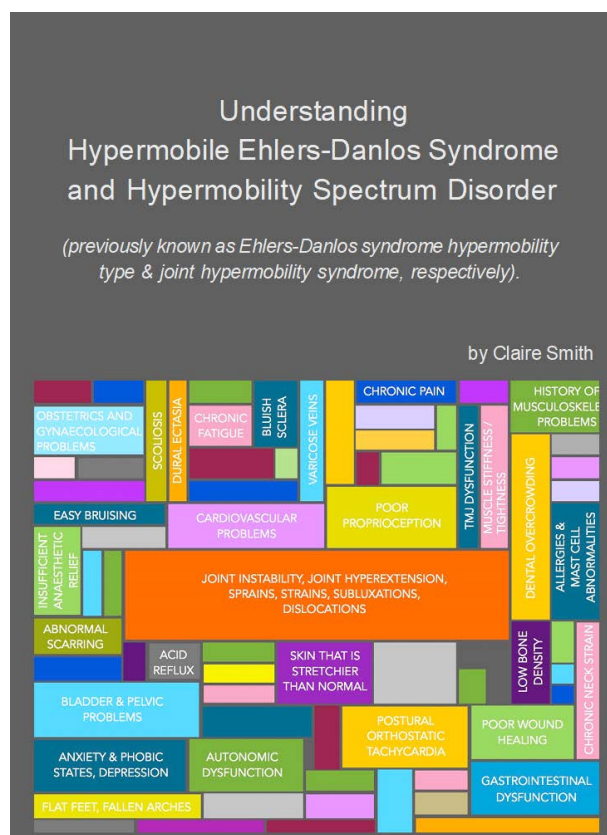


Initial diagnosis by clinical assessment - hEDS & HSD

The following extract is taken from:
Understanding hypermobile Ehlers-Danlos
Syndrome and Hypermobility Spectrum
Disorder

Chapter 3 Diagnosis and Management



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Initial diagnosis by clinical assessment

If a doctor suspects that a patient has a hypermobility related disorder, he or she may diagnose the patient themselves (using the criteria and scoring system shown on pages 121 and 122), or they may choose to refer the patient to a rheumatologist to undergo a clinical examination and assessment of their medical and family history. For some types of Ehlers-Danlos syndrome, genetic testing is also available to help confirm diagnosis but, at the current time, no genetic test is available for hEDS or HSD.

In this section, we will look at the route of 'diagnosis via clinical assessment', and at some of the services that may, in some instances, be involved in a patient's care.

A note to clinicians:

HEDS, and HSD are difficult to diagnose because patients often look well and present differently from each other. The severity of the wide ranging, multi-systemic symptoms, the joints that are affected and the level of pain and fatigue, experienced by those with hEDS or HSD, can vary greatly from day to day, or even hour to hour, and in each individual (3/HMSA 2014).

In terms of joint hypermobility, some individuals are very mobile, while others who were once mobile are no longer so. Patients generally fall into two broad groups: The first being those who suffer periods of pain and injury, usually lasting several weeks. After effective treatment, they can be relatively symptom-free for some time but experience symptoms intermittently. For patients in the other group, who may be more severely affected, each day is a struggle against pain and injury (Gurley-Green S. 2001). Patients from both groups report deterioration over time, which is contrary to much in the literature which suggests that symptoms decrease with age. Reported stiffening with age does not always bring less pain. On the contrary, many report increasingly painful symptoms as age advances (Arthritis Research UK / 1/EDNF.org, Uptodate.com).

Pain is the most common symptom reported. For patients it comes in varying degrees, and is often quite unbearable (Levy H.P. 2012, Chopra P. et al 2017).

Persistent pain can affect cognition and clarity, which limits good communication between patient and doctor (Gurley-Green S. 2001).

With such complex disorders it is often difficult for patients to tell their doctors the impact that hEDS, and HSD is having on them and their loved ones. In some cases, the whole family may need treatment/guidance, as this disorder can affect every aspect of daily and family life.

