



What are the Ehlers-Danlos syndromes?

The Ehlers-Danlos syndromes (EDS) are a group of connective tissue disorders that can be inherited and are varied, both in how they 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.

Connective tissue is the material in the body that binds together, supports, and separates different tissues and organs. Found between other tissues everywhere in the body, it provides strength and flexibility, and helps perform general functions as well as specialized services. Connective tissue disorders disrupt these most fundamental processes and structures of the body, so resulting problems can be widespread, in a wide range of severities, and affect areas that might seem to be otherwise unrelated.

Early diagnosis is crucial to positive patient health. Symptoms can be treated as they arise. Care is largely preventative, to support and manage EDS with the intent of keeping damage as minimal as possible. Specifics have to be tailored to those symptoms exhibited in the person with EDS. EDS are known to affect men and women of every race and ethnicity.

EDS are currently classified into thirteen subtypes. A person'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 EDS and other connective tissue disorders, so a definitive diagnosis for EDS when the gene mutation is known—all but hypermobile EDS—also calls for confirmation by testing to identify the responsible variant for the gene affected.

Please remember that an individual's experience with an EDS is their own, and may not necessarily be the same as another person's experience. Diagnostic criteria are meant solely to distinguish an EDS from other connective tissue disorders, and there are many more possible symptoms for each EDS than there are criteria.

Classical [COL5A1, COL5A2, rarely COL1A1]

Classical-like [TNXB]

Cardiac-valvular [COL1A2]

Vascular [COL3A1, rarely COL1A1]: possibility of shortened lifespan; arterial rupture is the most common cause of sudden death.

Hypermobile [no identified cause]

Arthrochalasia [COL1A1, COL1A2]

Dermatosparaxis [ADAMTS2]

Kyphoscoliotic [PLOD1, FKBP14]

Brittle cornea syndrome [ZNF469, PRDM5]

Spondylodysplastic [B4GALT7, B3GALT6, SLC39A13]

Musculocontractural [CHST14, DSE]

Myopathic [COL12A1]

Periodontal [C1R, C1S]

For more information,
ehlers-danlos.com