Joint Hypermobility: Diagnosis for Non-Specialists

This article describes how physician generalists can make useful working diagnoses of most patients with hypermobility syndromes, and proceed with effective treatment for them. It is primarily written for physicians. Individuals who suspect they may have a hypermobility condition may wish to read it through, and have their primary care practitioners use it to make a diagnosis.

Alan Spanos MD
Clinical Associate
UNC School of Medicine
Chapel Hill, NC
March 2019

Background

Joint hypermobility syndromes are a heterogeneous group of inherited conditions, with the common feature of lax or "hypermobile" joints. Joint laxity, looseness and hypermobility are synonyms for joints that move through wider ranges than normal, either actively or passively. Joint hypermobility syndromes typically present in childhood or adolescence. They are not rare: one ore more children in most high schools probably has such a condition. But most are either undiagnosed, or if diagnosed, are untreated. Often, these patients are dismissed with false platitudes such as, "It's only a name," "It's congenital so there's nothing you can do about it", or misdiagnosed as having fibromyalgia, or a psychiatric condition. These false diagnoses lead to unhelpful, and often harmful, treatments.

The best known hypermobility syndromes are the Ehlers Danlos Syndromes, and the much-less-common Marfan Syndrome. Since they are inherited, their diagnosis has been largely left to geneticists. But geneticists' appointments are routinely backlogged by many months, or even *several years*. So even when pediatricians or family doctors suspect a hypermobility condition, these patients remain undiagnosed and untreated - at just the time when their condition is getting severe enough to merit good medical attention.

An international body of experts recently spent several years reviewing diagnosis and treatment of these conditions.[1] One of their conclusions was that some of the most common hypermobility conditions *can be diagnosed by nonspecialists and do not require genetic tests for confirmation.* As for treatment, most of this is well known to medical generalists when applied to other conditions, and can be applied to hypermobility disorders to the great benefit of patients. Even if a referral is made to a geneticist, the patient can be given a *presumptive* diagnosis in the meantime, so they can be treated, rather than having their condition worsen through neglect or inappropriate treatment while they wait for the appointment.

This article deals only with *diagnosis* of hypermobility syndromes. Their *treatment* is covered in the accompanying article to this one, at the website *AlanSpanosMD.com*, entitled *Medical Care of Patients with a Hypermobility Disorder.*

How many hypermobility conditions?

In March 2017, an international group of experts redefined many of the inherited hypermobility conditions. They called the most common of these conditions "hypermobile spectrum disorder (HSD," and defined its diagnostic criteria so that it can be diagnosed by nonspecialists, without requiring confimatory genetic tests.

The spectrum referred also includes a redefined "hypermobile-type Ehlers Danlos Syndrome (hEDS)." This can be envisaged as on the right-hand end of the hypermobility spectrum. The generalist need not be concerned with the distinction between hEDS and HSD, since it's an artificial cut-point on a single spectrum. Moreover, *treatment is the same for hEDS as for HSD.* A generalist can make a diagnosis of HSD with confidence, but add "possible hEDS" to cover a possible future revision by a geneticist. If that occurs, it will not in no way invalidate the benefits of the treatment already given for "HSD."

In this article, I will refer to "HSD/hEDS" as the condition in question. I regret the need for this cumbersome acronym.. The rest of this article deals with patients identified as having HSD/hEDS using the new, 2017 criteria.

ICD-10 diagnostic codes include only one code for all kinds of Ehlers Danlos Syndromes, i.e. Q79.6. There is no new code for the new label of hypermobility spectrum disorder (HSD). For the time being, Q79.6 is the appropriate code to use for hypermobility spectrum disorder as well as any Ehlers Danlos Syndrome. Other, vaguer codes exist but are less well understood by insurance companies and others such as disability assessors.

~

The Joint Hypermobility Spectrum

In its new consensus documents, the International Consortium formalized the "hypermobility spectrum" concept, as a practical clinical tool.[2] This is depicted below.

Joint Hypermobility	**Hypermobility Spectrum Disorder**	**Hypermobile Ehlers Danlos Syndrome**
"JH"	**"HSD"**	**"hEDS"**
Hypermobility without problems	Hypermobility with pain and/or musculoskeletal complications	Hypermobility with other connective-tissue features

This spectrum moves from:

people with lax joints but *no problems* from them (JH), through
people with *problematic* loose joints (HSD), to
people with "hypermobile type Ehlers Danlos Syndrome" or hEDS.

