Citizen Participation and Public Petitions Committee

14th Meeting, 2023 (Session 6), Wednesday 4 October 2023

PE2038: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders

Petitioner	The Ehlers-Danlos Support UK
Petition summary	Calling on the Scottish Parliament to urge the Scottish Government to commission suitable NHS services for those with hypermobile EDS (hEDS) and hypermobility spectrum disorders (HSD), and to consult with patients on their design and delivery.
Webpage	https://petitions.parliament.scot/petitions/PE2038

Introduction

1. This is a new petition that was lodged on 24 July 2023.

2. A full summary of this petition and its aims can be found at **Annexe A**.

3. A SPICe briefing has been prepared to inform the Committee's consideration of the petition and can be found at **Annexe B.**

4. Every petition collects signatures while it remains under consideration. At the time of writing, 1,989 signatures have been received on this petition.

5. The Committee seeks views from the Scottish Government on all new petitions before they are formally considered. A response has been received from the Scottish Government and is included at **Annexe C** of this paper.

6. A submission has been provided by the petitioner. This is included at **Annexe D**.

Action

The Committee is invited to consider what action it wishes to take on this petition.

Clerk to the Committee

Annexe A

PE2038: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders

Petitioner

The Ehlers-Danlos Support UK

Date Lodged:

24/07/23

Petition summary

Calling on the Scottish Parliament to urge the Scottish Government to commission suitable NHS services for those with hypermobile EDS (hEDS) and hypermobility spectrum disorders (HSD), and to consult with patients on their design and delivery.

Previous action

Volunteers and members from The Ehlers-Danlos Support UK have met with Bob Doris MSP as a result of interactions via the Rare, Genetic and Undiagnosed Conditions Cross Party Group. Constituents have met with Mairi McAllan MSP, Angela Constance MSP, Bill Kidd MSP, Pauline McNeill MSP, Emma Roddick MSP and Daniel Johnson MSP.

Motions on behalf of constituents have been raised in the chamber of the Scottish Parliament by Martin Whitfield MSP and Michelle Thomson MSP.

A meeting with Emma Roddick MSP on 4 November 2022 resulted in Ms Roddick publicly calling for a pain pathway for those with EDS and others. She also agreed to speak to then Cabinet Secretary for Health Humza Yousaf about the issues and to raise a question in the Parliament about a care pathway for people with EDS.

Background information

hEDS is a connective tissue disorder with body-wide symptoms which can be disabling, affecting all aspects of life. Symptoms include musculoskeletal problems, chronic pain and fatigue, gastrointestinal disturbance, pelvic and bladder problems, anxiety and more. There is no single test, which makes diagnosis challenging. HSD presents many of the same symptoms and shares the same diagnostic challenge. Together, hEDS and HSD are thought to affect 1 in 500 people.

Historically, those showing symptoms have been referred to local rheumatology departments where service access is known to be inconsistent.

While there is a commissioned diagnostic service in England for rarer types of EDS (which can be accessed by people in Scotland), diagnosis and management has relied on the personal interest and knowledge of a small number of clinicians rather than formally commissioned services.

The situation described has resulted in inequalities in access to healthcare, causing anxiety, distress, and unnecessary pain and suffering for those waiting for diagnosis, those on inappropriate treatment pathways, and their families.

Annexe B

Briefing for the Citizen Participation and Public Petitions Committee on petition PE2038: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders lodged by Volunteers and members from The Ehlers-Danlos Support UK

Brief overview of issues raised by the petition

The petitioning support group are calling on the Scottish Parliament to urge the Scottish Government to commission suitable NHS services for those with hypermobile EDS (hEDS) and hypermobility spectrum disorders (HSD), and to consult with patients on their design and delivery.

Hypermobile Ehlers-Danlos syndrome (EDS)

According to the NHS UK website, “EDS are a group of rare inherited conditions that affect connective tissues in the body.” These tissues are the ones that provide support in skin, tendons, ligaments, blood vessels, internal organs and bones.

There are different types of EDS that can share symptoms such as joint hypermobility (increased joint movement), stretchy or fragile skin. Sometimes the condition is mild, but for others, symptoms can be severe and disabling, and in rare cases life threatening.

The cause is a fault or faults in certain genes associated with connective tissue, and the faults can be inherited from either or both parents. Sometimes though, the fault can arise spontaneously in the person affected.