Many patients have struggled for years with medical treatment that has made them worse, not better. Many receive fragmented and single-body-system, rather than holistic, care; something which is not only detrimental to their physical and psychosocial wellbeing but is also non-cost-effective for the health care provider. Inappropriate interventions and poor past experiences in these individuals have often resulted in increased psychological and physiological distress, and the understandable adoption of coping mechanisms that can sometimes become a barrier to further management. General practitioners often do not recognise that there may be a need to send their patients to specialised clinics. Patients are left feeling isolated and disbelieved; frustrated by the lack of effective treatment/service provider knowledge, and with no clear pathway available.

The route to diagnosis may depend on many factors, including the patient's GP's own knowledge of heritable disorders of connective tissue, and the original and 'presenting symptoms' for which a patient first attends a clinic. When taken individually, symptoms often lead down many dead ends and it is not until underlying hypermobility is recognised that the dots are finally joined and hEDS, or HSD is recognised. Unfortunately, it is not uncommon for patients to attend many appointments before this possibility is considered; in fact, the Hypermobility Syndromes Association state that for 55% of its members it takes more than 10 years to receive a diagnosis.

With this in mind, please remember:

- If the patient has struggled to obtain a diagnosis, the chances are that they may, in the past, have been made to believe that their symptoms are psychosomatic or that they may have injured themselves on purpose. This scenario is most common when the patient does not recall an 'accident', which would normally account for such pain or injury, or when their description of how an injury occurred could not (seemingly) account for such a severe injury in a normal patient.

Continued...

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- He/she may have high expectations, having waited many weeks for an appointment to see the consultant and having invested in the hope that the specialist might be able to help.
- They may be depressed as a result of their pain and disability. In addition, they may seem desperate, willing to do anything to be free from the pain.
- The patient may appear to be angry at all medical professionals after years of inadequate or inappropriate care.
- They may be defensive and anxious to prove that they need help, and they may therefore appear to be exaggerating their problem.
- Finally, they may be unclear what symptoms to relate, as the problems may have been present throughout their life and have become 'their normal.'

(Gurley-Green S. 2001 / Hamonet C. 2017)

Patient-led management and self management are currently the best approaches for the long-term treatment of both hEDS and HSD (along with multi-disciplinary referral where required). Unfortunately, patient-led management and self management are particularly problematical for the hEDS/HSD patient because they are disorders that can rob patients of control over their lives. All chronic illness and chronic pain cause feelings of loss of control. The problem is exacerbated with hEDS/HSD because there is not always a clear link between an activity and the onset of pain or injury; making it difficult for the patient to lower the level of pain, or avoid injury, by behavioral modification. This lack of a clear cause-and-effect relationship between an event and subsequent pain can make patients unsure of their body's limits, and can promote self-blame and poor self-esteem, ultimately leading to a lowering of the patient's strength through progressive physical inactivity (Gurley-Green S. 2001).

For successful self management, it is important for patients to feel some internal sense of control, to feel that they can change their lives for the better. This self-empowerment is often made more difficult because many hEDS/HSD patients have to go from one doctor or alternative practitioner to another in search of help. Many have complained of having to see a different physiotherapist each time they are referred for treatment, often with serious consequences relating to lack of continuity and conflicting advice.

Another aspect of this problem is that because pain often occurs some time after the activity that caused it, exercise programmes are difficult to maintain. The lack of regular appropriate exercise, combined with self-blame, may lead to weight gain for some patients. The resultant lowering of self-esteem fuels the downward spiral, putting both patient and doctor in a difficult position. Doctors correctly suggest to an already vulnerable patient that the weight gain is making the problem worse. This fuels the self-blame and accentuates the downward spiral (Gurley-Green S. 2001).

To support patients, it is important to recognise the impact of hEDS, and HSD, on their lives; research into the development of strategies of management, including physiotherapy and drug and cognitive therapies; research into patterns of symptoms to determine why, for example, many people who are very mobile have no pain while others who are less mobile are suffering; respond to the needs of the patient, and if unable to answer a patient's question, *explain that* to the patient whilst providing reassurance that you will work with them to find the answer.

Diagnostic criteria

The pathway to EDS/HSD diagnosis starts with a physical examination, using the Beighton Scale to assess how mobile joints are, a search for abnormal scarring and testing the skin to determine what it feels like and how much it stretches, as well as any additional tests they physician feels are needed. There should also be a look into a patients medical history to look for conditions and problems associated with EDS/HSD, and a discussion about family to help determine if an EDS/HSD was inherited (Ehlers-Danlos Society 2017).

