

About the Types of Ehlers-Danlos

The types of Ehlers-Danlos syndrome may appear clear and defined, but symptomatology rarely permits easy classification. Although most of the genes responsible are identified, not all of the mutations that cause EDS have been found yet.

HYPERMOBILITY [*distinctive cause unidentified*]: Most common, 1 in 10,000 to 15,000. Generalized joint hypermobility, skin involvement (possible hyperextensibility or smooth/soft skin), chronic joint pain, recurrent joint dislocations.

CLASSICAL [*COL5A1, COL5A2, COL1A1*]: Two to 5 in 100,000. Variable skin hyperextensibility, widened atrophic scars (tissue fragility), joint hypermobility, easy bruising; ± mulluscoid pseudotumors, subcutaneous spheroids; occasional internal organ fragility.

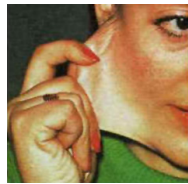
VASCULAR [*COL3A1*]: Rare at one in 100,000 to 250,000. Thin, translucent skin, arterial/intestinal/uterine fragility or rupture, extensive bruising, characteristic facial appearance; ± acrogeria, hypermobility of small joints, early onset varicose veins, and pneumothorax.

KYPHOSCOLIOTIC [*PLOD1*]: Very rare. Generalized joint laxity, severe muscle hypotonia at birth, scoliosis at birth (progressive), scleral fragility and rupture of the ocular globe; ± tissue fragility, easy bruising, arterial rupture and osteopenia.

ARTHROCHALASIA [*COL1A1 & COL1A2*]: Very rare. Severe generalized joint hypermobility with recurrent subluxations, congenital bilateral hip dislocation; ± skin hyperextensibility, tissue fragility (atrophic scars), easy bruising, muscle hypotonia and osteopenia.

DERMATOPARAXIS [*ADAMTS2*]: Extremely rare. Severe skin fragility, sagging redundant skin; ± soft doughy skin texture, easy bruising, premature rupture of fetal membranes, and large hernias (umbilical and inguinal).

OTHER reported types await definition or have been reported only in single families.



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EMERGENCY INFORMATION for VEDS

ARTERIAL RUPTURE IS THE MOST COMMON CAUSE OF SUDDEN DEATH.

- **Arterial or intestinal rupture commonly presents as acute abdominal or flank pain that can be diffuse or localized.**
- **Spontaneous arterial rupture is most likely to occur in a person's twenties or thirties, but can occur at any point in life.**
- **Cerebral arterial rupture may present with altered mental status and be mistaken for drug overdose.**
- **Mid-size arteries are commonly involved.**

Arterial, intestinal, or uterine fragility or rupture usually arise in EDS Vascular type, but should be investigated for any EDS type.

CAROTID-CAVERNOUS FISTULA: LIFE-THREATENING EMERGENCY

Redness, pain and prominence of one or both eyes and the sound of pulsations in their head can be manifestations of a life-threatening carotid-cavernous fistula:

- High pressure blood from the internal carotid artery can shunt blood inappropriately into the tissue around the eyes and into the eye itself, causing the symptoms;
- The life-threatening risk is that the high pressure blood will leak out of the confines of the blood vessels.

Seek immediate hospital-based medical attention, and inform emergency medical staff of the patient's VEDS and the critical risk of a carotid-cavernous fistula.

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Medical Resource Guide



Ehlers-Danlos
NATIONAL FOUNDATION

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INTRODUCTION

More than 1.5 million people around the world have Ehlers-Danlos syndrome (EDS).

Someone with EDS is born about every half hour.

Many won't know what's wrong with them...

Unless you recognize their EDS.

Those born with EDS can fight a wide range of problems daily, but many remain undiagnosed until a major medical emergency. Unaware that what they suffer isn't normal, some die without ever knowing differently, even though most forms of EDS can be seen in childhood. EDS affects more than one in 5,000 men and women, of every race and ethnicity.

