Hypermobile Ehlers-Danlos Syndrome

The Ehlers-Danlos Syndromes (EDS) are a heterogeneous group of heritable connective tissue disorders (HCTDs) characterised by joint hypermobility, skin hyperextensibility, and tissue fragility (Malfait et al. 2017).

At this point, the genetic cause for Hypermobile Ehlers-Danlos remains elusive. For that reason, among others, the criteria for diagnosing Hypermobile Ehlers-Danlos Syndrome were made more specific in 2017. The International Consortium on the Ehlers-Danlos Syndromes released a new nosology for all types of EDS, recognising 13 subtypes of EDS (where previously we had been working with 6 types).

During this process, the name for each type was modified slightly. EDS Hypermobility Type (which used to be known as EDS III, before it became EDS Hypermobility Type) has become Hypermobile EDS and shorted to hEDS.

A "clinical spectrum" of hypermobility has now also been identified and documented, and this ranges from Asymptomatic Joint Hypermobility through Generalised Hypermobility Spectrum Disorder, to Hypermobile EDS. See our handout on "Hypermobility Spectrum Disorders" for more information.

Beighton Score

Specifically for the purpose of diagnosing hEDS, the following cut-offs are recommended on the Beighton Score to ascertain whether someone has generalised joint hypermobility (GJH):

- ≥6 for pre-pubertal children and adolescents
- ≥5 for pubertal men and women up to the age of 50
- ≥4 for those >50 years of age

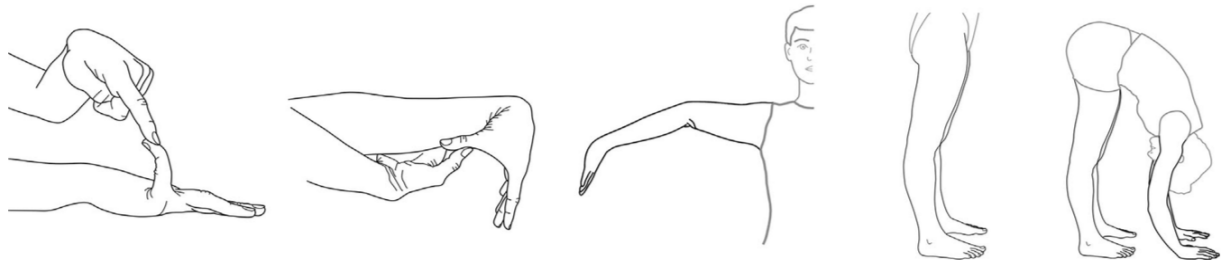

Figure 2. The Beighton scoring system. Each joint is measured using a goniometer and each side is scored independently as outlined [Juul-Kristensen et al., 2007]. (**A**) With the palm of the hand and forearm resting on a flat surface with the elbow flexed at 90°, if the metacarpal-phalangeal joint of the fifth finger can be hyperextended more than 90° with respect to the dorsum of the hand, it is considered positive, scoring 1 point. (**B**) With arms outstretched forward but hand pronated, if the thumb can be passively moved to touch the ipsilateral forearm it is considered positive scoring 1 point. (**C**) With the arms outstretched to the side and hand supine, if the elbow extends more than 10°, it is considered positive scoring 1 point. (**D**) While standing, with knees locked in genu recurvatum, if the knee extends more than 10°, it is considered positive scoring 1 point. (**E**) With knees locked straight and feet together, if the patient can bend forward to place the total palm of both hands flat on the floor just in front of the feet, it is considered positive scoring 1 point. The total possible score is 9. *Figure courtesy of Dr. Juul-Kirstensen.*

There is recognition that people with acquired joint limitations e.g. from surgery, wheelchair use, amputation etc. may not score accurately on the Beighton Scale. If the individual is 1 point under the cut-off for their age/life stage, The Five-Point Questionnaire can be used to help diagnose GJH.