There are two main reasons for locating these conditions "on a spectrum." One is that *more than one of these conditions is commonly found among different members of the same family.*. The second reason is that these conditions *grade into one another*. The distinction between adjacent ones is "fuzzy", and individuals may shift from one category to another over time. Genetic tests at present do not identify, let alone discriminate among, these categories.

Note that the spectrum is *not* one of increasingly lax joints: an asymptomatic person on the left may have joints looser than someone with hEDS on the far right. Rather, it is a spectrum of features *added on to the joint hypermobility*, that change the character of the overall condition.

Moving through the hypermobility spectrum

Joint looseness that is *localized* to one or a few joints, is generally due to obvious, non-inherited causes, notably injuries and arthritis. However, when *a number of joints in all limbs*, and perhaps also in the spine, are unusually lax, this condition has nearly always been present since early childhood, and is inherited. A crucial point is that many people with loose joints are not troubled by them much, if at all. They and their families may be aware of their unusual suppleness. As children they may have shown off the contortions they could do because of this. Many have excelled in gymnastics or ballet, or have found they could do yoga postures with ease, that were quite impossible for other people in the yoga class. Many such people will have little or no significant long term trouble from their loose joints. These people clearly do not have a disease or a syndrome. They are on the left end of the spectrum (see above), in that they have *hypermobile joints that are <u>not</u> causing trouble*, i.e. "joint hypermobility" or JH.

Moving to the middle of the spectrum, we have people with *loose joints that <u>are</u> causing chronic problems*, mainly pain, and/or joint displacements (subluxations and/or dislocations). This condition is now named "hypermobile spectrum disorder" or HSD. The pain may be localized to specific joints, or be more diffuse – hence the frequent misdiagnosis of "fibromyalgia." Patients often say they can feel some of their loose joints moving painfully out of place. They may sometimes have to manually put them back in place, especially fingers and toes. They may complain bitterly of unpleasant cracking feelings or sounds, with a feeling that the joints are moving loosely, especially in the neck. These symptoms commonly begin in childhood or adolescence. Often the teenager or adult reports that they were taken to the doctor many times as children for limb pains described as "growing pains." They have "hypermobile spectrum disorder" or HSD. The prevalence of HSD is not known precisely, but it is clearly rather common: most high schools seem to have one or several individuals with HSD. If its prevalence is at least one in five hundred, as seems likely, then at least twenty thousand people in North Carolina have HSD. *This is by far the most common hypermobility condition that generalists will see, and they can diagnose and treat it.*

Further to the right of HSD on the hypermobility spectrum is "hypermobile type Ehlers Danlos Syndrome" or hEDS (note the small "h"). This label is applied to people with hypermobile joints who *have a number of features of abnormal connective tissue <u>outside</u> the joints.* The other features may include soft and somewhat "stretchy" skin, slow healing of wounds and incisions, striae that are unexplained by weight change or pregnancy, abdominal hernias, and several others. The hEDS diagnosis requires presence of a number of these features, using a rather complex algorithm.

However, there is a continuum between people with HSD and hEDS, and the distinction between them is fuzzy, controversial and provisional. The committee that proposed this distinction admitted its artificiality, and planned to revise it every two years. (However, as of September 2021 they had not done so.) The distinction is of little or no relevance to treatment, and it is therefore unnecessary for the generalist to master it. *A diagnosis of "hypermobility spectrum disorder (HSD), possible hEDS" can be made by a generalist and is sufficient to direct treatment.* Further diagnostic refinement by a geneticist may be ordered, but is optional, and treatment should not wait on this.[3]

