Citizen Participation and Public Petitions Committee
Wednesday 9 October 2024
15th Meeting, 2024 (Session 6)

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

Introduction

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

1. The Committee last considered this petition at its meeting on 4 October 2023. At that meeting, the Committee agreed to write to the Scottish Government and the NHS National Services Division.

2. The petition summary is included in **Annexe A** and the Official Report of the Committee's last consideration of this petition is at **Annexe B**.

3. The Committee has received new written submissions from the Scottish Government, the National Services Division, the Petitioner and Emma Harper MSP which are set out in **Annexe C**.

4. Written submissions received prior to the Committee's last consideration can be found on the petition's webpage.

5. Further background information about this petition can be found in the SPICe briefing for this petition.

6. The Scottish Government gave its initial response to the petition on 6 September 2023.

7. Every petition collects signatures while it remains under consideration. At the time of writing, 2,232 signatures have been received on this petition.

Action

8. The Committee is invited to consider what action it wishes to take.

Clerks to the Committee
October 2024

Annexe A: Summary of petition

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 July 2023

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: Extract from Official Report of last consideration of PE2038 on 4 October 2023

The Convener: Our final new petition is PE2038, which has been lodged by Ehlers-Danlos Support UK and asks that suitable NHS services be commissioned for people with hypermobile Ehlers-Danlos syndrome, or HEDS, and hypermobility spectrum disorders, or HSD.

The SPICe briefing explains that Ehlers-Danlos syndromes are a group of rare inherited conditions that affect connective tissues in the body and that there are different types of EDS, including hypermobile EDS. The briefing also outlines other hypermobility spectrum disorders and notes that guidance on and guidelines for managing EDS have not been straightforward, due to some views that the evidence base is insufficient and varied opinions on the best way of managing the conditions.

The Scottish Government has outlined that diagnosis and patient care are provided by local and regional rheumatology services with the input of other specialities. Its submission highlights the Scottish rare disease action plan and states that the actions in the plan will address issues around the lack of signposting, referral pathways and overall care co-ordination, including for those living with HEDS and hypermobility spectrum disorders. The petitioner's written submission disputes the categorisation of HEDS and HSD as rare, stating low diagnosis of the issue and saying that four out of five people to whom it has spoken have not been diagnosed.

The submission also refers to a bid made in 2018 by Professor Stuart Ralston for a specialist centre that was supported by consultants, therapists and patients but not by the Scottish Society for Rheumatology. The petitioner highlights concern about access to services through rheumatology, stating its understanding that rheumatologists have been directed not to see people with non-inflammatory conditions.

In light of the submissions that we have received in addition to that from the petitioning organisation, do colleagues have any suggestions for action? If not, I suggest that we write to the national services division to ask whether it remains committed to producing a paper highlighting the issues and service gaps that people with EDS and HSD encounter; why the proposal in 2018 by Professor Stuart Ralston for a specialist EDS centre was rejected; and whether it has monitored the delivery of its commitment to encourage regional expertise and services in place of a specialist centre. We might also write to the Government to ask how it intends to engage with people with HEDS and HSD in taking forward actions under the rare disease action plan, either individually or through Ehlers-Danlos Support UK.

Are members content to proceed on that basis?

Members *indicated agreement.*

Annexe C: Written submissions

Scottish Government submission of 12 October 2023

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

Engagement with individuals living with hEDS and HSD has mainly been in two areas.

Individuals attended engagement events as part of the development of the Scottish Action Plan for Rare Diseases where we heard from members of the community about their lived experience.

The Scottish Government's Rare Diseases Policy Team receive correspondence from those living with are conditions though the Ministerial and Corporate Correspondence system. Such correspondence describing lived experience powerfully bring to life the challenges of living with a rare condition. These stories bring an important focus, not only raising awareness of individual rare conditions such as hEDS and HSD; but ensure the experiences of individual citizens are heard and their experience informs our thinking and future policies. The Scottish Government are listening, and will put lived experience at the heart of our actions to improve the lives of people in Scotland living with a rare condition in developing strategic policy for the NHS in Scotland.

With regards engagement with Ehlers-Danlos Support UK (EDS-UK), we can assure the Committee that it is the Rare Disease Implementation Board's intention to hold a number of "involvement meetings" early in 2024 which will be widely advertised. The Scottish Government has funded Genetic Alliance UK to carry out these meetings on our behalf.

The Scottish Government are also considering what additional stakeholder engagement activities may be required throughout 2024.

Officials would be happy to meet with EDS UK to explore their current priorities and discuss the work progressing under the current Rare Disease Action Plan. Finally, we'd invite EDS UK to contact Natalie Frankish (Policy and Engagement Manager for Scotland - Genetic Alliance UK) who is a member of the Scottish Government's Rare Disease Implementation Board (RDIB) and who could represent their views at the RDIB.