The 2017 International Criteria for hEDS (incorporating the Beighton Score for generalised joint hypermobility) has been laid out on page 121, **Box 2**. The criteria for other types of Ehlers-Danlos syndrome can be found on pages 127-128.

Please note that, when using the Beighton Score to diagnose generalised joint hypermobility, several additional points need to be taken into consideration.

These are shown in **Box 3** and include:

- The need for other symptoms to be present.
- Score adjustments that may need to be made relating to age, past surgeries etc.

Continued after Boxes 2 & 3...

Box 2. 2017 International Criteria for hypermobile Ehlers-Danlos syndrome

Incorporating the Beighton Score

Note: The clinical diagnosis of hypermobile Ehlers-Danlos syndrome (hEDS) requires the simultaneous presence of criteria 1 AND 2 AND 3.

Criterion 1

Generalised joint hypermobility (GJH) must be present

Using the Beighton Score (see page 122, **Box 3**) the cut-off for the definition of GJH is:

- 6 points or more (out of a total of 9) for pre-pubertal children and adolescents
- 5 points or more (out of a total of 9) for men and women post puberty up to 50 years of age
- 4 points or more (out of a total of 9) for men and women over 50 years of age

Important note:

Additional factors that **MUST** also be taken into consideration when using the Beighton score system are detailed on page 122, **Box 3 (points i - iii)**.

Criterion 3

All of the following prerequisites **MUST** be met

- 1) Absence of unusual skin fragility, which should prompt consideration of other types of EDS
- 2) Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions. In patients with an acquired connective tissue disorder (e.g. lupus, rheumatoid arthritis, etc.), additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted towards a diagnosis of hEDS in this situation.
- 3) Exclusion of alternative diagnoses that may also include joint hypermobility by means of hypotonia and/or connective tissue laxity. Alternative diagnoses and diagnostic categories include, but are not limited to, neuromuscular disorders (e.g. Bethlem myopathy), other hereditary disorders of connective tissue (e.g. other types of EDS, Loeys-Dietz syndrome, Marfan syndrome), and skeletal dysplasias (e.g. osteogenesis imperfecta). Exclusion of these considerations may be based upon history, physical examination, and/or molecular genetic testing, as indicated.

Criterion 2

Two or more among the following features **MUST** be present (e.g. A and B; A and C; B and C, or A and B and C)

Feature A - Systemic manifestations of a more generalised connective tissue disorder (NB/ a total of 5 must be present)

- 1) Unusually soft or velvety skin
- 2) Mild skin hyperextensibility
- 3) Unexplained striae such as striae distensae or rubrae (at the back, groins, thighs, breasts and/or abdomen in adolescents, men or prepubertal women without a history of significant gain or loss of body fat or weight)
- 4) Bilateral piezogenic papules of the heel
- 5) Recurrent or multiple abdominal hernia(s) (e.g., umbilical, inguinal, crural)
- 6) Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosiderotic scars as seen in classical EDS
- 7) Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
- 8) Dental crowding AND high or narrow palate
- 9) Arachnodactyly, as defined in one or more of the following: (i) positive wrist sign (Steinberg sign) on both sides; (ii) positive thumb sign (Walker sign) on both sides
- 10) An arm span-to-height ratio of equal to or greater than 1.05
- 11) Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- 12) Aortic root dilatation with Z-score greater than +2

Feature B - Positive family history

One or more first degree relative(s) must independently meet the current diagnostic criteria for hEDS (biological mother, father, brother, sister).

Feature C - Musculoskeletal complications

The patient must have at least one of the following three:

- 1) Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- 2) Chronic, widespread pain for 3 months or longer
- 3) Recurrent joint dislocations or clinically evident joint instability, in the absence of trauma (a or b):
 - a) Three or more atraumatic dislocations in the same joint, or two or more atraumatic dislocations in two different joints occurring at different times
 - b) Medical confirmation of joint instability at 2 or more sites not related to trauma

General Comment: Many other features are described in hEDS but most are not sufficiently test specific or test sensitive to be included in formal diagnostic criteria, at the moment. These include but are not limited to: sleep disturbance, fatigue, postural orthostatic tachycardia, functional gastrointestinal disorders, dysautonomia, anxiety, and depression. These other systemic manifestations may be more debilitating than the joint symptoms, often impair functionality and quality of life, and should always be determined during clinical encounters. While they are not part of the diagnostic criteria, the presence of such systemic manifestations may prompt consideration of hEDS in the differential diagnosis.