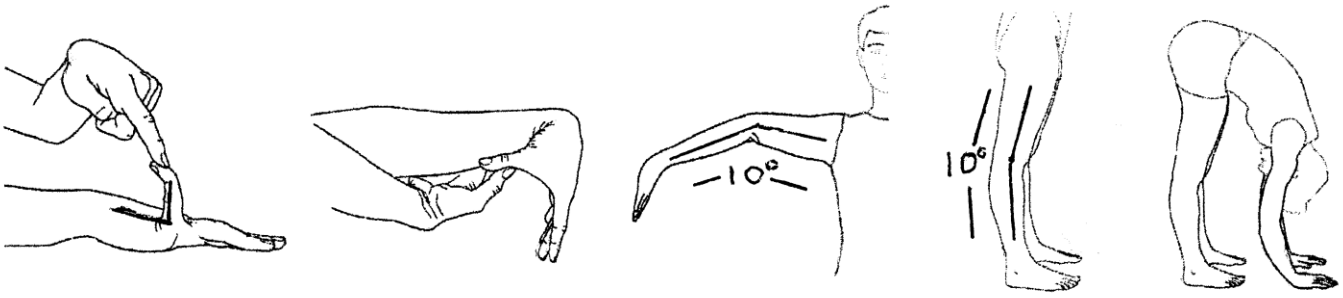
~

Joint Hypermobility: Diagnostic Procedure for the Generalist

Step 1. Does the patient have generalized joint hypermobility (GJH)?

The Beighton scale:

The Beighton scale has been used for several decades as the conventional tool for scoring joint hypermobility. Physical therapists are familiar with it, and many patients have discovered it online and checked it on themselves. It is appropriate for postpubertal adolescents and adults.[4]



With hand palm-down, pull little finger back. Is the angle between the base of the finger and the back of the hand at least 90°?[5]	Can the thumb be pulled to touch the volar (inner) side of the forearm?	With arm outstretched to side, and palm facing outward, does the elbow extend at least 10°?	Standing with knees locked, is the knee bent at least 10°?	With knees locked and feet together, can the whole palms of both hands be placed flat on the floor?
One point each side.	**One point each side.**	**One point each side.**	**One point each side.**	**One point.**

Interpretation:
A score of 5 or over identifies "generalized joint hypermobility" (GJH). But joints may stiffen slowly during adulthood, so a score less than 5 in an adult who is convinced they are "double-jointed" should prompt use of the Five Point Questionnaire, below. Conversely, in children before puberty, a score of 5 may be nonspecific; in these patients, a score of 6 is preferred to indicate GJH.

The Five-Point Questionnaire[6]

This questionnaire has been in use for over a decade. It can override a <u>negative</u> Beighton score if the physician thinks the history nonetheless suggests hypermobility through childhood and adolescence, in a patient who may have "stiffened up" through adulthood.

1 Can you (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 doing contortions, 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"?

Interpretation:
If the patient answers "Yes" to two questions, this suggests generalized joint hypermobility with ≥80% sensitivity and ≥80% specificity; success rate increases with each "yes" answer beyond two.

If the patient scores positive on either of the above instruments, then generalized joint hypermobility may be presumed. Go to next page.

Joint Hypermobility: Diagnostic Procedure for the Generalist

Step 2. Is there "musculoskeletal involvement" in addition to joint hypermobility?

Any of the following constitute musculoskeletal involvement, and count towards a diagnosis of HSD. More features add more confidence:

- **Chronic pain** In and around loose joints.
- **Injuries** Dislocations (e.g. shoulders), sprains (e.g. ankles), subluxations (e.g. patellae); cartilage damage (e.g. labral tear of hip or shoulder).
- **Joint damage** Osteoarthritis at an early age, in joints that are hypermobile, or have a history of hypermobility in the past.
- **Miscellaneous** Flat feet, misaligned bones in the elbow and big toe, mild-to-moderate scoliosis, kyphosis of the upper spine, excessive lumbar lordosis, and defective proprioception. This is a mixed bag of largely non-quantified items of dubious significance. It is probably rare that a patient be identified as having HSD, unless they have one of the first three criteria above (pain, injuries, joint damage).

In practice, it is the presence of symptomatic problems from any of the above that mainly justifies calling the patient's condition a medical syndrome (hypermobility spectrum disorder, HSD) rather than merely a constitutional feature of no medical importance (joint hypermobility, JH). Clearly then, an individual's condition may shift over time, as symptoms become sufficiently intrusive to constitute an ongoing problem. For people in the HSD category, this generally happens in childhood or early adolescence. A typical story would be of the child who enjoyed showing off contortions in elementary school, but gradually accumulates recurrent sprains and perhaps undiagnosed limb or back pain, so that by the time they are in high school, they are unable to keep up with their peers in sports. They are often told they have "growing pains".

