How is vEDS managed?

vEDS can cause a variety of symptoms in many different areas of the body, so people with vEDS may require multiple providers in different specialties to manage their care.

Key aspects of care focus on monitoring and managing arterial and organ fragility. It is recommended that people with vEDS have an emergency plan in place and make lifestyle modifications to minimize the risk of injury. Each person's care plan should address their individual needs.

How is vEDS inherited?

vEDS is inherited in an autosomal dominant pattern. This means if a person inherits the genetic variant from one of their parents, they will have vEDS. Each child of a parent with vEDS will have a 50% chance of having vEDS.

About half of people with vEDS are the first person in their family to have vEDS due to a random mutation. This is called a de novo mutation. Once a genetic variant is introduced through a de novo mutation, it can be inherited in an autosomal dominant pattern.

What resources are available to people with vEDS?

The Ehlers-Danlos Society offers a variety of resources for those affected by vEDS around the world.

- Helpline
- vEDS and vEDS parents Virtual Support Groups
- Healthcare Professionals Directory
- Support Group and Charity Directory
- Inspire
- Videos
- EDS ECHO

For more information on vEDS, scan the QR code below

The Ehlers Danlos Society

What is Vascular Ehlers-Danlos Syndrome (vEDS)?

We CARE For vEDS

What are the key signs and symptoms of vEDS?

vEDS may be suspected if a person has:

- A family history of vEDS
- Arterial rupture at a young age
- Bowel perforation
- Uterine rupture during pregnancy
- Carotid-cavernous sinus fistula

People with vEDS may also have:

- Easy bruising, unrelated to injury or at unusual sites
- Thin, translucent skin with visible veins
- Characteristic facial features, such as:
  - Thin lips
  - Narrow nose
  - Appearance of large eyes
  - Attached earlobes
- Spontaneous pneumothorax
- Small joint hypermobility
- Clubfoot

How is vEDS diagnosed?

Genetic testing is used to see if a person has the genetic variants that cause vEDS.

Genetic testing should be considered if a person has any of the major criteria or several minor criteria, particularly in people under 40.

Diagnostic Criteria for vEDS

Major Criteria

- Family history of vEDS with documented causative variant in *COL3A1*
- Arterial rupture at a young age
- Spontaneous sigmoid colon perforation in the absence of known diverticular disease or other bowel pathology
- Uterine rupture during the third trimester in the absence of previous C-section and/or severe peripartum perineum tears
- Carotid-cavernous sinus fistula (CCSF) formation in the absence of trauma

Minor Criteria

- Bruising unrelated to identified trauma and/or in unusual sites such as the cheeks and back
- Thin, translucent skin with increased venous visibility
- Characteristic facial appearance
- Spontaneous pneumothorax
- Acrogeria
- Talipes equinovarus
- Congenital hip dislocation
- Hypermobility of small joints
- Tendon and muscle rupture
- Keratoconus
- Gingival recession and gingival fragility
- Early-onset varicose veins (under age 30 and starting prior to pregnancy if female)

The Ehlers Danlos Society

We CARE For vEDS

What is vEDS?

vEDS // VASCULAR EHLERS-DANLOS SYNDROME

vEDS is a genetic disorder that causes connective tissue to be fragile, particularly in the blood vessels and organs. The complications of vEDS can be life-threatening and include aneurysm, dissection, and rupture of the arteries and rupture of organs. vEDS may also cause a variety of other symptoms, including extensive bruising and spontaneous pneumothorax.

What is the prevalence of vEDS?

vEDS is a rare disorder that affects roughly 1 in 100,000 – 200,000 people.

What causes vEDS?

vEDS is caused by differences in the genes called genetic variants. These genetic variants affect the connective tissue, which provides support, protection, and structure throughout the body.

vEDS is caused by genetic variants in the *COL3A1* gene. In extremely rare cases, there are also specific variants in the *COL1A1* gene that cause a genetic condition that can present similarly to vEDS.

GET IN TOUCH

For more information on The Ehlers-Danlos Society's work, to get involved, or to donate, please visit:

www.ehlers-danlos.com

/ehlers.danlos
/ehlers.danlos
/theedsociety
/edsocietyyoutube