Ehlers-Danlos Syndrome

Other Names

EDS; Ehlers Danlos Disease; E-D Syndrome

Category

Connective Tissue Diseases

Signs and Symptoms

The main symptoms of EDS involve the skin, muscles, skeleton, and blood vessels. Different types of EDS have different symptoms which are listed in the table below.

Most types of EDS symptoms include excessive flexibility of the joints (hypermobility). Many people with EDS also have soft, velvety skin that is very stretchy (elastic) and fragile, they tend to bruise easily, and some types of the condition also cause abnormal scarring

Types	Symptoms
Classical (cEDS)	Joint hypermobility, stretchy skin and abnormal scarring
Classical-like (clEDS)	Stretchy skin with velvety texture, but no abnormal scarring. Joint hypermobility, easily bruised and skin discolouration
Cardiac-valvular (cvEDS)	Causes severe problems with the valves that control the movement of blood through the heart. Also, skin that is thin, stretchy skin, abnormal scarring and bruising. Joint hypermobility
Vascular (vEDS)	Can cause unpredictable tearing (rupture) of blood vessels, leading to internal bleeding and other potentially life-threatening complications. Those with the vascular type EDS also have an increased risk of organ rupture, including tearing of the intestine and rupture of the uterus during pregnancy.
Hypermobile	Infants and children with hypermobility often have weak muscle tone, which can delay the development of motor skills such as sitting, standing, and walking. Loose joints are unstable and prone to dislocation and chronic pain
Arthrochalasia (aEDS)	Severe joint hypermobility. Infants have hypermobility and dislocations of both hips at birth, also stretchy skin
Dermatosparaxis (dEDS)	Loose skin that sags and wrinkles, and extra folds of skin may be present.

Kyphoscholiotic (kEDS)	Severe curvature of the spine that worsens over time and can interfere with breathing. Muscle weakness and hypermobility of the joints
Brittle Cornea Syndrome (BCS)	Thinness of the clear covering of the eye (the cornea) and other eye abnormalities
Spondylodysplastic (spEDS)	Short stature and skeletal abnormalities such as abnormally curved (bowed) limbs, muscle weakness
Musculocontractural (mcEDS)	Abnormalities of muscles, including hypotonia and permanently bent joints (contractures), characteristic facial features from birth, stretchy and easily bruised skin
Myopathic (mEDS)	Muscle weakness, hypermobility of finger joints, and shortening of finger muscles
Periodontal (pEDS)	Abnormalities of the teeth and gums

Cause/Inheritance

- Mutations in at least 19 genes have been found to be related to the different types of EDS.
- Inheritance can be either dominant (meaning only one inherited abnormal gene is needed to cause the condition, from one parent) or recessive (meaning two abnormal genes are needed to cause the condition, one from each parent)
    - Dominant types - classical, vascular, arthrochalasia, and periodontal forms of the disorder, and likely the hypermobile type
    - Recessive types - cardiac-valvular, dermatosparaxis, kyphoscoliotic, spondylodysplastic, and musculocontractural types of EDS, as well as brittle cornea syndrome
    - The myopathic type can have either dominant or recessive inheritance.

Treatment and Management

Treatments vary for each type of EDS and focus on avoiding serious or life-threatening complications. The main body systems which require treatment and/or management involve the skin, cardiovascular and musculoskeletal systems.

Skin

- Ascorbic acid (Vitamin C) may be recommended to help reduce the easy bruising that accompanies EDS.
- Any wounds, either surgical or accidental are stitched deeply, using a lot of stitches. Stitches are also used on the surface of the skin to realign skin to prevent scarring. Stitches are also left in for longer than usual to allow for better healing.

Cardiovascular

- EDS patients should have screening for high blood pressure and arterial disease and treatment should be started early on.
- Heavy sport and other strenuous activities should be avoided to ensure damage to blood vessels is avoided.

Musculoskeletal

- Patients may benefit from physical therapy, low-resistance exercise, and devices like braces, wheelchairs, and scooters.
- Calcium and vitamin D can help maximise bone density. DEXA bone density scans should be conducted every other year.

Treatments for other EDS types may include:

- Kyphoscoliotic type - patients should have routine eye exams as they are at risk for eyeball rupture, retinal detachment and glaucoma.
- Dermatosparaxis type- patients may benefit from protective bandages over exposed areas such as the skin of the elbows and knees.

Support Contacts International

- Ehlers-Danlos Society: a global community of patients, caregivers, health care professionals, and supporters, dedicated to saving and improving the lives of those affected by the Ehlers-Danlos syndromes, hypermobility spectrum disorders and related conditions. https://www.ehlers-danlos.com

Support Contacts Australia

- EDS Australia -This group is for Australians who have EDS and their families, a place for support, information, compassion and understanding. http://edsaus.ning.com

Facebook Support

- The Ehlers-Danlos Society https://www.facebook.com/ehlers.danlos/
- Ehlers Danlos Awareness https://www.facebook.com/EhlersDanlosAwarenessPage/

Sources

National Organisation for Rare Disorders (NORD) https://rarediseases.org/rare-diseases/ehlers-danlos-syndrome/
Genetics Home Reference: https://ghr.nlm.nih.gov/condition/ehlers-danlos-syndrome
Genetic and Rare Diseases Information Centre https://rarediseases.info.nih.gov/diseases/6322/ehlers-danlos-syndromes

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