Hypermobility

Hypermobility spectrum disorders (HSD) and hypermobile Ehlers-Danlos syndrome (hEDS) and are the most common symptomatic joint hypermobility conditions seen in family medicine.

Bottomline: Neither HSD nor hEDS requires a genetic assessment however hEDS has additional screening recommendations so it is important to differentiate between them.

What is hypermobility?

Joint hypermobility means that a person's joints have a greater range of motion than is expected or is typical. Hypermobility may be localized to a single joint, may only affect peripheral joints of the hands/feet, or may be generalized. Hypermobility that does not cause pain or other symptoms ie. asymptomatic joint hypermobility, does not need to be treated.

Issues with hypermobility arise when the joints lack stability which can result in subluxations, dislocations, sprains, and other injuries. This can cause both short-term and long-term pain, significantly impacting daily activities.

Hypermobility spectrum disorder

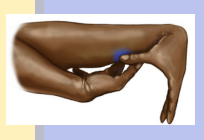
Hypermobility spectrum disorder (HSD) refers to the wide variation in both type and severity of symptoms people experience. Symptoms can include joint instability, mast cell activation syndrome, pain, fatigue, gastrointestinal issues, dysautonomia, headaches and anxiety. Individuals may experience all or some of these, and each to varying degrees.

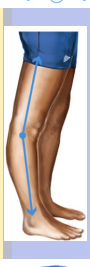
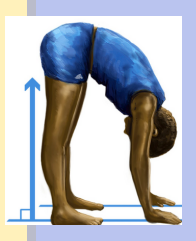
How is joint hypermobility identified?

One way to assess generalized joint hypermobility is with the Beighton Score, which measures joint hypermobility on a 9-point scale. One point is given for each item on the scale. A positive Beighton score is any score greater than or equal to 5/9 points in adults, 6/9 points in children (before puberty), and 4/9 points in adults over age 50.

Beighton Score

Joint assessment	Points	
	Right	Left
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	1	1
With arms outstretched forward but hand pronated, if the thumb can be passively moved to touch the ipsilateral forearm	1	1
With the arms outstretched to the side and hand supine, if the elbow extends more than 10°	1	1
While standing, with knees locked in genu recurvatum, if the knee extends more than 10°	1	1
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	1	1
Total score A positive Beighton score is any score greater than or equal to 5/9 points in adults, 6/9 points in children (before puberty), and 4/9 points in adults over age 50.		

images courtesy of EDS Society

How is HSD managed treated?

Management of HSD is supportive care likely requiring a multidisciplinary healthcare team. Find local specialists here.

What about the genetics of HSD?

A genetic etiology for HSD is not known. Genetic testing is not available. A **genetic assessment is not indicated.** Family members do not require assessment.

How does hypermobility spectrum disorder differ from hypermobile Ehlers-Danlos syndrome?

Check out the next page

Hypermobility

Bottomline: Neither hypermobile spectrum disorder nor hypermobile Ehlers-Danlos syndrome requires a genetic assessment however hEDS has additional screening recommendations so it is important to differentiate between them.

What is hypermobile Ehlers-Danlos syndrome?

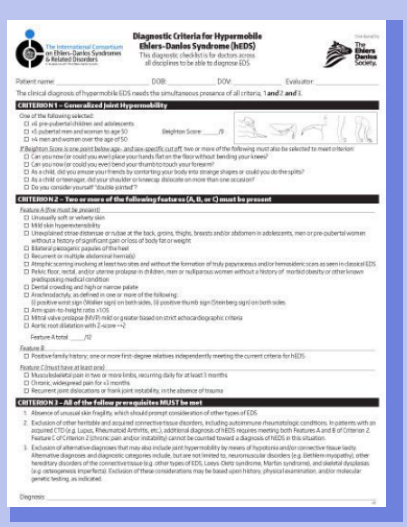
Ehlers-Danlos syndrome (EDS) is a group of connective tissue disorders caused by abnormalities in collagen production, processing or structure. Hypermobile Ehlers-Danlos is one of at least 13 EDS subtypes and is the most common.

How is hypermobile EDS diagnosed?

Hypermobile EDS (hEDS) is a **clinical diagnosis**. There are no confirmatory tests available.

A diagnostic check list can be found here and on the Ehlers-Danlos Society website.

An hEDS diagnosis is based on a person meeting three criteria: **(1)** Generalized joint hypermobility (assessed using the Beighton score); **(2)** Two or more manifestations of a connective tissue disorder e.g. a positive family history, recurrent joint dislocations, soft velvety skin; **(3)** absence of skin fragility (if present consider other subtypes of EDS) **and** exclusion of alternative diagnosis, heritable or acquired e.g. lupus, arthritis, Marfan syndrome.



Hypermobile EDS diagnostic checklist

Children and adolescents with generalized joint hypermobility can be assessed using the pediatric diagnostic framework.

Are there additional tests I should consider?

If considering a diagnosis of hEDS, an echocardiogram should be offered to rule out mitral valve prolapse and aortic root dilation.

What about genetics and hEDS?

The genetic cause of hEDS has not been identified and genetic testing is not available. A genetic assessment is not indicated unless another condition is suspected (e.g. another subtype of EDS, Marfan syndrome).

hEDS follows an autosomal dominant pattern of inheritance and first-degree relatives have a 50% chance to also have the condition.

Family history is an important part of the clinical diagnosis of hEDS. Asking about joint hypermobility, musculoskeletal symptoms, aneurysms, and genetic conditions is essential.

How is hEDS managed?

Offer an echocardiogram to rule out mitral valve prolapse and aortic root dilation.

hEDS is management is supportive, addressing the symptoms a person is experiencing. Because of the variety of symptoms, a multidisciplinary healthcare team may be required. Find local specialists here.

Further details on management of hEDS can be found on the Ehlers-Danlos Society site and in Yew et al. 2021 Am Fam Physician. [OpenAccess]

How do I know this is not another subtype of EDS?

Features of other EDS subtypes include: skin fragility with extensive atrophic scarring, very stretchy skin with velvety or doughy texture, arterial fragility with aneurysm/dissection/rupture, organ fragility and rupture.

If any of these features are present, referral to genetics is suggested and genetic testing to confirm a diagnosis may be offered.

How does hypermobility spectrum disorder differ from hypermobile Ehlers-Danlos syndrome?

Hypermobility spectrum disorder (HSD) does not meet diagnostic diagnostic criterion 2 and shows no signs/symptoms of skin fragility or an alternate diagnoses such as lupus, Marfan syndrome etc. No additional health screening (e.g. echocardiogram) for HSD is indicated.

Resources and references

The Ehlers-Danlos Society has information and support for the public and clinicians.

The Ehlers-Danlos Society

Yew KS, Kamps-Schmitt KA, Borge R. Hypermobile Ehlers-Danlos Syndrome and Hypermobility Spectrum Disorders. Am Fam Physician. 2021 Apr 15;103(8):481-492. [OpenAccess]