If the patient is troubled by chronic, or recurrent, problems as above, then "hypermobility spectrum disorder (HSD) or hypermobile Ehlers Danlos Syndrome (hEDS)" is the working diagnosis. Go to the next page.

~

Joint Hypermobility: Diagnostic Procedure for the Generalist

Step 3. Are there red flags for a dangerous inherited condition?

Several inherited conditions may *include* all the features of HSD/hEDS, but have *other additional features*. These conditions are very much more rare than HSD or hEDS, and many doctors will never see a case during their careers. The most well known are vascular-type Ehlers Danlos Syndrome (vEDS), and Marfan Syndrome. A short checklist of questions, described below, will usually detect red flags for these conditions.

1. Vascular Ehlers Danlos Syndrome (vEDS)[7]

This can present with generalized or regional hypermobility, but it also carries risks of possibly-fatal complications. In vEDS, there is a high risk of arterial rupture from an aneurysm or dissection, or rupture or bleeding of an abdominal viscus. The following questions screen for the very rare patient who may have vEDS.

Has anyone in your family, including distant relatives:

- collapsed and died suddenly at under 40 years of age?
- had an aneurysm diagnosed at under 40 years of age?
- had an unexplained rupture of the sigmoid colon?
- had a spontaneous pneumothorax?
- had rupture of the uterus, or catastrophic bleeding, in childbirth?
- been diagnosed with "vascular EDS" or "EDS Type 4?"
- Have you yourself had an aneurysm, colonic rupture, pneumothorax, or a diagnosis of Vascular EDS?

If the answer to any one of the above is "yes", then it would be prudent to call a geneticist and ask if this mandates an urgent appointment. The specialist may also suggest imaging studies to seek for aneurysms and to rule out dilatation of the proximal aorta.

---

2. Marfan Syndrome[8]

Patients with this condition may be diagnosed at any age. As well as having joint hypermobility, they are typically tall and thin with pectus excavatum or carinatum, and/or scoliosis, and various other musculoskeletal features. *However, the cardinal features are aortic root dilatation, and ectopia lentis (dislocated lenses).*

There are no firm guidelines on which patients should be screened for Marfan Syndrome, so any of the above features could prompt a referral to a geneticist, or a phone discussion with one. Also, it may be prudent to request echogram measurements of the aortic root in hypermobile patients who are:

- tall and thin
- have chest wall deformities
- have scoliosis
- have a history of lens abnormalities.

There are several, very much rarer conditions, that may present as HSD, but their existence need not prevent a *working or presumptive diagnosis.*

If vEDS and Marfan Syndrome are not suggested as above, then the patient's working diagnosis is "Hypermobility Spectrum Disorder (HSD), possible hEDS." Go to next page.

Hypermobility Spectrum Disorder (HSD): Clinical Implications

The treatment is what matters, not the label.

The distinction between HSD and hEDS is vague, artificial, and likely to change as the International Consortium that sets the criteria reassesses the issue every two years. *The patient and doctor need not be concerned about the label. What matters is that the treatment is just the same for HSD and hEDS.* The ICDM code for EDS is Q79.6 (as of March 2019). For the time being, this code may be used for HSD too.

We have no genetic tests to confirm HSD and hEDS

The hypermobility spectrum conditions, including hEDS, cannot, as yet, be identified by any genetic tests.

Prognosis is uncertain

Symptoms of HSD/hEDS may improve, worsen, stay the same, or go through long exacerbations and remissions. We have no tests that predict the long term outcome.

Inheritance pattern is highly uncertain

Usually there is more than one person in a family who is clearly "on the spectrum". But it is difficult to predict if an infant will develop HSD/hEDS, because joint laxity is normal in the first year or two of life. Delayed walking, unusual postures and repeated falling are of concern, and should be assessed by a pediatrician or pediatric physical therapist. Children with recurrent limb pains, often labelled "growing pains," often have HSD/hEDS.

