NHS

Ehlers Danlos Syndrome
National Diagnostic Service

Information for patients

# Hypermobile EDS (hEDS) and Hypermobility Spectrum Disorder (HSD)

## Patient information leaflet

# Hypermobile EDS (hEDS) and Hypermobility Spectrum Disorder (HSD)

## Introduction

This leaflet will give you more information about hypermobile Ehlers Danlos syndrome (hEDS) and Hypermobility Spectrum Disorder (HSD).

## What is Ehlers Danlos syndrome (EDS)?

The Ehlers Danlos syndromes are a collection of inherited conditions that fit into a larger group known as hereditary disorders of connective tissue. Connective tissues provide support in areas such as the skin, tendons, ligaments and bones.

There are several different, distinct types of EDS, but they have some features in common. These can include joint hypermobility (described below), stretchy skin and tissue fragility.

## What is joint hypermobility?

Hypermobile joints are joints that move further than the usual range. This can vary taking into account someone's age, sex and ethnic background. Many individuals have one or more hypermobile joints and factors such as muscle tone can increase the range of movement of a joint. For some this is not associated with any difficulties. For others the hypermobility can cause problems and it appears to co-exist with a number of other issues outlined in the diagnostic criteria.

## How is hypermobile EDS diagnosed?

The criteria for hypermobile EDS (hEDS) are stricter than they used to be, and it is hoped that that this will allow a genetic cause (or causes) of hEDS to be established in the future. People who were diagnosed with hEDS in the past (prior to 2017) may not now fulfil the diagnostic criteria for hEDS. However, it was agreed that individuals with a historical diagnosis of hEDS should not need to be reassessed, unless there is good reason, such as taking part in research into hEDS.

To be diagnosed with hEDS, one needs to meet three separate groups of criteria.

Criterion 1. An assessment of past and present hypermobility using a scoring system called the Beighton Score.
Criterion 2. This is divided into three features. To meet this criterion, an individual must meet two of these three features.

- **a.** Having at least five from a list of 12 signs and symptoms that can be identified by physical examination and additional investigations.
- **b.** Having a close family member (parent, child, brother or sister) who independently meets the criteria for a diagnosis of hypermobile EDS.
- **c.** Having significant pain or unstable joints.

Criterion 3. Other types of EDS and related connective tissue disorders need to have been considered by a doctor and ruled out.

Currently other signs and symptoms are not part of the 12 signs and symptoms mentioned above but an individual can still have these issues and fulfil criteria for hypermobile EDS. These include; postural tachycardia syndrome (POTS), which causes dizziness by an abnormally large increase in heart rate, mast cell activation disorder (MCAD), which is an immunological condition and gastrointestinal problems. Research has shown there is an association between joint hypermobility and the occurrence of these signs and symptoms but there is no proof that they are linked, or due to an underlying identical cause.

## How are Hypermobility Spectrum Disorders (HSD) diagnosed?

Hypermobility spectrum disorders (HSD) are a group of conditions related to joint hypermobility (previously referred to as benign joint hypermobility syndrome). HSD are diagnosed when clinical features of other diagnoses associated with hypermobility are not present. HSD, just like hypermobile EDS, can cause significant health problems which should be managed and supported appropriately.

## Is hypermobile EDS worse than Hypermobility Spectrum Disorders?

Not necessarily. In people with these diagnoses the same range of symptoms can be observed. What is important is that the symptoms are managed appropriately.

## What causes Hypermobility Spectrum Disorders?

Hypermobility spectrum disorders (HSD) are diagnosed clinically. A specific genetic cause has not yet been identified, although a genetic component is suspected, given the way this condition can run in families. The reason that a specific genetic cause has not yet been identified may be because:

- There may be more than one gene involved.
- There may be different causes in different families.

Sometimes there is only one person in a family who has HSD, but in some families there may be more than one person. Therefore, it is difficult to predict the chance of a parent passing these conditions on to their child. In addition, symptoms can vary, even within a family, with some family members being mildly affected and others having more severe problems. Sometimes one family member may be very hypermobile and have few other problems, while a relative may be less hypermobile but suffer from significant pain, dislocations, gut problems or POTS.

## Is there a test for hypermobile EDS or Hypermobility Spectrum Disorders?

The diagnosis of these conditions is made using clinical features, such as the signs and symptoms someone has, along with their medical and family history. There are currently no genetic tests available to confirm the diagnosis.

## What management is recommended for people with hypermobile EDS or Hypermobility Spectrum Disorders?

It is important to stay active.

- Regular gentle exercise, such as walking, cycling or swimming is encouraged to keep joints mobilised and to build up muscle tone around the joints to help stabilise them. Pilates can be beneficial in helping maintain core stability and to develop good posture. Care should be taken to choose appropriate physical activities, particularly if someone has had joint dislocations. A physiotherapist can help you to develop appropriate exercises which you can do at home and give advice on other suitable activities.
- Maintaining a healthy weight by sensible eating and appropriate exercise will help to avoid additional stress on the joints.
- Some people may benefit from a referral to medical specialities such as pain management, rheumatology, gastroenterology, physiotherapy or occupational therapy. This will depend upon individual needs.
- Your doctor may suggest a one-off heart scan (echocardiogram).

## Are there any recommendations for women with hypermobile EDS or Hypermobility Spectrum Disorders who are considering a pregnancy?

Hypermobile EDS (hEDS) and also Hypermobility Spectrum Disorders (HSD) are not usually associated with serious complications in pregnancy. We would suggest that women with these conditions inform their obstetrician (a doctor who specialises in the care of pregnant women) of their diagnosis.

There are a few points for pregnant women with hEDS and also with HSD to consider. Joint hypermobility tends to increase during pregnancy, due to the female hormones produced, and this may increase instability and joint pain. Exercises to strengthen the pelvic floor are particularly important for women with hEDS and also with HSD, as pelvic weakness can be a symptom in some people with these conditions.

## Where can I get more information and support?

Hypermobility Syndromes Association (HMSA)
Helpline: 03330 116 388
Web: www.hypermobility.org/

Ehlers-Danlos Support UK (EDS UK)
Helpline: 0800 907 8518
Web: www.ehlers-danlos.org
hEDS/HSD link: www.ehlers-danlos.org/what-is-eds/information-on-eds/hypermobile-eds-and-hypermobility-spectrum-disorders/

Helpline and support: helpline@ehlers-danlos.org

The Ehlers-Danlos Society – Europe Office
Tel: 0203 887 6132
Web: www.ehlers-danlos.com
Helpline: www.ehlers-danlos.com/eds-helpline/.

Stars (for POTS advice) https://www.heartrhythmalliance.org/stars/uk

PoTS UK (for POTS advice)
Web: www.potsuk.org
Email: support@potsuk.org