

The vascular form of Ehlers-Danlos syndrome (vEDS) is a connective tissue disorder that can affect many different parts of the body, including the skin, arteries, muscles, and organs. Connective tissue is a type of tissue in the body that helps to hold everything together, like a glue for your body.

People with vEDS can have some or many signs and symptoms, including skin issues (thin, translucent skin that shows signs of premature aging), easy bruising, hypermobile (extra flexible) fingers and toes, joint dislocations, collapsed lung, receding gums, a higher risk for an [aneurysm](#), and a characteristic facial appearance (that can include thin lips, a small chin, a thin nose, and large eyes). Not all people with vEDS will have all of these signs, and two people with vEDS may have very different symptoms, even within the same family. Individuals with vEDS also have a higher chance for their internal organs (particularly the uterus during pregnancy), arteries, and intestines to rupture due to the connective tissue not holding them together as well as it should. Babies who have vEDS have a higher chance to be born with clubfoot and congenital (from birth) hip dislocation.

Causes

Over 95% of cases of vEDS can be linked to pathogenic variants in the [COL3A1](#) gene, which are inherited in an [autosomal dominant](#) pattern. This means that a single copy of the pathogenic variant is enough to cause an individual to develop the condition, and anyone who carries the pathogenic variant has a 50% chance to pass it down to any children they have. The remaining individuals may have a genetic link to their vEDS, but it is in a gene that we do not yet know about. vEDS affects approximately 1 in 50,000 to 1 in 200,000 people. Because of the variability of vEDS and the thought that it is very rare, it is likely under-diagnosed in families that have non-vascular health issues. About 50% of people who have vEDS due to the [COL3A1](#) gene have a parent who also has it. The other 50% are the first ones in their family to have it (called *de novo*).

Diagnosing vEDS

While genetic testing for vEDS can be helpful to establish a diagnosis, medical providers may use other pieces of information from other lab tests or someone's medical and family history to make the diagnosis. Some red flags that can increase the chance for vEDS in a family include:

- [Aneurysms](#) (a bulging out on a blood vessel) that can rupture
- Rupture of the intestines
- Rupture of the uterus during pregnancy

- Other people in the family with a diagnosis or suspicion of vEDS
- Hypermobile joints/frequent joint dislocations
- Thin skin that bruises easily
- Collapsed lung
- Early-onset varicose veins

Many families may have these red flags in their family history and DO NOT have vEDS. However, someone with a strong pattern of these or other heart issues may be at a higher chance to have vEDS, and may benefit from talking about it more with a specialist, such as a cardiologist or a genetic counselor.

Medical Management for vEDS

Treatment for vEDS can sometimes vary depending on the individual person and their specific health concerns, and should be discussed with a medical provider who is familiar with vEDS. Some options for medical management can include using imaging, such as [ultrasound](#) and [magnetic resonance angiography](#), to try to see all of the arteries in the body to look for [aneurysms](#) or other abnormalities, and surgery can sometimes be required immediately if there is a rupture of an internal organ, artery, or intestines. Because of this increased risk for random rupture, people with vEDS should seek immediate medical attention for any sudden unexplained pain.

People with vEDS should try to avoid high-contact sports (such as football and rugby), as well as heavy weight lifting or weight training. Because of the increased risk for related complications, it may also be recommended that individuals who have vEDS avoid routine [colonoscopy screening](#) as well as any surgeries that are not absolutely necessary. It is important for people with vEDS to monitor their blood pressure. If their blood pressure is high, that can increase the chance for the veins to rupture, so if high blood pressure is found early treatment can be helpful to reduce the risk for complications.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Additional Resources

[Ehlers-Danlos Society Headquarters](#)

[Ehlers-Danlos Support Group](#)

[Ehlers-Danlos Syndrome Network C.A.R.E.S. Foundation](#)