Hypermobile Ehlers-Danlos Syndrome (hEDS)

Family History & Symptom Monitoring A Patient Guide for Pediatricians

WHAT IS hEDS? hEDS is a heritable disorder of connective tissue, the structural 'glue' of the body, causing joint instability, skin fragility, and systemic effects. Severity varies widely, from mild laxity and intermittent bracing to wheelchair use and complex multisystem involvement. Formal diagnosis in children is deferred until skeletal maturity due to naturally increased joint laxity in growing bodies, but a strong family history combined with systemic symptoms warrants active monitoring and symptom management now. A confirmed diagnosis is not required to take this child seriously. The parent presenting this form may themselves be diagnosed with hEDS and is bringing informed clinical context to this appointment.

~1 in 500 people affected	Avg. 10+ years to diagnosis	3:1 to 4:1 diagnosed are female	No cure: management-focused

HOW HEDS AFFECTS THE BODY — SYSTEMIC INVOLVEMENT:

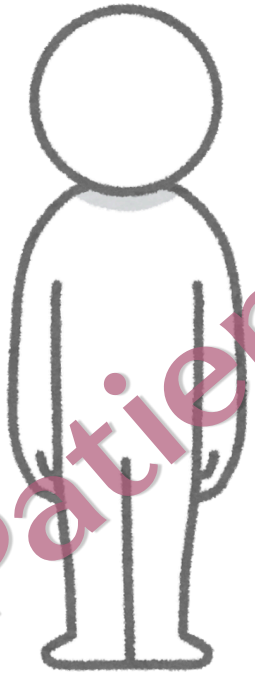
Patient has checked applicable symptoms

Neurological
- ☐ Migraines & headaches
- ☐ Brain fog/cognitive fatigue
- ☐ Small fiber neuropathy
- ☐ Proprioception deficits
- ☐ Anxiety/depression

Gastrointestinal
- ☐ IBS
- ☐ Gastroparesis/ delayed emptying
- ☐ GERD & acid reflux
- ☐ Food intolerances

Immune / MCAS
- ☐ MCAS — mast cell overactivation
- ☐ Flushing, hives, itching
- ☐ GI distress & food reactions
- ☐ Chemical/environmental sensitivity

Musculoskeletal
- ☐ Joint hypermobility & instability
- ☐ Subluxations & dislocations
- ☐ Chronic widespread pain
- ☐ Muscle fatigue & weakness
- ☐ Cervical instability (may contribute to headache, cranial nerve symptoms, or myelopathy)



Cardiovascular
- ☐ POTS — heart rate spikes on standing
- ☐ Blood pooling & dizziness
- ☐ Palpitations

Dermatological
- ☐ Soft, velvety, hyperextensible skin
- ☐ Stretch marks without weight change
- ☐ Easy bruising
- ☐ Poor wound healing

Genitourinary
- ☐ Pelvic floor dysfunction
- ☐ Bladder urgency/frequency
- ☐ Chronic pelvic pain
- ☐ Menstrual irregularities

Fatigue & Sleep
- ☐ Profound fatigue
- ☐ Non-restorative sleep
- ☐ Post-exertional malaise
- ☐ Chronic widespread pain at rest

The hEDS Trifecta: Frequently Co-Occurring Conditions

hEDS
Joint instability
Structurally abnormal connective tissue
Systemic symptoms

+

POTS
Heart rate spikes on standing
Dizziness & fatigue
Brain fog & cognitive dysfunction
Exercise intolerance

+

MCAS
Mast cell overactivation
Flushing, hives, itching
GI distress & food reactions
Chemical/environmental sensitivity

GROWING PAINS AND THE COST OF DISMISSAL: "Growing Pains" Is Not a Diagnosis Growing pains is a label, not a mechanism. In a child with a family history of hEDS presenting with joint pain, fatigue, headaches, and GI symptoms, attributing everything to growing pains closes a clinical conversation that should remain open. Pain that is severe enough to limit activity, disrupt sleep, or cause distress is not typical developmental discomfort regardless of age. Children with hEDS frequently spend their entire childhood being told their pain is normal, and reach adulthood with an average diagnostic delay of over ten years already behind them. Documenting symptoms, monitoring progression, and flagging for adult criteria review costs nothing and prevents years of unnecessary suffering.

DO

- Take family history of hEDS seriously as a significant diagnostic indicator
- Monitor and document joint hypermobility (Beighton score), pain patterns, and systemic symptoms at every well visit
- Flag the chart now for hEDS diagnostic criteria review at skeletal maturity. This is the most durable action this visit can produce
- Screen for dysautonomia and coordinate with cardiology if symptoms are present
- Refer to PT specializing in hypermobility for symptomatic joint instability even before formal diagnosis

DON'T

- Dismiss joint pain and instability as growing pains without considering hEDS family history
- Attribute fatigue, brain fog, and headaches to school stress or typical adolescent experience
- Assume lightheadedness and elevated heart rate on standing are normal without orthostatic assessment
- Assume the absence of a formal diagnosis means nothing is wrong or nothing needs managing
- Dismiss the parent's clinical knowledge. A parent with hEDS frequently understands the condition better than providers who have not encountered it