Hypermobile EDS or hEDS is the most common type of EDS (out of 13 types).

Range of symptoms of hEDS (from NHS UK website)

People with hEDS may have:

- joint hypermobility
- loose, unstable joints that dislocate easily
- joint pain and clicking joints
- extreme tiredness (fatigue)
- skin that bruises easily
- digestive problems, such as heartburn and constipation
- dizziness and an increased heart rate after standing up
- problems with internal organs, such as mitral valve problems or organ prolapse
- problems with bladder control (urinary incontinence).

Other hypermobility spectrum disorders

The petitioners refer to other hypermobility spectrum disorders or syndromes. There is an association, the Hypermobility Syndromes Association, which has more information about hypermobility. This site describes such syndromes as follows:

> "A hypermobile joint can bend beyond the typical range of movement. Many people are hypermobile (around 1 in 10) – and hypermobility is more common in women and children, and people of Afro-Caribbean and Asian descent. It is common in gymnasts, athletes, dancers and musicians. Many hypermobile people have no significant symptoms ('asymptomatic hypermobility').
>
> For a smaller number of people hypermobility is accompanied by symptoms (symptomatic hypermobility).
>
> Symptomatic hypermobility can be due to:
>
> A. Specific heritable disorders of connective tissue, like Ehlers-Danlos syndromes, Marfan syndrome, Stickler syndrome, osteogenesis imperfecta and others.

B. Joint shape, looser ligaments, or poor muscle tone (without a connective tissue disorder)

C. Other conditions (like Down's syndrome, Cerebral Palsy etc)

D. Injury or repeated stretching/training (for example in yoga enthusiasts and gymnasts)

We would say that people within groups A and B have **a hypermobility syndrome."**

It is important to note that joint hypermobility is relatively common (affecting around 1 in 30 people) and is unlikely to be caused by EDS. (NHS UK)

Diagnosis, treatment and management

There are no current tests to confirm whether someone has hEDS, and diagnosis is made based on medical history and physical examination. However, a GP might refer someone to a genetics service if one of the rare types of EDS is suspected, and other family members have symptoms.

There is no cure for hypermobility syndromes and standard treatment focuses on improving muscle strength and general fitness to better protect joints.

NHS UK outline current approaches to management of the condition:

"People with EDS may also benefit from support from a number of different healthcare professionals.

For example:

- a physiotherapist can teach you exercises to help strengthen your joints, avoid injuries and manage pain
- an occupational therapist can help you manage daily activities and give advice on equipment that may help you
- counselling and cognitive behavioural therapy (CBT) may be useful if you're struggling to cope with long-term pain
- for certain types of EDS, regular scans carried out in hospital can detect problems with internal organs

- genetic counselling can help you learn more about the cause of your condition, how it's inherited, and what the chances are of passing it on to your children".

NHS Inform, NHS Scotland's public facing website, covers treatment, focusing on self-care, physiotherapy, pain management and the options for some of the specific problems associated with the condition.

National guidance and guidelines

There are references to guidance and guidelines for managing the condition, but, according to the British Society for Rheumatology, the process for producing them has not been straightforward or without controversy. One reason cited is an insufficient evidence base, and that expert views vary widely on the best way to manage the conditions.

See also: Review: Ehlers-Danlos syndromes: state of the art on clinical practice guidelines - PMC (nih.gov)

The Ehlers-Danlos Society has formulated diagnostic criteria for doctors to use to aid diagnosis, but it is unclear whether it is used widely, or if it has been universally adopted.

The National Institute for Health and Care Excellence (NICE) also has some guidance for the referral for hypermobility in children. This is published as part of a Clinical Knowledge Summary for Hypermobility (Developmental rheumatology in children) in which EDS is referred to.

The British Medical Journal has published Best Practice guidance to aid diagnosis and treatment.

NHS services in the UK

This service specification for complex Ehlers Danlos (all ages) from NHS England suggests that there has been a specialist service, based in England "that is commissioned (by NHS England) for all eligible patients from England and Scotland", since 2013. The specification states that the service is provided by a consultant dermatologist in Sheffield Children's NHS Foundation Trust, North West London Hospitals NHS Trust and the John Radcliffe Hospital, Oxford.

The service provides diagnostic support for suspected complex EDS rather than ongoing treatment and support.

However, the specialist diagnostic service is not open to those with hypermobile EDS.

