Ehlers-Danlos syndrome

Ehlers-Danlos syndrome is a group of disorders that affect connective tissues supporting the skin, bones, blood vessels, and many other organs and tissues. Defects in connective tissues cause the signs and symptoms of these conditions, which range from mildly loose joints to life-threatening complications.

The various forms of Ehlers-Danlos syndrome have been classified in several different ways. Originally, 11 forms of Ehlers-Danlos syndrome were named using Roman numerals to indicate the types (type I, type II, and so on). In 1997, researchers proposed a simpler classification (the Villefranche nomenclature) that reduced the number of types to six and gave them descriptive names based on their major features. In 2017, the classification was updated to include rare forms of Ehlers-Danlos syndrome that were discovered more recently. The 2017 classification describes 13 types of Ehlers-Danlos syndrome.

An unusually large range of joint movement (hypermobility) occurs in most forms of Ehlers-Danlos syndrome, and it is a hallmark feature of the hypermobile type. Infants and children with hypermobility often have weak muscle tone (hypotonia), which can delay the development of motor skills such as sitting, standing, and walking. The loose joints are unstable and prone to dislocation and chronic pain. In the arthrochalasia type of Ehlers-Danlos syndrome, infants have hypermobility and dislocations of both hips at birth.

Many people with the Ehlers-Danlos syndromes have soft, velvety skin that is highly stretchy (elastic) and fragile. Affected individuals tend to bruise easily, and some types of the condition also cause abnormal scarring. People with the classical form of Ehlers-Danlos syndrome experience wounds that split open with little bleeding and leave scars that widen over time to create characteristic "cigarette paper" scars. The dermatosparaxis type of the disorder is characterized by loose skin that sags and wrinkles, and extra (redundant) folds of skin may be present.

Some forms of Ehlers-Danlos syndrome, notably the vascular type and to a lesser extent the kyphoscoliotic, classical, and classical-like types, can cause unpredictable tearing (rupture) of blood vessels, leading to internal bleeding and other potentially life-threatening complications. The vascular type of Ehlers-Danlos syndrome is also associated with an increased risk of organ rupture, including tearing of the intestine and rupture of the uterus during pregnancy.

Other types of Ehlers-Danlos syndrome have additional signs and symptoms. The cardiac-valvular type causes severe problems with the valves that control the movement of blood through the heart. People with the kyphoscoliotic type experience severe curvature of the spine that worsens over time and can interfere with breathing

by restricting lung expansion. A type of Ehlers-Danlos syndrome called brittle cornea syndrome is characterized by thinness of the clear covering of the eye (the cornea) and other eye abnormalities. The spondylodysplastic type features short stature and skeletal abnormalities such as abnormally curved (bowed) limbs. Abnormalities of muscles, including hypotonia and permanently bent joints (contractures), are among the characteristic signs of the musculocontractural and myopathic forms of Ehlers-Danlos syndrome. The periodontal type causes abnormalities of the teeth and gums.

Frequency

The combined prevalence of all types of Ehlers-Danlos syndrome appears to be at least 1 in 5,000 individuals worldwide. The hypermobile and classical forms are most common; the hypermobile type may affect as many as 1 in 5,000 to 20,000 people, while the classical type probably occurs in 1 in 20,000 to 40,000 people. Other forms of Ehlers-Danlos syndrome are rare, often with only a few cases or affected families described in the medical literature.

Causes

Mutations in at least 19 genes have been found to cause the Ehlers-Danlos syndromes. Mutations in the *COL5A1* or *COL5A2* gene, or rarely in the *COL1A1* gene, can cause the classical type. Mutations in the *TNXB* gene cause the classical-like type and have been reported in a very small percentage of cases of the hypermobile type (although in most people with this type, the cause is unknown). The cardiac-valvular type and some cases of the arthrochalasia type are caused by *COL1A2* gene mutations; mutations in the *COL1A1* gene have also been found in people with the arthrochalasia type. Most cases of the vascular type result from mutations in the *COL3A1* gene, although rarely this type is caused by certain *COL1A1* gene mutations. The dermatosparaxis type is caused by mutations in the *ADAMTS2* gene. *PLOD1* or *FKBP14* gene mutations result in the kyphoscoliotic type. Other rare forms of Ehlers-Danlos syndrome result from mutations in other genes.

Some of the genes associated with the Ehlers-Danlos syndromes, including *COL1A1*, *COL1A2*, *COL3A1*, *COL5A1*, and *COL5A2*, provide instructions for making pieces of several different types of collagen. These pieces assemble to form mature collagen molecules that give structure and strength to connective tissues throughout the body. Other genes, including *ADAMTS2*, *FKBP14*, *PLOD1*, and *TNXB*, provide instructions for making proteins that process, fold, or interact with collagen. Mutations in any of these genes disrupt the production or processing of collagen, preventing these molecules from being assembled properly. These changes weaken connective tissues in the skin, bones, and other parts of the body, resulting in the characteristic features of the Ehlers-Danlos syndromes.