CONSIDER / REFER

- PT specializing in hypermobility and proprioception. Symptomatic management does not require a formal diagnosis
- Cardiology or neurology referral if dysautonomia symptoms are present
- Orthostatic vitals assessment at well visits for symptomatic children
- Rheumatology referral if diagnostic clarification is appropriate at current age
- School accommodation letter for symptomatic children. Fatigue, pain, and dysautonomia affect attendance and performance
- Pelvic floor PT for adolescents with significant pelvic pain or menstrual symptoms
- GI referral if motility symptoms are present
- Formal chart flag: revisit hEDS diagnostic criteria at skeletal maturity. This is the single most actionable long-term step available before formal diagnosis is possible
- Genetics referral if family history includes multiple EDS subtypes or unclear subtype

COMMON MISDIAGNOSES IN hEDS PATIENTS PRESENTING TO ORTHOPEDICS		
Often Diagnosed As	**Consider Instead/Also**	**Key Differentiator**
Growing pains	hEDS joint instability and hypermobility	Pain that limits activity, disrupts sleep, or causes distress is not typical developmental discomfort
Anxiety disorder	hEDS with dysautonomia and central sensitization	Somatic symptoms are physiological, not psychological; POTS and mast cell activation produce anxiety-like
Functional abdominal pain	hEDS with GI dysmotility or MCAS	Connective tissue laxity affects gut motility; MCAS produces episodic GI symptoms; family history changes the
Juvenile idiopathic arthritis	hEDS joint instability	Normal inflammatory markers; hypermobility on exam; instability rather than synovitis
Depression	hEDS with fatigue, PEM, and dysautonomia	Fatigue and cognitive symptoms are physiological; misattribution delays appropriate management

PUBERTY, DYSAUTONOMIA, AND LIVEDO RETICULARIS: What to Watch For as hEDS-Affected Children Develop

Puberty is a significant inflection point for children with hEDS or hEDS family history. Hormonal changes, particularly the increase in relaxin and fluctuating estrogen and progesterone, amplify connective tissue laxity and autonomic instability. Dysautonomia symptoms including lightheadedness, elevated resting or postural heart rate, exercise intolerance, and fainting frequently emerge or worsen at this stage and are not normal adolescent experience. Menstrual pain and cycle-linked symptom flares are often significantly more severe in this population than in peers and should not be normalized without assessment. Livedo reticularis, a mottled, net-like skin discoloration caused by abnormal blood flow in superficial vessels, is a visible, objective connective tissue finding that is worth documenting when present. It is not cosmetic and its presence in a child with hEDS family history is clinically meaningful.

Sources: Malfait et al. 2017 (AJMG) | Tinkle et al. 2017 (AJMG) | Genetics in Medicine Open 2023 | ehlers-danlos.com | [illegible] | [illegible]

This document was prepared to provide clinical context for a child presenting with hEDS family history and active symptoms.
A confirmed diagnosis is not required to begin monitoring and symptom management. This reference is intended to support that conversation.

PLEASE COMPLETE THIS PAGE BEFORE YOUR CHILD'S APPOINTMENT AND BRING IT WITH YOU

CURRENT MEDICATIONS & SUPPLEMENTS

WHAT HELPS:

WHAT MAKES IT WORSE:

CURRENT SYMPTOM SEVERITY: use pain scale (eg. 4) to rate severity

Joint pain/instability severity/frequency:

Fatigue severity/frequency:

GI symptoms type/frequency:

Heart rate/dizziness severity/frequency:

Additional symptoms:

WHAT WE NEED FROM TODAY'S APPOINTMENT

Referrals needed:

Questions I have:

Medication changes to discuss:

My primary concern today:

Other:

MANKOSKI PAIN SCALE

Use this scale when rating your pain severity in CURRENT SYMPTOM SEVERITY

#	What the pain is like	Typical treatment	In my own words
0	No pain.	*No medication needed.*	*"I feel completely normal."*
1	Very minor annoyance — occasional minor twinges.	*No medication needed.*	*"Hardly notice it."*
2	Minor annoyance — occasional strong twinges.	*No medication needed.*	*"Annoying but manageable."*
3	Annoying enough to be distracting.	*Mild OTC painkillers may help.*	*"Hard to ignore; affects my focus."*
4	Can be ignored if very focused, but still distracting.	*Mild OTC painkillers relieve pain for 3–4 hours.*	*"Getting in the way of tasks."*
5	Can't be ignored for more than 30 minutes.	*Mild OTC painkillers reduce pain for 3–4 hours.*	*"Stops me mid-task."*
6	Can't be ignored. Can still go to work and participate in social activities.	*Stronger prescription pain relief needed; works 3-4 hours.*	*"Present all the time. I push through."*
7	Difficult to concentrate; interferes with sleep. Can still function with effort.	*Stronger painkillers only partially effective.*	*"Hard to function. Sleep is disrupted."*
8	Physical activity severely limited. Can read/converse with effort. Nausea possible.	*Strongest painkillers minimally effective.*	*"Mostly bed-bound. May feel nauseated."*
9	Unable to speak. Crying out or moaning uncontrollably. Near delirium.	*Strongest painkillers only partially effective.*	*"Cannot communicate. Losing control."*
10	Unconscious. Pain causes passing out.	*Strongest painkillers only partially effective.*	*"Passed out or on the verge of it."*

Mankoski Pain Scale developed by Andrew Mankoski, 1995. Adapted for patient communication. Not a clinical diagnostic tool.

IMPORTANT NOTE FOR HEDS PATIENTS & PROVIDERS:

People with hEDS often have an altered pain baseline due to central sensitization — a process in which the nervous system becomes increasingly sensitized to pain signals over time.

A "7" for this patient may be what others feel as a "9."
Please do not compare severity numbers to those of patients without chronic illness.

This scale helps us communicate.
It is not a measure of tolerance, willpower, or how "bad" things really are.