TAKEAWAYS

What Should Providers Look For?

- History of joint dislocations or subluxations
- Family history of hypermobility or connective tissue issues
- Chronic unexplained pain or fatigue
- Easy bruising without bleeding disorders
- Gastrointestinal dysmotility (e.g., chronic constipation, nausea)
- Autoimmunity, autoinflammation, or mast cell activation disease
- Dysautonomia / POTS
- Symptoms that are multisystemic, difficult to localize, or have unclear origins

Fighting Misconceptions

- hEDS is not rare; it's underdiagnosed
- Joint hypermobility may decrease with age or be affected by prior injuries or surgeries; its absence on exam does not rule out a diagnosis of hEDS
- Chronic pain, fatigue, and dysautonomia are genuine, disabling symptoms, not psychological in origin

How Providers Can Support Patients

- Validate symptoms — patients with EDS often face years of dismissal
- Coordinate care across specialties (PT, cardiology, GI, allergy, immunology, genetics, pain management)
- Consider referring to EDS-experienced specialists
- Provide school/work accommodations when needed

LEARN MORE

References:

1. Phenotypic Clusters and Multimorbidity in Hypermobile Ehlers-Danlos Syndrome. Petrucci, Taylor et al.
2. Hypermobile Ehlers-Danlos syndromes: Complex phenotypes, challenging diagnoses, and poorly understood causes. Gensemer, Cortney et al.
3. Preprint - Defining the Clinical Complexity of hEDS and HSD: A Global Survey of Diagnostic Challenge, Comorbidities, and Unmet Needs. Daylor, Victoria et al.
4. Co-occurrence of Tethered Cord Syndrome and Cervical Spine Instability in Hypermobile Ehlers-Danlos Syndrome. Gensemer, et al.

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UNDERSTANDING Hypermobile EHLERS-DANLOS SYNDROME

Learn more about hypermobile Ehlers-Danlos syndrome (hEDS) and the resources available to support individuals and families.

The Norris Lab

WHAT IS HYPERMOBILE EHLERS-DANLOS SYNDROME?

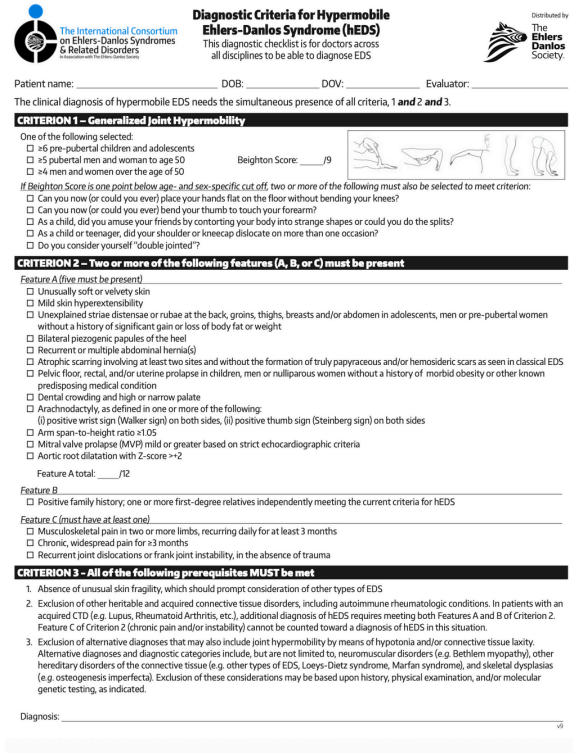
The Ehlers-Danlos syndromes are a group of 14 heritable connective disorders. The most common subtype is the hypermobile type (hEDS).

Prevalence

The prevalence of hypermobile EDS is currently unknown, but it is believed to be more common than rare [2]

DIAGNOSIS

Any provider can make a clinical diagnosis of hEDS with the 2017 criteria provided by the International Consortium on Ehlers-Danlos Syndromes & Related Disorders



The International Consortium on Ehlers-Danlos Syndromes & Related Disorders

Diagnostic Criteria for Hypermobile Ehlers-Danlos Syndrome (hEDS)

This diagnostic checklist is for doctors across all disciplines to be able to diagnose EDS

Distributed by The Ehlers Danlos Society

Patient name: _____ DOB: _____ DOV: _____ Evaluator: _____

The clinical diagnosis of hypermobile EDS needs the simultaneous presence of all criteria, 1 **and** 2 **and** 3.

CRITERION 1 – Generalized Joint Hypermobility

One of the following selected:
- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men and woman to age 50
- ☐ ≥4 men and women over the age of 50

Beighton Score: _____/9

If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:
- ☐ Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- ☐ Can you now (or could you ever) bend your thumb to touch your forearm?
- ☐ As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- ☐ As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- ☐ Do you consider yourself "double jointed"?

