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."

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15 August 2023

The purpose of this briefing is to provide a brief overview of issues raised by the petition. SPICe research specialists are not able to discuss the content of petition briefings with petitioners or other members of the public. However, if you have any comments on any petition briefing you can email us at spice@parliament.scot

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