(Criteria: Malfait F et al. 2017)

Box 3. The Beighton Score to diagnose generalised joint hypermobility

The Beighton Score has been used for many years as an indicator of generalised joint hypermobility (GJH).

- **Score 1 point** if you can bend forward from the waist, with your knees straight and put your palms flat on the floor
- **Score 1 point** for each elbow that you can hyperextend backwards (see picture below)
- **Score 1 point** for each knee you can hyperextend backwards (see picture below)
- **Score 1 point** for each thumb back you can bend back to meet your forearm (see picture below)
- **Score 1 point** for each little finger you can bend back beyond 90 degrees towards the back of the hand (see picture below)

The International Classification 2017 defines the cut-off for the definition of GJH using the Beighton Score as:

6 points or more (out of a total of 9) for pre-pubertal children and adolescents

5 points or more (out of a total of 9) for men and women post puberty up to 50 years of age

4 points or more (out of a total of 9) for men and women over 50 years of age

Important notes:

As a clinical tool the Beighton Scoring System (above) can be a quick and straightforward thing to do **BUT** there are additional factors that **must** be taken into consideration :

i. A high Beighton score by itself does not automatically mean that an individual has a hypermobility related disorder - other symptoms and signs need to also be present. Patients with Hypermobility Spectrum Disorder usually experience one or more of the following musculoskeletal manifestations (Castori M. et al 2017):

Pain: Occasional, recurring pain is a natural result of the trauma, but chronic pain can develop—perhaps because of unusual sensitivity to pain (hyperalgesia), perhaps because of an impaired connective tissue function (as suggested by the discovery of small fiber neuropathy in adults with classical, hypermobile, and vascular EDS).

Musculoskeletal and/or soft tissue trauma: Macrotrauma includes dislocation, subluxations, and connected soft tissue damage (ligaments, tendons, muscles). It can cause acute pain and loss of joint function. Microtrauma are injuries too small for them to be noticed as they happen. Over time, they may make one susceptible to recurrent or persistent pain, and possibly early joint degeneration like osteoarthritis.

Disturbed Proprioception: Proprioception is the sense of the relative position of parts of the body and how much effort is needed for movement—can be reduced. Not understanding where our joints are and how much muscle strength it takes to use them can lead to a cycle that increasingly limits our abilities to manage every day life.

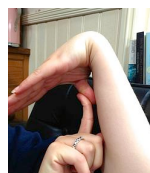
Other Musculoskeletal Traits: Those with GJH often have other minor musculoskeletal physical traits, which may be the result of the interactions between “softer” musculoskeletal tissues and mechanical forces during growth. These include flat feet (flexible type), misaligned bones in the elbow and big toes,

mild to moderate scoliosis (side to side curvature of the spine), kyphosis (outward curvature) of the upper spine and lordosis (inner curvature) of the lower spine. There may be an indirect association with mild reduced bone mass as a result of many factors—lack of proprioception, muscle weakness, and the resulting reduced activity (All above Ehlers Danlos Society 2017a)

ii. Score adjustments should be made for injury/trauma, past surgeries, wheelchair use, amputations and age etc. If a patient scores one point below the required total but answers ‘yes’ to two or more of the questions listed in the five-point questionnaire (see below), GJH can still be diagnosed (Malfait F. et al 2017).

- 1) As a child did you amuse your friends by contorting your body into strange shapes, or could you do the splits?
- 2) As a child or teenager, did your shoulder or knee-cap dislocate on more than one occasion?
- 3) Do you consider yourself ‘double-jointed’?
- 4) Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- 5) Can you now (or could you ever) bend your thumb back to touch your forearm? (All above: Hakim A. 2017 HMSA).

iii. A low score should be considered with caution when assessing someone, as hypermobility can be present at a number of sites that are not counted in the Beighton Score. For example, at the jaw joint (the ‘TMJ’), neck (cervical spine), shoulders, mid (thoracic) spine, hips, ankles and feet. Joints outside these scores should be assessed also, and in particular if they are sites of pain / injury. For example, the shoulder, hip and ankle are common sites of pain and instability but are not in the Beighton Score – in such a situation, only using the Beighton Score to decide whether hypermobility might explain a presentation is inappropriate (Hakim A. 2017 HMSA).



