Hypermobile Ehlers-Danlos Syndrome

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FAST FACTS

6 years
of age, minimum age for assessment of hEDS

30 minutes, 5 days/week
of low-impact exercise such as swimming or riding a recumbent bike recommended for management of hEDS

10%
of the population may be considered hypermobile

no known genetic cause,
but it typically runs in families

Hypermobile Ehlers-Danlos Syndrome (hEDS) is the most common form of a group of inherited connective tissue disorders. hEDS is generally characterized by joint hypermobility, skin hyperextensibility, and tissue fragility. The diagnosis of hEDS is clinical; there is no identifiable genetic cause yet, so no test is available. Hypermobility presents across a spectrum ranging from asymptomatic joint hypermobility to hEDS. There is no cure for hEDS.

ASSESSMENT

Perform a standard health history and physical exam, with probing history questions for both patient and family.

HPE RED FLAGS

Prior to referring for hEDS, consider other conditions which require additional workup, referral or testing.

Personal History
- Unusual skin fragility (which should drive consideration of other EDS types)
- Skeletal dysplasia (e.g., osteogenesis imperfecta)
- Spasticity
- Low muscle tone
- Common neuromuscular manifestations due to a known condition
- Rheumatologic symptoms

Family History
Self or first-degree relative:
- Aortic disease/aortic root dilation
- Aneurysm
- Organ rupture
- Bowel perforation
- Other genetic or acquired connective tissue disorders

MANAGEMENT/TREATMENT

Treat based on symptoms, and refer to specialists for help with associated complications/issues and further education:
- Do low-impact exercise for 30 minutes 5 days a week.
- Hydrate and keep track of daily water intake.
- Manage and prevent injury over the long-term through strengthening, proprioceptive training, joint protection and endurance training (as guided by OT/PT). Consider nontraditional interventions like massage, yoga, meditation and acupuncture. Do not recommend joint/spinal manipulation therapy due to joint laxity/instability.
- Recommend NSAIDS, heating pads and cold packs as needed for pain. Other medications can help for certain types of pain in specific situations which often need expert input to ensure the balance between benefit and risk is maintained.
- Treat associated symptoms, which may be more debilitating and have more impact on daily living than the joint symptoms—including anxiety, depression, dysautonomia, fatigue, functional GI disorders, headaches, postural orthotic tachycardia and sleep disturbances.

WHEN TO REFER

1. Consider other disorders or conditions before referring for hEDS.
2. Refer based on the presence or absence of Red Flags. See algorithm on reverse side for more specific information.

If you have clinical questions about patients with Hypermobile Ehlers-Danlos Syndrome, email EDSforDocs@cchmc.org.

If you would like additional copies of this tool, or would like more information, please contact the Physician Outreach and Engagement team at Cincinnati Children's.

Tool developed by Cincinnati Children's physician-hospital organization (known as Tri-State Child Health Services, Inc.) and staff in the James M. Anderson Center for Health Systems Excellence. Developed using expert consensus and informed by Best Evidence Statements, Care Practice Guidelines, and other evidence-based documents as available. For Evidence-Based Care Guidelines and references, see www.cincinnatichildrens.org/evidence.

Hypermobile Ehlers-Danlos Syndrome

Patient Presents

Standard Workup

- Personal History
- Family History
- Physical Exam

HPE Red Flags

Personal History

- Unusual skin fragility (which should drive consideration of other EDS types)
- Skeletal dysplasia (e.g., osteogenesis imperfecta)
- Spasticity
- Low muscle tone
- Common neuromuscular manifestations due to a known condition
- Rheumatologic symptoms – prolonged morning stiffness, limping, asymmetric joint pains, daily persistent joint swelling (>6 weeks duration

Family History

Self or first-degree relative:

- Aortic disease/aortic root dilation
- Aneurysm
- Organ rupture
- Bowel perforation
- Other genetic connective tissue disorders (e.g., Marfan, Loey's-Dietz or other forms of EDS)
- Other acquired connective tissue disorders (e.g., lupus, mixed connective tissue disorders)

Any Red Flags?

Yes

Refer accordingly:

General Genetics

- Classic or other type of EDS, except vascular EDS
- Unusual skin fragility (consider other types of EDS)
- Spasticity
- Low muscle tone or common neuromuscular manifestations due to a known condition — confirmed or suspected, such as chromosomal disorders or muscular dystrophy
- First degree relative with:
  - Bowel perforation
  - Low muscle tone
  - Common neuromuscular conditions

Skeletal Dysplasia Clinic

- Skeletal dysplasia (e.g., osteogenesis imperfecta)

Cardiovascular Genetics

- Aortic root dilation
- Ectopia lentis
- Other genetic connective tissue disorders, e.g., Marfan, Loey's-Dietz, vascular EDS
- Personal history of bowel perforation or organ rupture
- First degree relative history of thoracic aortic aneurysm/dissection

Rheumatology

- Rheumatologic symptoms, e.g., prolonged morning stiffness, limping, asymmetric joint pains, daily persistent joint swelling
- Personal history or first-degree relative with other acquired connective tissue disorders, e.g., lupus, mixed connective tissue disorders

No

Refer accordingly:

Genetics Hypermobility Clinic

- Symptomatic hypermobility

OT/PT

- Joint hypermobility with or without joint pain or instability, including hEDS, other types of EDS, other Hypermobility Spectrum Disorders, joint laxity or other generalized hypermobility

Consider diagnosis based on 2017 diagnostic criteria found at www.ehlers-danlos.com.

To make an hEDS diagnosis, all three criteria (1, 2, 3) must be met.

1. Generalized joint hypermobility (as defined by the Beighton score)
   a. ≥6 for pre-pubertal children and adolescents
   b. ≥5 for pubertal adolescents and young adults

2. Two or more of the following must be present
   a. Minimum 5 of 12 systemic manifestations of a more generalized connective tissue disorder (www.ehlers-danlos.com/heds-diagnostic-checklist/)
   b. One or more first-degree relatives independently meeting current diagnostic criteria for hEDS
   c. Musculoskeletal complications of joint instability

3. Meet ALL of these prerequisites
   a. Absence of unusual skin fragility
   b. Other heritable and acquired connective tissue disorders including autoimmune rheumatologic conditions have been considered
   c. Alternative diagnoses that may include joint hypermobility by means of hypotonia/connective tissue laxity have been ruled out

For urgent issues, or to speak with the specialist on call 24/7, call the Physician Priority Link at 1-888-987-7997.

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