Ehlers-Danlos Support UK state that:

> "In 2017, the National Services Division in Scotland committed to producing a paper for regional directors of planning, highlighting which issues and service gaps people with EDS and HSD encounter. To our knowledge, this has not been written. In 2019, a proposal to the National Services Division for a specialist EDS centre for Scotland was rejected, despite almost unanimous support from healthcare professionals. A commitment was made instead to enhance regional expertise and services. This has not yet happened."

In February 2018, the Scottish Government published 'Rare Disease Scotland: Progress report', which was an update on work carried out against 51 commitments in the UK Strategy for Rare Diseases (2013). The report refers to EDS.

A petition was submitted to the UK Parliament, calling to 'Provide dedicated funding for diagnosis/treatment of hypermobile EDS and HSD'.

The UK Government responded to the petition on 5 July 2023, stating that:

> "There are no plans for a national service for diagnosis or treatment of hEDS and HSD. Our plans for musculoskeletal conditions will be outlined in the major conditions strategy."

Anne Jepson
Senior Researcher
15 August 2023

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Annexe C

Scottish Government submission of 6 September 2023

PE2038/A: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders

I refer you to your email of 24 July asking for the Scottish Government's views on the actions called for in petition PE2038. I have addressed the requests contained in the bullet points of the petition below.

- **Calling on the Scottish Parliament to urge the Scottish Government to commission suitable NHS services for those with hypermobile EDS (hEDS) and hypermobility spectrum disorders (HSD), and to consult with patients on their design and delivery.**

The Scottish Government is absolutely committed to ensuring that people living with a rare condition like hEDS and HSD are able to access the best possible care and support, and benefit from healthcare services that are safe, effective, and person-centred. It is the responsibility of the Scottish Government to set out policies which will tackle these issues, and it is expected all NHS Boards to provide high quality care accordingly. In general, it is the responsibility of NHS Boards to ensure sufficient workforce is in place to deliver safe services and the Scottish Government continue to support all NHS boards to ensure they do this.

Where specialist care and clinical expertise is available for rare conditions we are committed to making sure they are available to patients through the commissioning of specialist services. Diagnosis and patient care/symptom management for hEDS and HDS is provided by local and regional rheumatology services with the input of other specialities such as clinical genetics, chronic pain services and colorectal surgery as required. Where Scotland doesn't have such specialised treatment, there are options for patients to access the care they need, if available, elsewhere in the UK. The cross-border referral application would be made by the patient's secondary care specialist or consultant if they deem that the treatment required is not available in Scotland. The application is evaluated by NHS National Services

Division who then provide funding if the application shows that the referral is to access a proven, evidence-based, specialist intervention, that is not available elsewhere in NHS Scotland. If the person in question does not yet have a secondary care specialist or consultant involved in their care, then this will fall under the responsibility of their GP.

During the development of the Scottish Rare Disease Action Plan, which was published last December, we engaged with the rare condition community and listened to the issues they raised, such as those highlighted in the petition.

The Action Plan is structured around the four shared priorities of the UK Rare Diseases Framework:

- faster diagnosis
- raising awareness
- better co-ordination of care
- improved access to specialist care

The actions that have been set out in this first iteration are to be worked on over the next 18–24 months, in collaboration with Scotland's NHS and with third-sector partners such as Genetic Alliance UK and other key stakeholders and patient groups. The actions will address issues around lack of sign posting, referral pathways and overall co-ordination of care for people with rare conditions, including those living with hEDS and HDS.

Another vital component to this is continuing to work with the rare condition community as we implement the Action Plan, to ensure that those with lived experience can tell us the most about what needs to change to improve their lives. This will include people who have conditions such as hEDS and HSD.

We are committed to raising awareness and education of rare conditions, such as hEDS and HSD, across all health care professionals, including physiotherapists and orthopaedic units. There are over 7,000 rare conditions and it is not possible for healthcare professionals to receive comprehensive training on every condition. However, it is important to ensure an understanding of rare conditions are part of the curriculum for trainee healthcare professionals. We are also seeking to promote Continuing Professional Development linked to rare conditions which will be available throughout the career of a healthcare

professional. There will also be work around appropriate signposting to reliable sources of information in order to help healthcare professionals navigate the complex clinical guidance that exists for many rare conditions.

I hope this response is helpful both to the Committee and to the petitioner.