Pictures showing a patient touching their thumb to their wrist; bending their little finger back; a patient's knees and elbow in hyper-extension; bending forwards and placing their palms flat to the floor (all past the range of movement that is considered 'normal')

Additional comment: Establishing whether a joint is hypermobile or not is a relatively easy task and it is carried out by (i) using professional tools, such as the orthopaedic goniometer; (ii) following specific procedures (e.g. Juul-Kristensen B. et al., 2007; and (iii) comparing the measured range of motion (ROM) with normal parameters. (Tinkle B. et al 2017)

If there is any concern that a patient may have a connective tissue disorder other than HSD or hEDS, they should be referred for expert opinion, and further assessment might include genetic testing, vascular imaging and ophthalmic review.

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- The need for all joints to be assessed (not just those described in the scale).

The latter is well illustrated by a parent of children diagnosed with HSD, who herself has the condition, she writes:

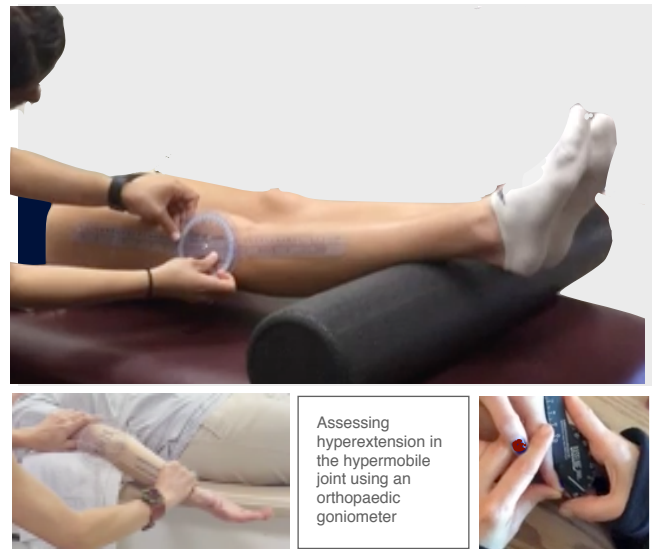
'My daughter can't touch her thumb to her wrist let alone her palms to the ground, but my son and I both can. My daughter's elbows, shoulder, hips and thumbs ARE hypermobile. My son scores very low number on the Beighton scale; he can't touch his thumb to wrist, but he can bend all his fingers together up to 90 degrees backwards, and also has hypermobile shoulders, hips, patella and other joints!'

Reaching a diagnosis

Hypermobile Ehlers-Danlos syndrome

Having clinically assessed a patient using the diagnostic assessment criterion shown in **Boxes 2 and 3** (pages 121 and 122), and considered and excluded all other differential diagnoses (criterion 3, **Box 2**), those who meet the criteria for hypermobile Ehlers-Danlos syndrome (hEDS) should receive this diagnosis. (NB/ An 'EDS Types Chart' showing each subtype, inheritance patterns, and genetic basis and the protein involved (where known) can be found on page 187.

As we have seen in the previous two chapters, there are a range of conditions which can accompany hEDS, although, as yet, there is not enough data for them to become diagnostic criteria (while they are clearly associated with hEDS, they are not proven to be the *result* of hEDS). Some of these include sleep disturbance, fatigue, cardiovascular autonomic dysfunction (such as postural orthostatic tachycardia); anxiety and depression; mechanical and neuropathic bowel dysfunction (hernia, reflux, sluggish bowel and constipation), chronic bowel inflammation (inc. mast cell activation); myopia, astigmatism; poor response to local anaesthetic; pelvic floor weakness, rectal and/or uterine prolapse, chronic bladder inflammation (inc. mast cell activation); influence of progesterone - worsening musculoskeletal symptoms; also heavy and painful menstrual cycle; musculoskeletal and pelvic complications of pregnancy; and anxiety disorders, such as panic disorder and agoraphobia (Hakim A.J. (HMSA) 2017 / 2/Ehlers-Danlos Society 2017)



Assessing hyperextension in the hypermobile joint using an orthopaedic goniometer

For the patient, these other systemic manifestations may be more debilitating than the joint symptoms: often impairing functionality and quality of life, and should always be determined during clinical encounters. While they are not part of the diagnostic criteria, the presence of such systemic manifestations may prompt consideration of hEDS in the differential diagnosis (2/Ehlers-Danlos Society 2017).