The Five-Point Questionnaire. Adapted From [Grahame and Hakim, 2003]

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

A "yes" answer to two or more questions suggests joint hypermobility with 80–85% sensitivity and 80–90% specificity

Diagnosis of Hypermobile EDS (hEDS)

A diagnosis of hEDS should be assigned only in those who meet ALL of the criteria below. It's important to be strict with assessment of criterion in order to help reduce heterogeneity within the diagnostic category, and to help future efforts to discover underlying genetic causes for the condition (Malfait et al. 2017).

A diagnosis of hEDS requires the patients' symptoms meet Criteria 1 AND Criteria 2 AND Criteria 3.

Criterion 1: Generalised Joint Hypermobility

- As diagnosed using the Beighton Score (+/- The Five-Point Questionnaire)

Criterion 2: Two or More Among the following features (A-C) MUST be present (e.g. A + B; A + C; B + C; A + B + C)

Feature A: (A total of 5 must be present)

- Unusually soft or velvety skin
- Mild skin hyperextensibility
- 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
- Bilateral piezogenic papules of the heel
- Recurrent or multiple abdominal hernia(s) (e.g., umbilical, inguinal, crural)
- Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known pre- disposing medical condition
- Dental crowding and high or narrow palate
- 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

Criterion 2: Feature A continued

- Arm span-to-height ≥1.05
- Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- Aortic root dilatation with Z-score > +2

Criterion 2: Feature B: Positive Family History with one or more first-degree relatives independently meeting the current diagnostic criteria for hEDS.

Criterion 2: Feature C: Musculoskeletal complications (must have at least one)

- Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- Chronic, widespread pain for ≥3 months
- Recurrent joint dislocations or frank 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 two or more sites not related to trauma

Criterion 3: All the Following Prerequisites MUST be met

- Absence of unusual skin fragility, which should prompt consideration of other types of EDS

- 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.

- 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., myopathic EDS, Bethlem myopathy), other HCTD (e.g., other types of EDS, Loeys–Dietz syndrome, Marfan syndrome), and skeletal dysplasias (e.g., OI). Exclusion of these considerations may be based upon history, physical examination, and/or molecular genetic testing, as indicated.

There are more explanatory notes available in the original article "The 2017 International Classification of the Ehlers-Danlos Syndromes" (Malfait et al. 2017)

A number of other symptoms & syndromes have been found to be associated with hEDS. While these are not included in the diagnostic criteria, the presence of co-morbidities is not uncommon.

These can include:

- Sleep disturbance
- Fatigue
- Postural Orthostatic Tachycardia Syndrome
- Functional Gastrointestinal disorders
- Dysautonomia
- Anxiety
- Depression

What does the change in diagnostic criteria mean for the person living with a hypermobility condition?

Theoretically, the change in the criteria has affected some patients to the point that they no longer meet the criteria for Hypermobile EDS. This has been concerning for a lot of people with hypermobility conditions, and understandably so. They have just had the diagnosis they have been working with for X number of years ''taken away from them'', if you will. While the Ehlers-Danlos Society has tried to placate concerned patients by saying their diagnosis won't be ''taken away from them'', in the grand scheme of things, this goes against the whole point of narrowing down the diagnostic criteria in the first place. The Consortium have declared that we need to reduce the heterogeneity within the hEDS diagnostic category, and that can only happen if those who are already diagnosed, as well as those diagnosed in the future, are in fact given the correct diagnosis. The Ehlers-Danlos Society (2017) have since stated: ''If someone was diagnosed with hEDS before the 2017 criteria, there's no cause to seek a new diagnosis unless they decide to participate in new research or need to be reassessed for some other reason''.

Typically, those who don't meet the criteria for hEDS anymore, meet the criteria for one of the Hypermobility Spectrum Disorder subtypes, but this also needs to be assessed.

Our approach at Hypermobility Connect is to try to encourage people to see this change of diagnosis in an alternate way. This change in criteria means that people have now received a far more accurate diagnosis. They are still living with a disorder that sits on the Hypermobility Spectrum, which can be *just as disabling, painful, and frustrating*, it's just no longer considered to be Hypermobile EDS at this point in time.

In a lot of ways, this change in diagnosis means nothing other than a label change. The management of hEDS and G-HSD is essentially the same. Management is largely symptom and prevention focused in both conditions.