Some genes associated with recently described types of Ehlers-Danlos syndrome have functions that appear to be unrelated to collagen. For many of these genes, it is not clear how mutations lead to hypermobility, elastic skin, and other features of these conditions.

Inheritance Pattern

The inheritance pattern of the Ehlers-Danlos syndromes varies by type. The classical, vascular, arthrochalasia, and periodontal forms of the disorder, and likely the hypermobile type, have an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder. In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new (de novo) gene mutations and occur in people with no history of the disorder in their family.

The classical-like, cardiac-valvular, dermatosparaxis, kyphoscoliotic, spondylodysplastic, and musculocontractural types of Ehlers-Danlos syndrome, as well as brittle cornea syndrome, are inherited in an autosomal recessive pattern. In autosomal recessive inheritance, two copies of a gene in each cell are altered. Most often, the parents of an individual with an autosomal recessive disorder are carriers of one copy of the altered gene but do not show signs and symptoms of the disorder.

The myopathic type of Ehlers-Danlos syndrome can have either an autosomal dominant or autosomal recessive pattern of inheritance.

Other Names for This Condition

- EDS
- Ehlers Danlos disease

Diagnosis & Management

Formal Diagnostic Criteria

- Bowen JM, Sobey GJ, Burrows NP, Colombi M, Lavallee ME, Malfait F, Francomano CA. Ehlers-Danlos syndrome, classical type. Am J Med Genet C Semin Med Genet. 2017 Mar;175(1):27-39. doi: 10.1002/ajmg.c.31548. Epub 2017 Feb 13. Review.
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Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Ehlers-Danlos syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0013720/
- Genetic Testing Registry: Ehlers-Danlos syndrome Beasley Cohen type
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1837462/
- Genetic Testing Registry: Ehlers-Danlos syndrome dysfibronectinemic type
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857038/
- Genetic Testing Registry: Ehlers-Danlos syndrome progeroid type
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C4552003/
- Genetic Testing Registry: Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C3281160/
- Genetic Testing Registry: Ehlers-Danlos syndrome, autosomal recessive, cardiac valvular form
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C4303789/
- Genetic Testing Registry: Ehlers-Danlos syndrome, classic type
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C4552122/

- Genetic Testing Registry: Ehlers-Danlos syndrome, familial joint laxity type https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268349/
- Genetic Testing Registry: Ehlers-Danlos syndrome, hydroxylysine-deficient https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268342/
- Genetic Testing Registry: Ehlers-Danlos syndrome, musculocontractural type https://www.ncbi.nlm.nih.gov/gtr/conditions/C1866294/
- Genetic Testing Registry: Ehlers-Danlos syndrome, musculocontractural type 2 https://www.ncbi.nlm.nih.gov/gtr/conditions/C3809845/
- Genetic Testing Registry: Ehlers-Danlos syndrome, periodontal type, 2 https://www.ncbi.nlm.nih.gov/gtr/conditions/C4310681/
- Genetic Testing Registry: Ehlers-Danlos syndrome, procollagen proteinase deficient https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551623/
- Genetic Testing Registry: Ehlers-Danlos syndrome, progeroid type, 2 https://www.ncbi.nlm.nih.gov/gtr/conditions/C3809210/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 2 atypical https://www.ncbi.nlm.nih.gov/gtr/conditions/CN071419/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 3 https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268337/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 4 https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268338/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 4 variant https://www.ncbi.nlm.nih.gov/gtr/conditions/CN071423/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 5 https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268341/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 7A https://www.ncbi.nlm.nih.gov/gtr/conditions/C3508773/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 7B https://www.ncbi.nlm.nih.gov/gtr/conditions/C1851801/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type 8 https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551499/
- Genetic Testing Registry: Ehlers-Danlos syndrome, type vii, autosomal recessive https://www.ncbi.nlm.nih.gov/gtr/conditions/C2700425/
- Genetic Testing Registry: Ehlers-Danlos syndrome, unspecified autosomal dominant https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220679/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22ehlers-danlos+syndrome%22

Other Diagnosis and Management Resources

- GeneReview: Classic Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1244
- GeneReview: Hypermobile Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1279
- GeneReview: PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1462
- GeneReview: Vascular Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1494

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Ehlers-Danlos Syndrome
  https://medlineplus.gov/ency/article/001468.htm
- Encyclopedia: Hyperelastic Skin
  https://medlineplus.gov/ency/article/003280.htm
- Encyclopedia: Hypermobile Joints
  https://medlineplus.gov/ency/article/003295.htm
- Health Topic: Ehlers-Danlos Syndrome
  https://medlineplus.gov/ehlersdanlossyndrome.html

Genetic and Rare Diseases Information Center

- Arthrochalasia Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/2084/arthrochalasia-ehlers-danlos-syndrome
- Brittle cornea syndrome
  https://rarediseases.info.nih.gov/diseases/1019/brittle-cornea-syndrome
- Cardiac-Valvular Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/12613/cardiac-valvular-ehlers-danlos-syndrome
- Classical Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/2088/classical-ehlers-danlos-syndrome
- Classical-like Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/8507/classical-like-ehlers-danlos-syndrome