Hypermobility Spectrum Disorder

Those individuals with hypermobility-related problems that meet the Beighton Score criteria (see **Box 3**, page 122), and who experience one or more of the secondary musculoskeletal manifestations described in **Box 3** point (i), but who do not meet the full criteria for hEDS or any other heritable disorder of connective tissue, should be given the diagnosis of hypermobility spectrum disorder (HSD) (Castori M. et al 2017 / Hakim A. 2017c). The subtype of HSD is then distinguished based on *which* joints are hypermobile (see **Box 4**).

Cautionary points - hEDS and HSD

- 1) As stressed in Chapter 1, musculoskeletal manifestations fall within a wide spectrum of severity (both in terms of the physical signs and severity of symptoms).
- 2) As with the hypermobile type of Ehlers-Danlos syndrome, individuals with HSD may also be affected by one or more of the comorbid disorders commonly associated with joint hypermobility. For example, postural orthostatic tachycardia, fatigue, functional gastrointestinal disorders, pelvic and bladder dysfunction etc (a larger list of examples can be found on page 42). The 2017 Classification recognises that they are present in some people with EDS, and HSD, and that health care professionals should be looking for them and treating them (Hakim HMSA 2017 / Castori M. et al 2017 / 3/Ehlers-Danlos Society).

3) Where a patient's diagnosis is placed within the spectrum of hypermobility related disorders (also see diagram and post-it note in **Box 5** at the end of this article) does not necessarily represent severity of symptoms experienced, it represents the *range* of symptoms experienced. Indeed, one person's hypermobile Ehlers-Danlos syndrome joint pain may be less severe than another's hypermobility spectrum disorder joint pain. The overall spectrum isn't strictly linear from least to most severe, it represents the *range* of symptoms seen in patients from single joint issues through to the 'syndrome of disorders' seen in hEDS .

Box 4.

When symptomatic joint hypermobility is experienced but the symptoms do not meet the diagnostic criteria for defined syndromes or disorders that feature hypermobility (such as those described on page 18) a patient may be given a diagnosis of 'hypermobility spectrum disorder (HSD).

HSD form part of a single continuous spectrum ranging from asymptomatic joint hypermobility at one end, through the various hypermobility spectrum disorders, and on to hEDS (Castori M. et al 2017).

Four different HSD may be identified:

Generalised (joint) hypermobility spectrum disorder (G-HSD):
Diagnosed where generalised joint hypermobility has been objectively assessed (e.g. by the Beighton score) plus one or more secondary musculoskeletal manifestations as identified in **Box 3 (point i)**, page 122. In these patients, the pattern and severity of the involvement of the musculoskeletal system should be carefully assessed in order to explore the possibility of a hEDS. In this category usually fall most patients with GJH and additional musculoskeletal manifestations but who do not meet the full diagnostic criteria for hEDS.

Peripheral (joint) hypermobility spectrum disorder (P-HSD):
Diagnosed where joint hypermobility is limited to hands and feet plus one or more secondary musculoskeletal manifestations identified in **Box 3(i)** page 122.

Localised (joint) hypermobility spectrum disorder (L-HSD):
Diagnosed where joint hypermobility is found at single joints or group of joints plus one or more secondary musculoskeletal manifestations regionally related to the hypermobile joint(s) (see **Box 3(i)**, page 122).