What we need to be aware of is the idea of ''downgrading'' which seems prevalent in the EDS/Hypermobility Community. There is a belief that hEDS is ''worse'' than HSD. And because patients have often struggled to have their condition recognised and their pain validated, many have a tendency to want the diagnosis that ''sounds the worst'' and therefore attracts more empathy & sympathy from others. This is where education has to come in to play. A correct diagnosis is always better than the most severe sounding, but we have a long way to go in helping people to see it this way. It's a big change.

What it looks like

Hypermobile EDS is a pervasive condition. The criterion in the diagnostic nosology above cover the following body areas or systems:

MUSCULOSKELETAL Joint hypermobility Joint pain Joint instability/dislocations
SKIN Hyperextensibility Scarring Abnormal stretch marks
INTERNAL ORGAN STRUCTURES Hernias Prolapses
DENTAL High palate Crowding of teeth
CARDIAC Mitral Valve Prolapse Aortic Root Dilation

Plus the non-diagnostic related co-morbidities

- Sleep
- Fatigue
- Mood
- Gastrointestinal
- Autonomic System

Patients with hEDS present in a myriad of ways. There is so much diversity within the hEDS diagnostic criteria, which means that no two people living with Hypermobile EDS will present the same way.

The most common reason for someone to present for diagnosis in this hEDS sub-type of Ehlers-Danlos Syndrome is joint hypermobility, joint instability and chronic pain. They also often experience debilitating fatigue.

A person living with significant joint hypermobility, pain and fatigue will often be struggling with their life roles (e.g. parenting, working, schooling) as well as many of the tasks within these roles (e.g. cooking, cleaning, studying, driving, looking after children). Some will have made helpful adaptations (e.g. found ways around difficult tasks), others will have found unhealthy ones (e.g. avoiding leaving the house), while others may not have made any changes at all.

If the person has been experiencing symptoms for some time, they will often present using aids, braces, splints and may or may not be taking considerable doses of pain medication. Most of these things are necessary in the life of person severely affected by hEDS, especially when they haven't been managed well (or at all).

More often than not, they will *not* have had access to specialist services for hEDS. There are no adult genetic specialists services in Australia for the Ehlers-Danlos Syndromes. There is a scattering of private practice health professionals who are knowledgeable enough to be able to work with patients with these conditions. Typically, what they have learned in this area of practice is from treating patients with hypermobility, through trial and error.

At this time, the database that we have of these practitioners is not available to the public. However, names of practitioners can be provided on request, if any are located in the patient's area and have agreed for their details to be shared.

The serious lack of resources is why we desperately need to educate medical & health professionals about Hypermobile EDS and the Hypermobility Spectrum Disorders. Specifically, how to diagnose (for medical practitioners) and how to help someone living with hypermobility to learn to manage their condition (allied health). At the moment, patients are generally learning how to do this from each other, in patient forums and Facebook groups, which means their management plans are not tailored to their individual needs, and more often than not are not appropriate for the individual. Patients are not getting the best healthcare possible because of lack of health professional education in the area of hypermobility conditions.

If you are a medical or health professional and this document has found its way into your hands, we would like to invite you to **join our database:** www.hypermobilityconnect.com/join-our-list

If you're a person living with hypermobility **join our community mailing list** www.hypermobilityconnect.com/mailing-list-sign

HYPERMOBILITY CONNECT

Recognise Educate Accept

Information compiled by
MICHELLE O'SULLIVAN BAPPSC(OT) GRAD CERT LOSS, GRIEF & TRAUMA COUNSELLING, BASED ON:

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THE 2017 INTERNATIONAL CLASSIFICATION OF THE EHLERS–DANLOS SYNDROMES. AM J MED GENET PART C SEMIN MED GENET 175C:8–26.
Hypermobile Ehlers-Danlos syndrome (hEDS) vs. Hypermobility Spectrum Disorders (HSD): What's the Difference?
https://ehlers-danlos.com/wp-content/uploads/hEDSvHSD.pdf