Planning and Quality Division

National Services Division submission of 13 October 2023

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

Thank-you for your recent correspondence from the Citizen Participation and Public Petitions Committee regarding a petition 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.

We were sorry to hear that those with symptoms suggestive of hEDS and HSD as well as those with a diagnosis of these conditions continue to describe difficulties in obtaining timely, equitable, person-centered diagnosis, treatment, and ongoing support to meet their needs.

National Services Division (NSD) previously facilitated a Short Life Working Group (SLWG) bringing together several clinical staff from across Scotland and the UK as well as representatives of the patient advocacy group with a view to understanding the needs of this group of patients and scoping how services could be delivered.

Although patients from Scotland can access NHS England highly specialist diagnostic EDS services located in Sheffield or London via NSD's close working relationship with the English commissioners, such services have a very specific remit and patient referral criteria and are not commissioned to provide ongoing support or treatment following diagnosis. It was therefore the recommendation of the SLWG that there was a need for specific specialist expertise in Scotland to improve patient care.

An NHS Lothian application led by Professor Ralston to develop a national specialist service to deliver care for this group of patients based upon the recommendations of the SLWG was however not supported back in 2019 by the National Specialist Services Committee and therefore the application did not progress to designation.

The reasons for non-progression included the lack of support for the proposal from the Scottish Society of Rheumatology, the need for initial assessment and treatment by local rheumatology services prior to referral to the national service and that care might be better delivered through the development of a set of clinical guidelines, a patient pathway of care or a networked community of practice.

On this basis the recommendation of the National Specialist Services Committee (NSSC) was that this need should be passed back to the clinical community for further development.

The ongoing diagnostic, treatment, and care needs of hEDS and HSD patients are the responsibility of individual Health Boards across Scotland who commission and/or provide rheumatology, clinical genetic, cardiology, gastroenterology, pain management, physiotherapy, and occupational therapy services which these individuals often require.

The responsibilities of NSD do not extend to the performance management, planning, operational delivery, or scrutiny of such services and therefore it is not possible for NSD to undertake a needs or service gap analysis nor monitor service developments.

The option remains open to the clinical community to formally register a new application for consideration of designation of a national specialist service or managed clinical network however such an application would require the endorsement of the Chief Executive of the Health Board where the clinician is in

clinical practice. The designation process now follows an annualized cycle with registration for the next cycle closing on 31 March 2024 for consideration during year 2024/2025.

We hope that this information is helpful to you however should you have any further queries please do not hesitate to contact us.

Petitioner submission of 13 September 2024

PE2038/E: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders

We have new evidence to support the provision of suitable NHS services for people with hypermobile Ehlers-Danlos syndrome (hEDS) and hypermobility spectrum disorders (HSD). This evidence was presented by EDS UK, researchers Kathryn Berg and Dervil Dockrell and a range of Healthcare professionals at an exhibition in Holyrood on the 7-9 May 2024 during EDS awareness month and a roundtable hosted by Michael Marra MSP on the 4 September 2024.

Evidence that hEDS/HSD is not rare and evidence of lack of NHS services

hEDS/HSD are not rare types of Ehlers-Danlos syndromes, they are rarely diagnosed. A study of diagnosed prevalence in Northumberland, Frankel B et al 2023, showed that as many as 1 in 227 had a diagnosis. Demmler et al 2019 found that 1 in 500 people had a diagnosis of hEDS/HSD. That's between 11 and 24 thousand people living with hEDS/HSD in Scotland. These are only the people who actually have a diagnosis. Berg K and Dockrell D 2024 hEDS-START study found an average 20 year wait for diagnosis with 22% of people paying privately to get a diagnosis, 1 in 4 leaving Scotland to access hEDS/HSD related healthcare elsewhere and 1 in 7 being diagnosed in England. This demonstrates a clear failure to provide suitable NHS services for people with hEDS/HSD in Scotland.

National Services Division response PE2038/D

In October 2023, the Committee wrote to the National Services Division (NSD) to ask why the 2018 proposal for a specialist centre was rejected. They were also asked to confirm if the commitment from the NSD to produce a paper for regional directors of planning highlighting which issues and service gaps people with EDS and HSD encounter still stood. We are not aware of any paper being produced and there is no acknowledgement of this in their response.

We would also like to draw your attention to the suggested alternatives given by NSD to our original application for a specialist service "The reasons for non-progression included the lack of support for the proposal from the Scottish Society of Rheumatology, the need for initial assessment and treatment by local rheumatology services prior to referral to the national service and ***that care might be better delivered through the development of a set of clinical guidelines, a patient pathway of care or a networked community of practice***". This is exactly what we are trying to achieve but we are being told from Health Improvement Scotland there is not enough evidence to support SIGN guidelines. This is despite the evidence

provided above and over 6,000 people providing their lived experience evidence in the hEDS START project.