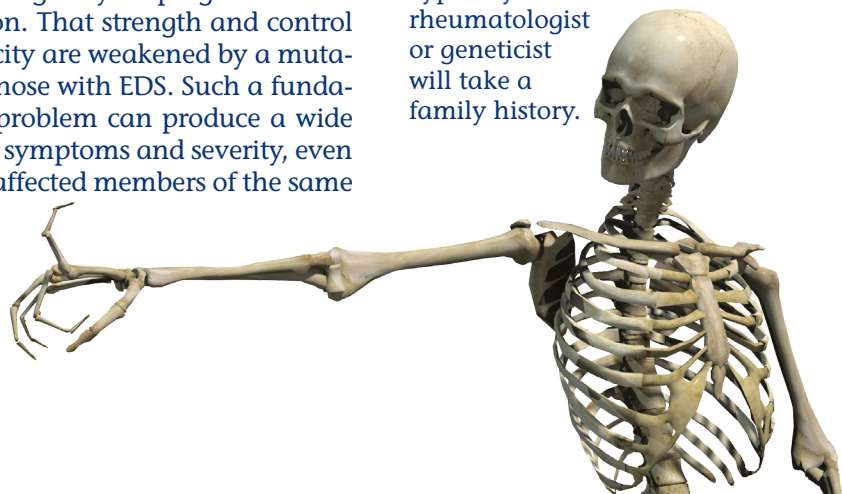
Ehlers-Danlos syndrome is a group of at least six inherited collagen disorders; each type is thought to be the result of a unique defect, but not all of the genes involved have been found.

Collagen usually gives connective tissue strength by helping it resist deformation. That strength and control of elasticity are weakened by a mutation in those with EDS. Such a fundamental problem can produce a wide range of symptoms and severity, even among affected members of the same family.

DIAGNOSTIC ADVICE

Listen to your instinct if you suspect a collagen disorder like EDS: early diagnosis is crucial to positive patient health. EDS cannot be diagnosed from this simple brochure, but some guidelines can be offered.

- The most common types of EDS include some degree of joint hypermobility, and may also involve:
 - pain, chronic and widespread;
 - joint instability;
 - spontaneous subluxations/dislocations.
- Current recommendations for assessing skin hyperextensibility are:
 - find test sites where skin is not damaged/scarred nor where there is redundant skin as on the back of the hand or the elbow;
 - the usual choice is the palm side of the forearm, where normal is up to 1–1.5 cm stretch;
 - pull the skin outward until there is resistance;
 - not every site must show hyperextensibility.
- Genetic testing at present can be useful for determining some EDS types, but cannot rule out the presence of EDS.
- Typically a rheumatologist or geneticist will take a family history.



- Hypermobility is tested using the Beighton scale, which adds a point for extreme range-of-motion in each of nine joints; results should be viewed with these cautions:
 - high Beighton scores should never be the sole criterion for EDS diagnosis;
 - children are naturally more hypermobile than adults;
 - aging adults score as less hypermobile than earlier in life;
 - there are hypermobile joints not tested by Beighton that should also be considered.

TREATMENT OPTIONS

Diagnosis of EDS is the beginning, because although there is no cure, many of the effects are treatable.

- Baseline echocardiogram should be performed and routine follow-ups scheduled:
 - retest children with normal results every one to two years, and adults with normal results every three to five years;
 - any enlargement of the aorta should be monitored every one to two years.
- People with EDS are predisposed to:
 - functional bowel disorders (gastroparesis, irritable bowel syndrome);
 - cardiovascular autonomic dysfunction (postural orthostatic tachycardia syndrome, neurally mediated hypotension). Many with EDS experience low blood pressure, lightheadedness, and

fast heart rates. Treat low blood pressure by increasing salt and fluids; medications may have side effects.

- Low bone density is common in EDS. Consider a DEXA scan at diagnosis, particularly in the presence of risk factors, such as someone with reduced activity, and certainly no later than midlife.
- It is important to find an accommodating dentist because of temporomandibular disorders, facial/jaw pain, bone/tooth density issues, and gum disease that is particularly exacerbated by neglect.
- A physical therapist who is highly knowledgeable and deeply understanding about connective tissue and joint dysfunction can be useful to long-term health:
 - use low resistance exercise to gradually increase muscle tone and to help stabilize loose joints;
 - minimize joint trauma by avoiding joint hyperextension and any high resistance or impact activities;
 - just as it often takes years for joint laxity to cause significant pain and instability, it can take at least months and usually years to gradually reverse the process via muscle toning exercise.
- Both psychologist and psychiatrist are recommended for simple quality of life and sleep disturbance. EDS probably contributes organic problems, and poor mental health frequently results from enduring constant physiological failures.

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This **Medical Resource Guide** brings you highlights of information found at **ednf.org**, available whenever and however you need important resources about EDS, from a quick summary for a patient waiting in your office, to in-depth articles and media. EDNF publishes **Medical Resource Guides**.

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