CRITERION 2 – Two or more of the following features (A, B, or C) must be present

Feature A (five must be present)
- ☐ Unusually soft or velvety skin
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal 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)
- ☐ 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 predisposing medical condition
- ☐ Dental crowding and high or narrow palate
- ☐ Arachnodactyly, as defined in one or more of the following: (i) positive wrist sign (Walker sign) on both sides, (ii) positive thumb sign (Steinberg sign) on both sides
- ☐ Arm span-to-height ratio ≥1.05
- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: _____/12

Feature B
- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (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

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 CTD (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 toward 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 the 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.

Diagnosis: _____

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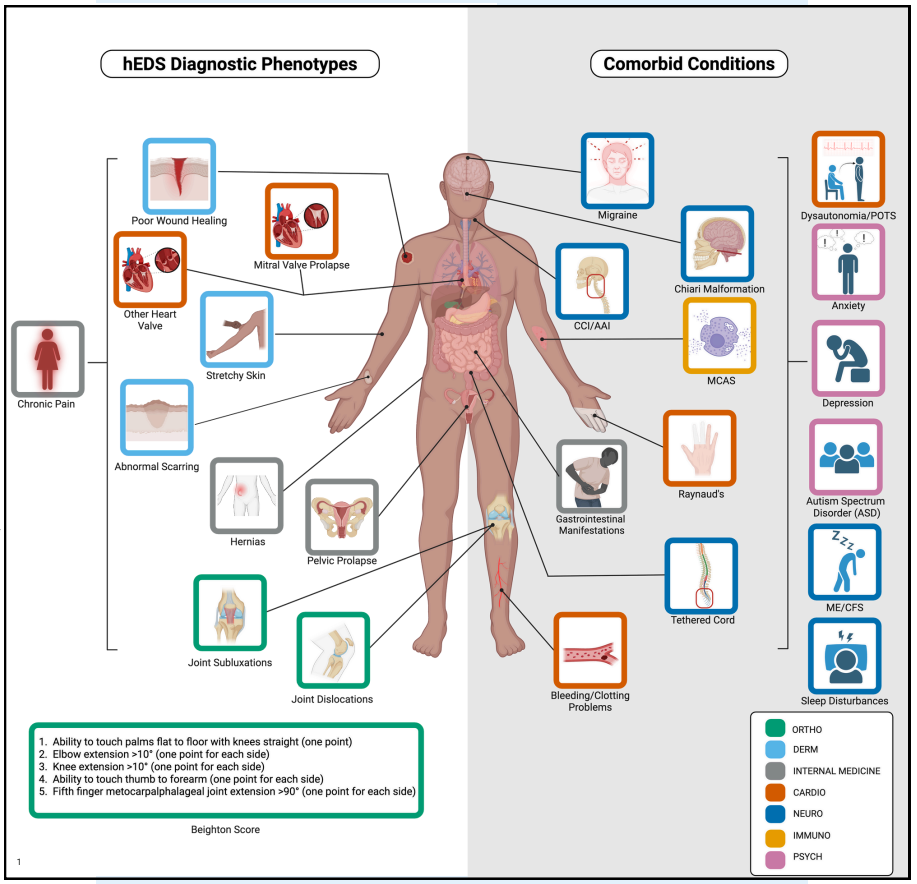
SYMPTOMS AND COMORBIDITIES

hEDS is so much more than hypermobile joints!



Epidemiologic and clinical insights:

- **hEDS is more often diagnosed in women and girls** [2,3]
- **Symptoms typically begin in adolescence, with diagnosis often delayed into adulthood** [3]
- **Multisystemic symptoms are frequently misattributed to anxiety or deconditioning, especially in young women and gender minorities** [3]
- **Family history of hypermobility or chronic pain may be present** [1,2,3]

If you can't connect the issues, think connective tissues!

HOW PRACTITIONERS CAN SUPPORT PATIENTS WITH HEDS

Unlike conditions managed within a single specialty, hEDS often requires coordinated care across multiple disciplines due to its multisystem involvement. Effective management relies on a collaborative, team-based approach—where each provider contributes their expertise to address the diverse ways hEDS can impact a patient's health and quality of life.

Clinical Approach

- Listen closely to patient-reported symptoms, even when they span specialties
- Refer to specialty providers familiar with hypermobility disorders (e.g. physical therapy)
- Order orthostatic vitals to evaluate for comorbid conditions (e.g., POTS, dysautonomia)
- Consult genetic counseling to rule out other subtypes of EDS
- Use shared decision-making to tailor treatment plans to patient needs and goals

Why Early Recognition Matters

Patients with hEDS can face an average diagnostic delay of up to 22 years.[3] Early identification can:

1. Enable timely symptom management, improving function and quality of life
2. Help prevent progressive joint damage, instability, and cardiovascular complications
3. Connect patients to appropriate rehab, autonomic care, GI support, and behavioral healthcare