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

Information for patients

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

A member of staff will talk to you about the information in this leaflet but you may want to keep it to read again later.

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, 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 new diagnostic criteria.

What is hypermobile EDS and why has the definition changed?

The names 'hypermobile EDS' and 'joint hypermobility syndrome' have been used interchangeably for several years. However it was decided at an international meeting in 2016 that the criteria for all types of EDS should be updated. The criteria for hypermobile EDS are now stricter and it is hoped that that this will allow the genetic cause (or causes) of hypermobile EDS to be established in the future.

People who were diagnosed with hypermobile EDS in the past may not fulfil the new diagnostic criteria for hypermobile EDS. However, it was agreed that individuals with

the old diagnosis should not need to be reassessed, unless there is good reason, such as taking part in research into hypermobile EDS.

It was agreed that the term 'joint hypermobility syndrome' would no longer be used and that Hypermobility Spectrum Disorder is a better description of the range of signs and symptoms seen. Following the 2016 meeting, new international criteria for diagnosing hypermobile EDS were published in 2017. This is summarised in the next section.

What are the new diagnostic criteria for hypermobile EDS?

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

1. An assessment of present and historical hypermobility using a scoring system called the Beighton Score.
2. This is divided into three features. To meet this criterion, an individual must fulfil two of these three features.
    a. Having at least 5 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.
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 including postural tachycardia syndrome, mast cell activation disorder and gastrointestinal problems 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. 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.

More detailed information about hypermobile EDS can be found on the EDS Society website, details of which are listed at the end of this leaflet.

Is hypermobile EDS worse than HSD?

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 are the different types of HSD?

There are new categories of HSD depending on which joints are involved. These are:

- Generalised HSD, involving a number of large joints like knees, shoulders and elbows
- Peripheral HSD, involving joints in the hands and feet
- Localised HSD, involving a single joint or small group of joints in the same area

- Historical HSD, where people were previously hypermobile, and have become stiffer over time

What are the main symptoms and signs of hypermobility spectrum disorder?

Individuals with HSD may have some of the following:

- Hypermobile joints that can move beyond normal limits (hyperextend)
- Loose, unstable joints that can lead to partial or full dislocations
- Joint pain and fatigue
- Decreased bone mass, which is usually only mild in HSD
- Co-ordination problems and difficulties in movements (dyspraxia)
- Early onset degeneration of joint cartilage and the underlying bone (osteoarthritis). This mainly causes stiffness.
- Easy bruising
- Gastrointestinal dysfunction such as constipation or irritable bowel disease
- Postural tachycardia syndrome (POTS) causing fast heart rate, dizziness and fainting
- Heart valve abnormality (mitral valve prolapse) which is usually only mild in HSD
- Uterine, rectal or bladder prolapse (slipping down/forward of the organ)
- Urinary dysfunction such as loss of bladder control, urgency and urinary tract infections

What causes HSD and how does it occur?

The exact cause or causes of HSD are unknown. The features of HSD suggest that there is a problem with connective tissues and possibly collagen. Collagen is the main group of structural proteins found in connective tissue.

Sometimes there is only one person in a family who has HSD, but in some families there may be more than one. Therefore, it is difficult to predict the chance of a parent passing HSD on to their child. There are possible explanations for this:

- There may be more than one gene involved
- There may be different causes in different families.
- There may be an effect of different genes in combination with the environment and lifestyle factors. This is known as multifactorial.

Can family members have different symptoms?

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 HSD?

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

What management is recommended for people with HSD?

It is important to stay active and learn the skills and knowledge to self-manage your symptoms..

- 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 or other activities which focus on strengthening and improving balance and co-ordination are beneficial in helping maintain core stability and to develop good posture. The most important goal is to find an activity that you enjoy. A physiotherapist can help you to develop appropriate exercises which can be carried out at home and give advice on other suitable activities and goals you wish to achieve.
- 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 HSD who are considering a pregnancy?

Hypermobility spectrum disorder is not usually associated with serious complications in pregnancy. We would suggest that women with HSD 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 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 HSD, as pelvic weakness can be a symptom in some HSD patients.

Other sources of information and support:

Hypermobility Syndromes Association (HMSA)
Tel: **03330 116 388**
Web: http://hypermobility.org/

EDS UK - Ehlers-Danlos Support UK
Tel: 0800 907 8518
Web: www.ehlers-danlos.org
Email: info@ehlers-danlos.org

The Ehlers-Danlos Society – Europe Office
Tel: 0203 887 6132
Web: www.ehlers-danlos.com
E-mail: info@ehlers-danlos.com

Stars (for POTS advice)
Web: www.stars.org.uk

PoTS UK
Web: www.potsuk.org
Email: info@potsuk.org

This information leaflet was written by the NHS Ehlers Danlos Syndrome National Diagnostic Service. For more information please contact the appropriate EDS team:

North of England:
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EDS National Diagnostic Service
Level 8V
Northwick Park and St Mark's Hospital
Watford Rd
Harrow
HA1 3UJ
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General Trust information

Patient Advice and Liaison Service (PALS)

PALS is a confidential service for people who would like information, help or advice about the services provided by any of our hospitals. Please call 0800 783 4372 between 9.30am and 4.30pm or e-mail LNWH-tr.PALS@nhs.net. Please note that this service does not provide clinical advice so please contact the relevant department directly to discuss any concerns or queries about your upcoming test, examination or operation.

If you would like this information in an easy to read format, large print, braille, different format or language, please contact the PALS team on 020 8869 5118 or email lnwh-tr.PALS@nhs.net. We will do our best to meet your needs.