Understanding *EDS & HSD*

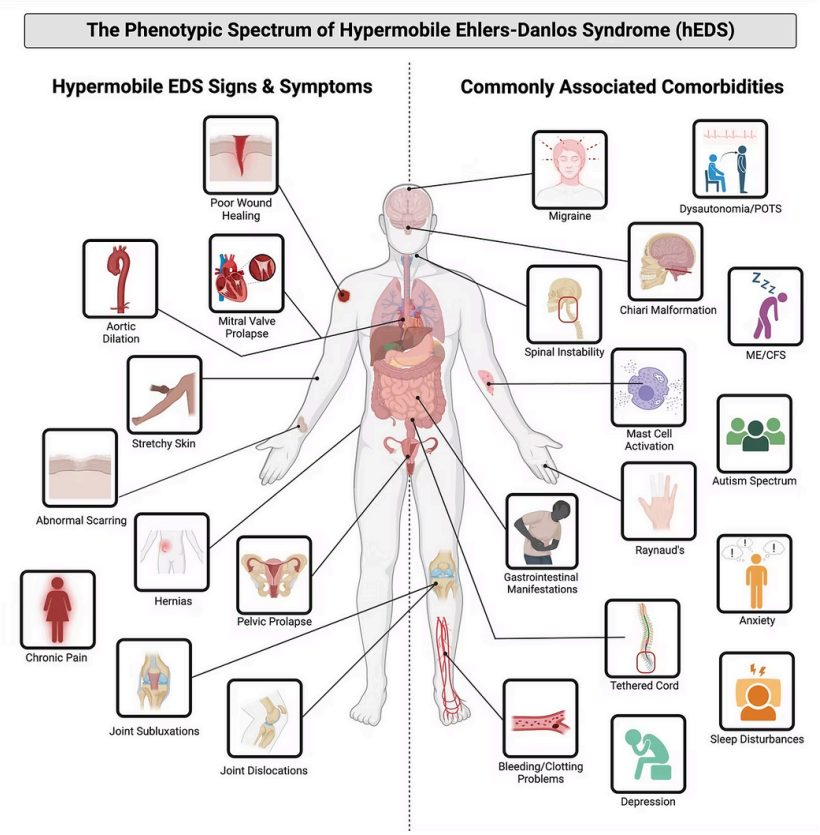
EDS RESOURCES | THE NORRIS LAB | FEB 2025

What are EDS & HSD?

The Ehlers-Danlos syndromes (EDS) are a group of 14 genetic connective tissue disorders that affect multiple systems within the body, particularly affecting connective tissue such as the skin, bones, cartilage, and blood vessels throughout the whole body. Hypermobility Spectrum Disorders (HSD) encompass connective tissue disorders with features of joint hypermobility and related symptoms which do not fully meet the current criteria for hypermobile EDS (hEDS).

Hypermobile Ehlers-Danlos Syndrome (hEDS)

hEDS is the most common subtype, inherited in what is suspected to be an autosomal dominant pattern. Unlike other EDS types, hEDS does not currently have a known genetic marker.



Common Symptoms:

- **Joint Hypermobility:** Unusual flexibility, joint instability, subluxations, and dislocations.
- **Skin Changes:** Soft, stretchy skin, easy bruising, and slow wound healing.
- **Chronic Pain & Fatigue:** Widespread pain, joint pain, and persistent exhaustion.
- **Cardiovascular Issues:** Mitral valve prolapse, aortic root dilation, and possible valve defects such as aortic, pulmonary, and tricuspid.
- **Gastrointestinal Issues:** IBS-like symptoms such as bloating, abdominal pain, nausea, reflux, delayed gastric emptying and intestinal dysmotility.
- **Laryngological Issues:** Dysphonia, dysphagia, hoarseness, vocal cord hemorrhage.
- **Dysautonomia:** Dizziness, fainting, and temperature regulation issues.

Common Co-morbidities & Features:

- **Neurological & Nervous System Issues:** Craniocervical instability, Chiari malformation, Tethered cord syndrome, Raynaud's phenomenon, and neuropathies such as peripheral neuropathy and or small fiber neuropathy.
- **Immune and Allergic Disorders:** Mast cell activation syndrome, Immune dysfunction
- **Musculoskeletal & Structural Issues:** TMJ dysfunction, scoliosis, flat feet, pelvic organ prolapse.
- **Autonomic Issues:** Postural orthostatic tachycardia syndrome (POTS)
- **Gastrointestinal Disorders:** Gastroparesis, IBS, SIBO, Eosinophilic esophagitis, Visceroptosis, and abdominal hernia.
- **Gynecological & Reproductive Disorders:** Interstitial cystitis, Endometriosis, Pelvic floor dysfunction, Pelvic organ prolapse, PCOS
- **Vascular Compression Syndromes:** Thoracic outlet syndrome, Median arcuate ligament syndrome (MALS), Nutcracker syndrome, Superior mesenteric artery syndrome (SMAS)
- **Other:** Potential overlap of autoimmune conditions

Diagnosis & Testing:

hEDS is diagnosed based on the 2017 International Classification of the Ehlers-Danlos Syndromes criteria consisting of three criterion:

1. **Generalized Joint Hypermobility** (using the Beighton Score - a hypermobility screening tool)
2. **Additional Symptoms & Comorbidities** (3 feature checklist containing required characteristics)
3. **Exclusion of Other Conditions** (e.g., Marfan Syndrome, Lupus, other CTDs)

Why Early Diagnosis Matters:

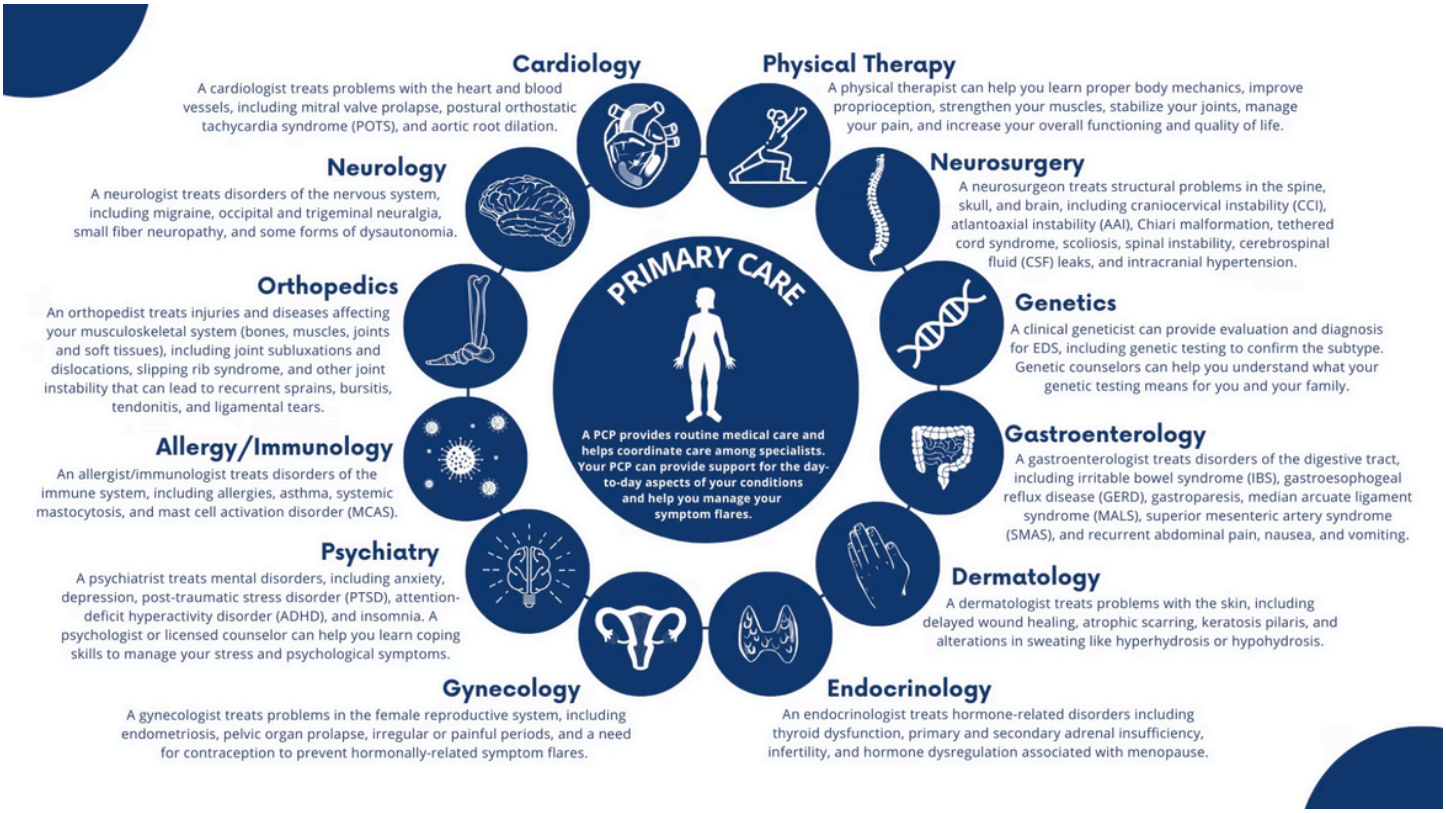
- Provides guidance on symptom management to improve quality of life
- Helps prevent complications like joint damage & cardiovascular issues
- Allows access to tailored treatments & support networks

Management & Treatment:

Although there is no cure, current symptom management strategies include:

- **Physical Therapy:** Strengthening muscles to stabilize joints
- **Pain Management:** Medications, bracing, and lifestyle modifications
- **Cardiac Monitoring:** Regular echocardiograms if necessary
- **Gastrointestinal Support:** Specialized diets, hydration, and medications

What Providers Could You See for EDS?



About the Norris Lab: The Norris Lab is a multidisciplinary environment studying cardiovascular disease and EDS. Consisting of motivated students, technicians, and postdocs who utilize molecular, biochemical and biomechanical tools to further understand the genetic and molecular underpinnings of Hypermobile Ehlers-Danlos Syndrome.