Scottish Government response PE2038/C

The Scottish Government has cited the Scottish Rare Disease Action Plan (SRDAP) in its response. EDS UK has participated in events for SRDAP and has used these opportunities to advocate on behalf of members living with the rarer forms of EDS, recognising that the plan's broad actions to improve awareness of rare conditions more generally will, eventually, result in improvements in the experience of care for people with rare conditions. To maintain momentum we would welcome the opportunity to meet with officials to discuss EDS UK's current priorities and work progressing under the SRDAP as offered.

However, the development of specialist services for individual rare conditions is not within the scope of the SRDAP. The remit of Scotland's Rare Disease Implementation Board does not extend to decision making relating to the commissioning of specialist services, clinical pathways and guidelines in Scotland. Therefore, the SRDAP is not an appropriate route to address the urgent unmet clinical needs of the hEDS/HSD community in Scotland that this petition aims to tackle and is therefore not a solution. The evidence shows hEDS/HSD is not rare and is therefore not covered by the SRDAP.

What is happening in other UK nations

Through the EDS UK petitions, in May 2024 NHS Wales committed to co-creating a hypermobility pathway for primary care to help GPs diagnose and manage these conditions. Their Health Minister also gave EDS UK the contact for the Welsh Rare Disease Partnership to look at the rarer types of EDS. In England, a debate was held in Westminster and the Health and Secondary Care Minister offered a meeting with NHS England. The election was announced after the debate and so the meeting was postponed but we are confident this will be honoured. In Northern Ireland, we have meetings planned with MLAs to secure a similar debate.

What we need across the UK

Across all four nations we are seeking the following:

- A pathway for NHS diagnosis and care for hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders.
- NICE/SIGN guidelines for Ehlers-Danlos syndromes and hypermobility spectrum disorders.
- A coordinated, multidisciplinary approach to diagnosis and care for people with hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders and their associated co-morbidities.
- Support and training for healthcare professionals to deliver this.

What we have already done to provide a starting point

The hEDS-START study held a patient engagement event with EDS UK where a wish list for a pathway of care and a list of values to underpin a regional service in Scotland were developed. This study provides a lived experience perspective and evidence base.

We also have the EDS UK GP toolkit which they are using in Wales as a basis for the primary care pathway.

We remain willing to engage with the Scottish Government in finding a solution for implementing the pathway, guidelines and training that are so desperately needed.

Emma Harper MSP submission of 13 September 2024

PE2038/F: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders

I'm sorry that I couldn't be there in person today due to a committee clash, but I am glad to be able to share some thoughts regardless on an issue very close to my heart.

Over the last three years, I've had the pleasure of meeting many others who struggle daily with normal tasks, like walking, sitting, typing, and living. I know that too many struggle also to get the help that they need with their pain and other symptoms due to a lack of diagnosis or available treatment.

When we meet each other, we will often get excited when the other mentions something like neck headaches, the difference in pain between crisp packet crepitus and everyday joint clicking, or getting toothache in your hip. Nobody is happy that others feel these things, but most patients with chronic pain will experience a type of gaslighting by the world; being told over and over that the bizarre is normal, the exhausting is routine, and the excruciating is mild. The joy of hearing someone else acknowledge and validate your experience is so extreme because of the dismissal we've experienced before.

Doctors cannot be expected to know everything all the time. There are so many so-called rare conditions which affect many thousands of people in Scotland. I do not expect every GP to look at hypermobile joints, migraines, gastro issues, and chronic pain and think "EDS". I do not expect them to see chronic pain and know exactly what the cause is. But there should be a nationally agreed standard for pain pathways to ensure that people do not fall through the cracks, left to suffer.

We know that earlier diagnosis and earlier treatment for EDS mean you are less likely to develop more severe symptoms. We know that quality of life can increase drastically, and the cost to the NHS should, therefore, also be lower.

Living with chronic pain can seem impossible. I have had times where I can barely move, barely think through the pain, but the receptionist at the end of the phone simply asks me to try paracetamol and ibuprofen for a few days. I've been taken to hospital with medication-induced hepatitis because I have relied on too much of the wrong medication for too long. These are very common situations, and it is very common for people with chronic pain to end up suicidal.

EDS Support UK has been clear in its calls for a pain pathway and formally commissioned services for people with EDS – with input from lived experience. Many people giving evidence and energy to this committee have been clear why this is necessary.

I hope that the Committee will progress this petition, which gives voice and hope to many of our constituents who are suffering unnecessarily.