training on hypermobility and this related to their confidence in assessing and managing HSD [23,3]. This suggests that more work is required to improve the education and awareness of HSD.

This study explored the experiences of patients with HSD in the management of their shoulder instability. All patients had symptoms of shoulder instability but only one of the participants had seen a specialist shoulder physiotherapist with regards to their shoulder instability and their experience improved as a result. Advice around relocating the shoulder independently was reported as an important component of management. These experiences can be explored in greater depth in further research using a larger group of participants to confirm the themes identified in this small exploratory study. The findings of this study support those of other studies that have explored patients experiences in the diagnosis and management of HSD [3,11,24]. Future research could focus on the types of treatments patients received and what interventions and strategies they found the most effective. This may assist in developing services and care pathways for this patient group. It would also be beneficial for developing training strategies amongst physiotherapists.

This was a small exploratory study using a limited number of participants that were selected from one NHS trust. All patients had attended physiotherapy but had not been treated by the lead researcher reducing the risk of bias. Due to the small sample size caution must be taken when drawing conclusions from the findings as they may not be transferable to other contexts. It is unlikely that data saturation was achieved due to the small number of participants. Repeating the study with multiple focus groups and more participants, would also allow the identification of any additional themes. This study provides the basis for developing a larger study with a greater number of participants and addressing some of the limitations of the study. This would include having two independent researchers analyse the transcripts, analysis by one researcher could affect the reliability of the findings. However, the findings are evidenced through quotes from the data collected. As only one patient had received treatment from a shoulder specialist for their shoulder instability symptoms, further research would be essential to explore this further. A further limitation of the study was that as patients' symptoms presented in childhood they were reporting on both past and more recent experiences, this raises the possibility of recall bias with regards to their diagnosis and treatment. All patients within the group reported feelings of anxiety and depression but this was not explored in further detail within this study. The psychosocial factors associated with HSD would be a potential area for further research in the future.

Conclusions

HSD/hEDS are complex conditions where care pathways are suboptimal and are not underpinned by robust evidence. Delays in diagnosis and variable management can result in high levels of frustration and anxiety amongst this patient group. Earlier diagnosis and increased awareness amongst health care professionals can improve long-term management. Although physiotherapy is the most widely accepted treatment, patient experiences are variable and there is currently a lack evidence for the most effective methods of treatment. These are exploratory findings from a small study, and they need to be confirmed from a larger study. There is also a need for further research exploring the best educational strategies for improving awareness amongst health care professionals and exploring patients experiences of physiotherapy interventions for HSD and shoulder instability.

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