Historical (joint) hypermobility spectrum disorder (H-HSD):
Self-reported (historical) symptomatic generalised joint hypermobility (e.g., by the five-point questionnaire - see page 122, **Box 3, point (ii)** with negative Beighton score plus one or more secondary musculoskeletal manifestations as identified in **Box 3 (i)** page 122; in these cases, physical examination aimed at excluding the alternative diagnoses of G-HSD, P-HSD, and L-HSD as well as other rheumatologic conditions is mandatory. (All above: Castori M. 2017)

Type	Beighton score	Musculo-skeletal involvement	Notes
Asymptomatic GJH	Positive	Absent	
Asymptomatic PJH	Usually negative	Absent	JH typically limited to hands and/or feet
Asymptomatic LJH	Negative	Absent	JH typically limited to single joints or body parts
G-HSD	Positive	Present	
P-HSD	Usually negative	Present	JH typically limited to hands and/or feet
L-HSD	Negative	Present	JH typically limited to single joints or body parts
H-HSD	Negative	Present	Historical presence of JH
hEDS	Positive	Possible	

Source: 5/Ehlers-Danlos Society

'HEDS and HSD can be equal in severity, and both need similar management, validation and care.'

3/Ehlers-Danlos Society)

Is a formal diagnosis necessary?

The issue of whether or not a formal diagnosis is beneficial is a question often asked in patient forums.

As there is no definitive genetic test, and treatment is symptom based, doctors may advise against a formal diagnosis on the basis that, in many patients, symptoms can be effectively managed without a formal diagnosis. Some doctors also fear that 'labeling' patients with a diagnosis can lead to fear, anxiety, hypervigilism, and reluctance to exercise, resulting in a downward spiraling of health.

Where patients are left to find information themselves, and resort to searching the internet for answers or to creating their own (not necessarily beneficial) coping mechanisms, diagnosis without adequate support may be a risk. However, where a patient is signposted to appropriate educational sources after diagnosis (for example, to a charity whose information is accredited under NHS England's Information Standard), diagnosis may significantly reduce patient anxiety, be personally empowering and save NHS funding in terms of inappropriate tests and treatments.

Whether it's best to seek, or push for, a formal diagnosis is dependent on lots of factors, and is a very personal choice on the patients part. The

main thing is that a patient gets the right treatment/management advice. If they are getting the support they need, then a formal diagnosis might not be needed. If problems are not being addressed however, then, again, a diagnosis may help.

Some of the many reasons patients seek diagnosis are listed below:

- The confirmation and validation a diagnosis brings can often be a huge relief to patients, many who have been seeking answers for many years or who have been made to feel disbelieved.
- A diagnosis helps doctors know to look for related, co-morbid conditions frequently seen with EDS and to have an idea where to look when new symptoms occur.
- An official diagnosis can alert surgeons and therapists to changes in how they do carry out procedures and potential problems with anaesthesia and healing issues.
- A diagnosis can help a physiotherapist understand that alterations to "traditional or rehabilitative" physiotherapy may be required in order for it to work successfully for patients.
- The official diagnosis may be important to receive services from some specialists.
- A diagnosis can be helpful in order to obtain accommodations at work or for a child's requirements for PE or with sports activities to prevent injuries (ref: Supporting Pupils at school with Medical Conditions' Department of Education 2014).
- Outside of the UK, a diagnosis is helpful if you ever have to file for disability benefits (in the UK, disability benefits are based on symptoms rather than diagnosis).
- Some patients find that receiving diagnosis makes it easier to make appointments through private medical insurers.
- A diagnosis provides researchers with a better idea of the incidence of EDS.
- A diagnosis can help other family members understand what may be happening with them and help them to be diagnosed.
- A diagnosis can be important when advice is required to make informed decisions about family planning.

As discussed in Chapter 1, an international registry for EDS is planned and, over the next few years, it is likely that formal diagnosis will be encouraged in order to identify the global EDS population, raising

Diagnosing children

A baby is born with 300 bones which gradually fuse and strengthen as they progress through toddlerhood and childhood. By the time they reach adulthood they have 206. Due to this process

(ossification), it can be difficult for clinicians to tell whether or not a child has excessive laxity in comparison to other children of the same age. In some instances, it may be necessary to wait until they are around 6 years of age or older (Tinkle B. 2010) and, even at that point, unless there is clear evidence that a strong family history or genetic family link is involved, it is unlikely that a child will be given a formal HSD or hEDS diagnosis - any such diagnosis would usually be reserved until the child reaches their teens or adulthood and a clear history can be established.