HSD/hEDS is a multisystem condition

For unknown reasons, people with loose painful joints commonly have various other problems. These should be asked about, and treated individually. The commonest are:

1. **Abdominal pains**, often with constipation, resembling irritable bowel syndrome, and often with gastro-esophageal reflux.[9]
2. **Sleep disorder**, most commonly hypersomnia, so patients sleep longer than normal. They may also need to nap every afternoon. Disrupted sleep is also common.
3. **Rapid exhaustion** with mild or moderate exertion, of unknown mechanism. True muscle weakness may also be present.
4. **Orthostatic intolerance**,[10] causing patients to be wary of standing in long supermarket lines etc. Patients may have syncope unless they sit or lie down when they feel bad in such situations. Cardiovascular parameters may, or may not, be disturbed.
5. **Neuropathic pain**[11] of various sorts and various distributions.
6. **Headaches**, which vary from typical migraines, to headaches originating in the cervical joints.[11,12]
7. **Anxiety**, often beginning in early childhood, which may be severe.[13]
8. **Easy bruising**, but without abnormal clotting studies.[14]
9. **Slow healing** of wounds and surgical incisions.[14]
10. **Poor response to local analgesics**, so patients need large and repeated doses for dentistry etc.[14]
11. **Clumsiness**, generally dating from early childhood. It appears to be due to defective proprioception.[14]
12. **Dysmenorrhea**, often starting soon after menarche.[14]

Usually only a few of the above are problematic enough to require medical treatment. The article on my website, *Medical Care of Patients with a Hypermobility Disorder*, summarizes treatments, most of which can be managed by the patient's primary care practitioner.[15]

Alan Spanos MD
March 2019

Dr Spanos is in Chapel Hill, NC.
Other articles are on his website at *www.AlanSpanosMD.com.*

References

[1] The International Consortium on Ehlers-Danlos Syndromes and Related Disorders published its findings in a series of articles in a special issue of the American Journal of Medical Genetics in 2017, i.e. Am J Med Genet Part C Semin Med Genet 175C.

[2] A Framework for the Classification of Joint Hypermobility and Related Conditions. Castori M, Tinkle B et al. 2017. Am J Med Genet Part C Semin Med Genet 175C.

[3] See endnote 2, pp 154-156 for a detailed discussion of the proposed Hypermobility Spectrum Disorders. I have omitted items that do not seem necessary for diagnosis by the medical generalist.

[4] Modified from Malfait F, Francomano C et al. 2017. The 2017 International Classification of the Ehlers–Danlos Syndromes. Am J Med Genet Part C Semin Med Genet 175C.

[5] Note that hyperextension of the terminal phalanx, as illustrated in this diagram, is irrelevant: the angle measured is between the metacarpal and the proximal phalanx.

[6] See page 17 of article referenced in endnote 4. The questionnaire was originally presented by Hakim AJ and Graham R. 2003. A simple questionnaire to detect hypermobility. Int J Clin Pract 57:163-166. It has been used in many clinics since then.

[7] Details available at the website of the Ehlers Danlos Society, *www.ehlers-danlos.com*.

[8] The website of the Marfan Foundation at *marfan.org* has useful literature for patients and their physicians on how to proceed if Marfan Syndrome is suspected.

[9] Fikree A, Chelimsky G, Collins H, Kovacic K, Aziz Q. 2017. Gastrointestinal involvement in the Ehlers–Danlos syndromes. Am J Med Genet Part C Semin Med Genet 175C.

[10] Hakim A, O'Callaghan C, et al. 2017. Cardiovascular autonomic dysfunction in Ehlers–Danlos syndrome - Hypermobile type. Am J Med Genet Part C Semin Med Genet 175C.

[11] Chopra P, Tinkle B, et al. 2017. Pain management in the Ehlers–Danlos syndromes. Am J Med Genet Part C Semin Med Genet175C.

[12] Henderson Sr. FC, Austin C, et al. 2017. Neurological and spinal manifestations of the Ehlers–Danlos syndromes. Am J Med Genet Part C Semin Med Genet 175C.

[13] Bulbena A, Baeza-Velasco C, et al. 2017. Psychiatric and psychological aspects in the Ehlers–Danlos syndromes. Am J Med Genet Part C Semin Med Genet 175C.

[14] Tinkle B, Castori M, et al. 2017. Hypermobile Ehlers–Danlos syndrome (a.k.a. Ehlers–Danlos syndrome Type III and Ehlers–Danlos syndrome hypermobility type): Clinical description and natural history. Am J Med Genet Part C Semin Med Genet 175C.

[15] Article "Medical Care of Patients with a Hypermobility Disorder," at *www.AlanSpanosMD.com*.