

These are the Ehlers-Danlos syndromes

The Ehlers-Danlos syndromes 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, as an individual's experience with an EDS is their own, and may not necessarily be the same as another person's experience. EDS are known to affect men and women of every race and ethnicity.

These are the types of Ehlers-Danlos

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.

For those who meet the minimal clinical requirements for an EDS subtype – but who have no access to molecular confirmation; or whose genetic testing shows one (or more) gene variants of uncertain significance in the genes identified for one of the EDS subtypes; or in whom no causative variants are identified in any of the EDS-subtype-specific genes – a “provisional clinical diagnosis” of an EDS subtype can be made. These patients should be followed clinically, but alternative diagnoses and expanded molecular testing should be considered.

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] affects skin, wound healing, and joints. Joints can stretch beyond normal, causing pain. Skin that can be stretched further than it should is easily torn, and doesn't repair itself well, causing disfiguring scarring.

Classical-like [TNXB] is distinguished by generalized joint hypermobility; hyperextensible, soft and/or velvety skin without the typical atrophic scarring seen in classical EDS; and easy bruising.

Cardiac-valvular [COL1A2] involves severe cardiac-valvular disease that requires valve replacement surgery in conjunction with variable skin hyperextensibility, atrophic scarring, and joint hypermobility.

Vascular [COL3A1, rarely COL1A1] is the most serious type due to the possibility of shortened lifespan. Minor trauma can lead to extensive bruising and skin tears. Arterial rupture is the most common cause of sudden death. In childhood easy bruising is most often noticed, perhaps accompanied by striking skin lucency and visibility of blood vessels. Additional features can include unusual bruising without identified cause, premature aging of hands and feet, early onset varicose veins, and characteristic facial features with prominent eyes. In the absence of a family history, the diagnosis of vascular EDS is often not considered until after a vessel or hollow organ rupture.

Hypermobile EDS as yet has no identified distinctive cause; it evolves over time. The “hypermobility” phase (the first years of life) involves contortionism and propensity for sprains and dislocations; pain, often limited to lower limbs, also with fine motor or repetitive tasks; easy fatigability may be a feature, together with voiding dysfunction. The “pain” phase (starting 2nd to 4th decade of life) includes more widespread and progressively worsening musculoskeletal pain, pelvic pain in women, and headache; exacerbation of fatigue; and is often associated with additional complaints. The “stiffness” phase (observed in a few adults and elderly only) results in general reduction of joint mobility; significant reduction in functionality due to disabling symptoms of pain and fatigue, and limitations from reduced muscle mass and weakness, prior injuries, and arthritis. The clinical description of hEDS in the medical literature has expanded considerably to include more features, such as chronic pain, chronic fatigue, dysautonomia, and anxiety among other associated symptoms.

Arthrochalasia [*COL1A1, COL1A2*] displays severe generalized joint hypermobility, congenital bilateral hip dislocation, and recurrent subluxations and dislocations of both small and large joints.

Dermatosparaxis [*ADAMTS2*] causes extreme skin fragility with redundant, almost lax skin, and severe susceptibility of bruising.

Kyphoscoliotic [*PLOD1, FKBP14*] *PLOD1* form results in abnormal spine curvature at birth, reduced muscle tone, and joint hypermobility; the *FKBP14* form is characterized by kyphoscoliosis, severe reduced muscle tone at birth with muscle atrophy, joint hypermobility, and congenital hearing loss.

Brittle cornea syndrome [*ZNF469, PRDM5*] displays thin, fragile cornea, with an increased risk for spontaneous corneal rupture

Spondylodysplastic [*B4GALT7, B3GALT6, SLC39A13*] *B4GALT7* produces short stature and intellectual disability. The *B3GALT6* form results in characteristic head and facial features; abnormal spine curvature; joint hypermobility and deformed, rigid joints; short stature; osteoporosis with multiple fractures; and intellectual disability. *SLC39A13* is characterized by moderate short stature; hyperelastic, velvety, thin skin with an easily visible venous pattern, and bruisability which leads to atrophic scars; slender, tapering fingers, wrinkled palms, and considerable atrophy; and distal joint hypermobility which later results in contractures.

Musculocontractural [*CHST14, DSE*] results in distinctive head and facial features; multiple deformed and rigid joints at birth, including adducted thumbs and club foot; characteristic skin features including fine palmar creases; peculiar finger

shapes; progressive spinal and foot deformities; large subcutaneous hematomas; and ophthalmological and urogenital involvement.

Myopathic [*COL12A1*] involves muscle weakness that is present in infancy or childhood and is associated with proximal large joint rigidity and distal joint hypermobility. The muscle weakness tends to get better with age until young adulthood with some deterioration in the 4th decade.

Periodontal [*C1R, C1S*] results in early-onset inflammation of the tissue around teeth, with extensive gum destruction and loss of teeth starting in childhood or adolescence.

The Ehlers-Danlos Society

The Ehlers-Danlos Society is a global community of patients, caregivers, healthcare professionals, and supporters, dedicated to saving and improving the lives of those affected by the Ehlers-Danlos syndromes and related disorders.

We support collaborative research initiatives, awareness campaigns, advocacy, community-building, and care for the EDS population.

Our goals are worldwide awareness – and a better quality of life for all who suffer from these conditions. Research is at the center of what we do, so that one day we will have a cure.

Our strength begins with hope.

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What are the Ehlers-Danlos syndromes?



The
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