Planning and Quality Division

Annexe D

Petitioner submission of 20 September 2023

PE2038/B: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders

Hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders (hEDS/HSD) are not rare types of Ehlers-Danlos syndromes, they are just rarely diagnosed. Demmler et al 2019 found that 1 in 500 people had a diagnosis of hEDS/HSD.

In 2018 Professor Stuart Ralston (Rheumatologist) put in a bid for a specialist centre for the treatment and diagnosis of patients with hEDS / complex EDS. The aim being to establish a centre for the assessment, and management of patients whose needs are not being adequately met at a local level. The centre would accept referrals from secondary care in NHS Boards throughout Scotland, where it was felt that the patients' needs could not be adequately addressed locally. This was backed up by consultants, therapists, and patients but was not supported by Scottish Society for Rheumatology; they did not see a need for a specialist centre, as they believed the services were already available to patients. At this time there was also a commitment from National Services Division to produce a paper for regional directors of planning highlighting which issues and service gaps people with EDS and HSD encounter. To our knowledge this still hasn't been written.

There is a lack of people being seen by rheumatology, nearly all referrals are bounced back to GPs. Therefore, most people if they can afford it, pay to see a rheumatologist privately. This is the same for physiotherapy and other Musculoskeletal (MSK) services. It is our understanding that Rheumatology have been directed not to see people with non-inflammatory conditions. EDS is not an inflammatory condition, but it is a connective tissue disorder (CTD) and musculoskeletal (MSK) condition causing multi-systemic issues. Both MSK and CTD conditions normally fall under the remit of rheumatology.

A 2020 report on behalf of EDS Support UK discusses the experience of young people with EDS/HSD in Scotland. The report titled Ehlers-Danlos Syndrome and Young People in Scotland: The Agency Needed to

Prosper (by Anne-Marie Martin) states that young patients in Scotland perceive healthcare professional attitudes to be negative. This is particularly prevalent in young females who report that their symptoms are not believed. The diagnostic journey for EDS/HSD patients is problematic and the report revealed an inequity of service access in Scotland, with most patients surveyed reporting that they had to travel to London to receive a diagnosis. The report advocates for better education and training of health professionals in Scotland and the need for better service access geographically.

We have 850 members in Scotland located across every health board. The majority of whom are living with hEDS/HSD. Meetings are run twice monthly online and in person and we provide 1:1 support. We have lived experience from members and our volunteers who have 7+ years of experience supporting our members. EDS Support UK are currently represented at Cross Party Groups for Rare, Genetic and Undiagnosed Conditions, Arthritis and Musculoskeletal conditions as well as Chronic Pain.

In our experience very few people with hEDS/HSD have been accepted for an out of country referral and through our experience of supporting members there is no specialist centre for people to be sent to.

There is clearly still work to be done. In our experience 4 out of 5 of the people we speak to are undiagnosed and seeking help in getting a diagnosis and / or help to manage their condition.

We had hoped to obtain Scotland-wide data for patients diagnosed with hEDS/HSD but ICH-10 coding only found 321 patients from across Scotland, which suggests hEDS/HSD is not being recorded. In 2019 the old classification of EDS 3 was still being used. If the Scottish Government is basing its budgets on coding, it will be almost impossible to determine accurate data about who has hEDS/HSD which in turn leads to the issue of funding/commissioning of services for hEDS/HSD which is so badly needed.

Csecs et al 2022 found there appears to be a greater prevalence of neurodiversity for people with hypermobility compared to those in the general UK population. This often leads to patients being misdiagnosed and or dismissed entirely. This will account for a large number of our members waiting years for a diagnosis. Researchers and scientists in Scotland and across Europe are looking at changes to the connective tissue in the brain, as well as the body, to help understand the prevalence of neurodiversity.

A specialist service can provide patients with the confidence that the treatment options being offered are rooted in research, knowledge, experience and understanding of their condition. It will ensure that the impacts of surgery are well understood. This is rarely the case at present. We believe a specialist service will lead to more research opportunities from within the UK and internationally. Through our volunteers we already collaborate with researchers at the universities of Edinburgh and Strathclyde. We also have contacts with a hEDS specialist service and a genetics laboratory in North America.

We hope you can see why we are calling for services for hEDS/HSD to be funded or commissioned in Scotland. Our members have nowhere else to go.