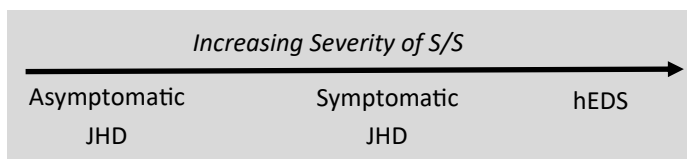


Hypermobile Ehlers-Danlos Syndrome (hEDS)

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BACKGROUND

- EDS is a heterogeneous group of heritable connective tissue (CT) disorders, caused by abnormal collagen formation^{2,6}
 - Prevalence:** 1 in 5,000 for all types of EDS, unknown prevalence of hEDS specifically due to change in dx criteria
- hEDS is 1 of 13 different types of EDS and is on the severe end of the spectrum of joint hypermobility disorders (JHD)^{4,5}



- Risk Factors**^{2,6}
 - Non-Caucasian, Female, Family Hx

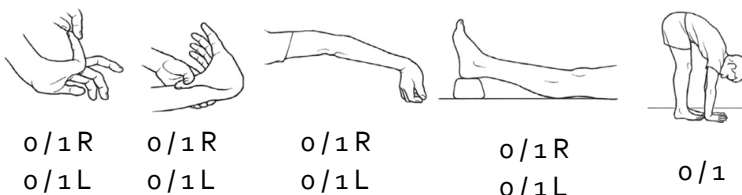
DIAGNOSTIC CRITERIA

***Unlike other types of EDS, there is no known genetic markers to diagnose hEDS. Thus, it is a clinical diagnosis of exclusion^{2,4-6}*

Clinical Diagnosis (presence of 3 criteria)^{1,5}

- Generalized Joint Hypermobility**
 - ⇒ Beighton Score commonly used (see below)
 - ≥6 (pre-pubertal children)
 - ≥5 (post-pubertal, up to 50 yo)
 - ≥4 (+50 yo)
- At least 2 of the following:**
 - ⇒ Systemic manifestations of CT disorders (see S/S)
 - ⇒ Positive family hx
 - ⇒ MSK complications
- Absence of excessive skin fragility, exclusion of other CT disorders (Marfan, Loey-Dietz) or other causes of hypermobility (hypotonia, skeletal dysplasia)**

Beighton Scoring (0-9 points; yes = 1 point, no = 0 points)²



SIGNS, SYMPTOMS & COMORBIDITIES^{1,2,4}

***Unclear distinction between presentations that are caused by EDS and those that are often seen as comorbidities*

- Pain (bilateral, chronic) 🚩
- Extensible, soft, "velvety" skin
- Fatigue 🚩
- Postural Orthostatic Tachycardia Syndrome (POTS) 🚩
- Orthostatic Intolerance 🚩
- GI Disorders (IBS, GERD, diarrhea) 🚩
- Hx of dislocations or subluxations
- Osteoarthritis
- Headache
- TMJ Dysfunction
- Sleep Disturbances 🚩
- Depression, Anxiety
- Kinesiophobia
- Pelvic Floor Dysfunction (UI) 🚩
- Proprioception & Balance Problems

KEY: Systemic S/S & Red Flag Criteria 🚩

REFERRALS & PT ROLE

Most cases of undiagnosed hEDS will warrant a **non-immediate physician referral**. However, if a patient presents with a subluxation or s/s that would require emergent management in the general population, make an **immediate physician referral**.

PT Treatment^{3,4}

- Patient education, pain management and lifestyle modifications
- Low-impact resistance training, aquatic exercise, joint stabilization, proprioception training, bracing and splinting.
- PTs should be aware of potential complications and monitor patients closely

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