- Dermatosparaxis Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/2089/dermatosparaxis-ehlers-danlos-syndrome
- Ehlers-Danlos syndrome, dysfibronectinemic type
  https://rarediseases.info.nih.gov/diseases/8508/ehlers-danlos-syndrome-dysfibronectinemic-type
- Ehlers-Danlos syndromes
  https://rarediseases.info.nih.gov/diseases/6322/ehlers-danlos-syndromes
- Hypermobile Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/2081/hypermobile-ehlers-danlos-syndrome
- Kyphoscoliotic Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/2083/kyphoscoliotic-ehlers-danlos-syndrome
- Musculocontractural Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/8486/musculocontractural-ehlers-danlos-syndrome
- Periodontal Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/12474/periodontal-ehlers-danlos-syndrome
- Spondylodysplastic Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/9991/spondylodysplastic-ehlers-danlos-syndrome
- Vascular Ehlers-Danlos syndrome
  https://rarediseases.info.nih.gov/diseases/2082/vascular-ehlers-danlos-syndrome

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Heritable Disorders of Connective Tissue
  https://www.niams.nih.gov/health-topics/heritable-disorders-connective-tissue

Educational Resources

- MalaCards: ehlers-danlos syndrome
  https://www.malacards.org/card/ehlers_danlos_syndrome
- Merck Manual Consumer Version
  https://www.merckmanuals.com/home/children-s-health-issues/connective-tissue-disorders-in-children/ehlers-danlos-syndrome
- Orphanet: Ehlers-Danlos syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98249

Patient Support and Advocacy Resources

- Ehlers-Danlos National Foundation
  https://www.ehlers-danlos.com/
- Ehlers-Danlos Support Group (UK)
  https://www.ehlers-danlos.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/ehlers-danlos-syndrome/
- RareConnect
  https://www.rareconnect.org/en/community/ehlers-danlos-syndrome
- Resource List from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/ehlers.html
- The Hypermobility Syndromes Association (UK)
  http://hypermobility.org/

Clinical Information from GeneReviews

- Classic Ehlers-Danlos Syndrome
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- Hypermobile Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1279
- PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1462
- Vascular Ehlers-Danlos Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1494

Scientific Articles on PubMed

- PubMed
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Catalog of Genes and Diseases from OMIM

- BRITTLE CORNEA SYNDROME 1
  http://omim.org/entry/229200
- EHLERS-DANLOS SYNDROME WITH PLATELET DYSFUNCTION FROM FIBRONECTIN ABNORMALITY
  http://omim.org/entry/225310
- EHLERS-DANLOS SYNDROME, ARTHROCHALASIA TYPE, 1
  http://omim.org/entry/130060

- EHLERS-DANLOS SYNDROME, AUTOSOMAL DOMINANT, TYPE UNSPECIFIED
  http://omim.org/entry/130090
- EHLERS-DANLOS SYNDROME, BEASLEY-COHEN TYPE
  http://omim.org/entry/608763
- EHLERS-DANLOS SYNDROME, CARDIAC VALVULAR TYPE
  http://omim.org/entry/225320
- EHLERS-DANLOS SYNDROME, CLASSIC-LIKE
  http://omim.org/entry/606408
- EHLERS-DANLOS SYNDROME, CLASSIC TYPE, 1
  http://omim.org/entry/130000
- EHLERS-DANLOS SYNDROME, DERMATOSPARAXIS TYPE
  http://omim.org/entry/225410
- EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE
  http://omim.org/entry/130020
- EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC TYPE, 1
  http://omim.org/entry/225400
- EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC TYPE, 2
  http://omim.org/entry/614557
- EHLERS-DANLOS SYNDROME, MUSCULOCONTRACTURAL TYPE, 1
  http://omim.org/entry/601776
- EHLERS-DANLOS SYNDROME, MUSCULOCONTRACTURAL TYPE, 2
  http://omim.org/entry/615539
- EHLERS-DANLOS SYNDROME, PERIODONTAL TYPE, 1
  http://omim.org/entry/130080
- EHLERS-DANLOS SYNDROME, PERIODONTAL TYPE, 2
  http://omim.org/entry/617174
- EHLERS-DANLOS SYNDROME, SPONDYLODYSPLASTIC TYPE, 1
  http://omim.org/entry/130070
- EHLERS-DANLOS SYNDROME, SPONDYLODYSPLASTIC TYPE, 2
  http://omim.org/entry/615349
- EHLERS-DANLOS SYNDROME, TYPE V
  http://omim.org/entry/305200
- EHLERS-DANLOS SYNDROME, VASCULAR TYPE
  http://omim.org/entry/130050

Medical Genetics Database from MedGen

- Ehlers-Danlos syndrome
  https://www.ncbi.nlm.nih.gov/medgen/41720

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