Ehlers-Danlos Syndromes

The Arc of New Jersey Family Institute
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1. What is Ehlers-Danlos syndromes?

The Ehlers-Danlos syndromes are a group of connective tissue disorders that can be inherited and are varied both in how affect the body and in their genetic causes. They are generally characterized by joint hypermobility (joints that stretch further than normal), skin hyperextensibility (skin that can be stretched further than normal), and tissue fragility.

The Ehlers-Danlos syndromes (EDS) are currently classified into thirteen subtypes. Each EDS subtype has a set of clinical criteria that help guide diagnosis; a patient's physical signs and symptoms will be matched up to the major and minor criteria to identify the subtype that is the most complete fit. There is substantial symptom overlap between the EDS subtypes and the other connective tissue disorders including hypermobility spectrum disorders, as well as a lot of variability, so a definitive diagnosis for all the EDS subtypes when the gene mutation is known—all but hypermobile EDS (hEDS)—also calls for confirmation by testing to identify the responsible variant for the gene affected in each subtype.

2. **What are the symptoms of Ehlers-Danlos syndromes?**
   Clinical manifestations of an Ehlers-Danlos syndrome are most often joint and skin related and may include:

- **Joints**
  Joint hypermobility; loose/unstable joints which are prone to frequent dislocations and/or subluxations; joint pain; hyperextensible joints (they move beyond the joint's normal range); early onset of osteoarthritis.
- **Skin**
  Soft velvety-like skin; variable skin hyper-extensibility; fragile skin that tears or bruises easily (bruising may be severe); severe scarring; slow and poor wound healing; development of molluscoid pseudo tumors (fleshy lesions associated with scars over pressure areas).
- **Miscellaneous/Less Common**
  Chronic, early onset, debilitating musculoskeletal pain (usually associated with the Hypermobility Type); arterial/intestinal/uterine fragility or rupture (usually associated with the Vascular Type); scoliosis at birth and scleral fragility (associated with the Kyphoscoliosis Type); poor muscle tone (associated with the Arthrochalasia Type); mitral valve prolapse; and gum disease.

Each type of Ehlers-Danlos syndrome is defined as a distinct problem in connective tissue. Connective tissue is what the body uses to provide strength and elasticity; normal connective tissue holds strong proteins that allow tissue to be stretched but not beyond its limit, and then safely return that tissue to normal. Connective tissue is found throughout the body, and Ehlers-Danlos syndromes are structural problems. An analogy: If one builds a house with faulty materials, say half the necessary wood or with soft aluminum nails, it is certain there will be problems. Some problems are more likely to show up than others, but because those materials were used everywhere and are not necessarily visible, one can be surprised by where a problem shows up or how serious it is. It is much the same thing with an Ehlers-Danlos Syndrome and connective tissue.

3. **How is EDS diagnosed?**
   At this time, research statistics of the Ehlers-Danlos syndromes show the total prevalence as 1 in 2,500 to 1 in 5,000 people. Recent clinical experience suggests that Ehlers-Danlos syndrome may be more common. The conditions are known to affect both males and females of all racial and ethnic backgrounds. If you think you might have one of the Ehlers-Danlos syndromes (EDS) or hypermobility spectrum disorders (HSD), and particularly if someone in your immediate family has been diagnosed, ask your doctor if a diagnosis fits your symptoms. If they choose to, any doctor who can diagnosis a disease is able to diagnose EDS/HSD; but most likely you'll be given a referral to a geneticist, because EDS are genetic disorders and geneticists are most adept at distinguishing between those diseases, as well as in doing any testing necessary to differentiate EDS/HSD from the more than 200 other heritable connective tissue disorders.

   The two known inheritance patterns for the Ehlers-Danlos syndromes include autosomal dominant and autosomal recessive. Regardless of the inheritance pattern, we have no choice in which genes we pass on to our children.

4. **Prevention:**
   If you have a personal or family history of Ehlers-Danlos syndrome and you're thinking about starting a family, you may benefit from talking to a genetic counselor — a health care professional trained to assess the risk of inherited disorders. Genetic counseling can help you understand the inheritance pattern of the type of Ehlers-Danlos syndrome that affects you and the risks it poses for your children.

For more information, visit:

- The Ehlers-Danlos Society: www.ehlers-danlos.com
- Mayo Clinic: www.mayoclinic.org/diseases-conditions/ehlers-danlos-syndrome/symptoms-causes/syc-20362125
- EDS Awareness: www.edsawareness.com