In the meantime, if the child is struggling with pain and other problems, a child's GP can refer to a paediatrician or direct to physiotherapy. They may also benefit from a referral to podiatry and occupational therapy. Additional support and/or adaptations may also be needed in school.

Image by: Jim Gathany, CDC - under Creative Commons Licence



awareness, standards of care, clinical trials, facilitating scientific identification of the underlying genetic cause(s) of hEDS and, in the future, a treatment or cure.

It seems clear therefore that, although not right for everyone, formal diagnosis has value that should certainly be considered.

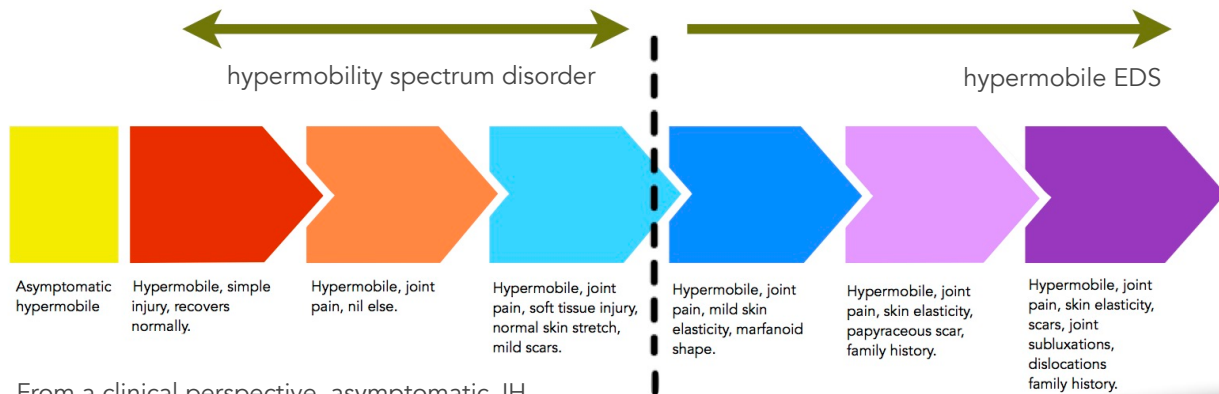
Concerns relating to differential diagnosis

If there is any concern that a patient may have a connective tissue disorder other than HSD or hEDS, they should be referred for expert opinion. Further assessment might include vascular imaging, ophthalmic review and genetic testing; geneticists are most adept at distinguishing between the types of Ehlers-Danlos syndrome, as well as differentiating EDS from the more than 200 other heritable connective tissue disorders (1/ Ehlers-Danlos Society 2017).

Management strategy and multi-disciplinary referral, physiotherapy; occupational therapy; joint protection; splinting, bracing, taping, compression-ware and orthotics; relief of severe pain, and using pain killers effectively; self management; cognitive behavioural therapy; complementary and physical therapies; making the most of your health care team; dental implications, and surgical issues are all discussed in the remainder of this chapter.

Box 5.

In the 2017 Classification/criteria, the International Consortium on Ehlers-Danlos syndrome attempt to better define the signs and symptoms of hEDS, and to address the key problem of distinguishing hEDS from symptomatic hypermobility.



From a clinical perspective, asymptomatic JH, HSDs, and hEDS can all be brought back to a single continuous spectrum ranging from isolated JH to hEDS passing through the various HSDs. As you travel left to right in the examples it becomes more and more possible to consider hypermobile Ehlers-Danlos syndrome as a diagnosis.

The 2017 Criteria

Above Image based on information provided by Dr A. Hakim in the HMSA Statement of Position February 2017

Important: Where a patient is placed within the spectrum does not necessarily represent the 'severity' of symptoms experienced, it represents the 'range' of symptoms experienced. Indeed, one person's hypermobile Ehlers-Danlos syndrome joint pain may be less severe than another's hypermobility spectrum disorder joint pain. The overall spectrum isn't strictly linear; from least to most severe, it represents the range of symptoms seen in patients; from single joint issues through to the 'syndrome of disorders' seen in hEDS. Symptoms of both disorders can be equal in severity and, importantly, need similar management, validation and care (see more on page 134). (Wicks D. and Hakim A.J. 2017)

Extract © Claire Smith

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