

What is Vascular Ehlers-Danlos Syndrome (vEDS)?

Vascular Ehlers-Danlos Syndrome (vEDS) is an inherited connective tissue (CT) disorder that is caused by defects in collagen, an important protein that provides strength and resistance to CT in order to support and connect the organs and other structures of the body. It is generally considered the most severe form of Ehlers-Danlos syndrome (EDS). Common symptoms include thin, translucent skin, easy bruising, characteristic facial appearance, and fragile arteries, muscles and internal organs. vEDS is reported to affect 1 in 50,000 to 200,000 individuals.

How is it Transmitted?

Vascular EDS (vEDS) is an autosomal dominant condition caused by a mutation (change) in the COL3A1 gene. This means that if a child receives a copy of the mutated gene, they will be affected by vEDS. Also, a parent with vEDS has a 50% chance of passing it onto their children. Approximately half of patients inherit the pathogenic (disease-causing) variant from a parent, while the other half develop new (spontaneous) pathogenic variants.

How is it Diagnosed?

Diagnosis of vEDS is based on specific major and minor clinical features. It should be suspected if a person has at least one major feature, such as early arterial rupture or dissection (before age 40), intestinal rupture, uterine rupture during pregnancy without a prior C-section, or a family history of vEDS. It may also be suspected if several minor features are present, including easy bruising, thin or translucent skin, clubfoot, small joint hypermobility (fingers and toes), or congenital hip dislocation. The diagnosis is confirmed through genetic testing that identifies a disease-causing variant in the COL3A1 gene.

Is There a Treatment for vEDS?

Currently, there is no cure for vEDS. The treatment of vEDS relies on careful surveillance, lifestyle adaptations, and treatment of the manifestations. Surveillance includes regular arterial screening with ultrasounds and blood pressure monitoring. Affected individuals are instructed to seek immediate medical attention for sudden, unexplained pain. Treatment may include medical or surgical management for arterial complications, bowel rupture, or uterine rupture during pregnancy. In the coming years, gene therapy may become an option.

What Can I do Every Day?

- 1 Always carry an alert card and/or wear medical alert jewelry.
- 2 Follow prescribed medical follow-up, in collaboration with your attending physician.
- 3 Inform the people around you about warning signs, so that they can help you, if necessary.
- 4 Let them know you have an alert card or medical alert jewelry.
- 5 Inform your doctor if you are planning to become pregnant or are in early pregnancy.

What Physical Activity Can I Practice?

Do: Participate in regular, light to moderate physical activity where you can speak easily without stopping to catch your breath. Examples include walking, light jogging, cycling, swimming, light weightlifting, yoga, pilates, and tai chi. Attend rehabilitation programs to manage pain and increase muscular strength.


Avoid: Competitive and contact sports (ex: football, hockey); sports with a high risk of shock; heavy lifting; repetitive or excessive joint movement; activities that lead to a sudden or large increase in blood pressure; muscle straining.

Depending on the involvement of the blood vessels, some patients will have to be more careful and do even lighter exercises.

vEDS Syndrome Manifestation & Recommendations

Body System & Manifestations

Treatment and Recommendations



Cardiovascular (Heart & Blood Vessels):

Arterial rupture, aneurysm (bulge), and/or dissection (tear); rupture of the chordae tendineae (cords that connect to heart valves) or ventricle; varicose veins and chest pain


Imaging: X-rays, CT scans and MRIs are done to image the heart and arteries. Echocardiograms should be performed at least every three years if normal at baseline. Conventional arterial angiography should be discouraged because it has been associated with added new complications. **Medication:** Beta-blockers and angiotensin converting enzyme inhibitors or angiotensin 2 receptor blockers might be beneficial to reduce the progression of arterial dilation and improve cardiac function. **Surgery:** Conventional or endoscopic surgery might be a lifesaving approach in the case of a life-threatening arterial aneurysm (bulge), rupture, or dissection (tear). Alternative methods include tourniquet occlusion, stent placement, and digital compression of the affected artery. Radiofrequency or laser energy can be used in the treatment of larger varicose veins.



Gastrointestinal:

Perforation (holes) in the gastrointestinal tract; abdominal pain; constipation/diarrhea; heartburn (reflux disease); nausea/vomiting

Imaging: Chest x-rays and CT scans are used to image the intestines and colon, make a diagnosis, and identify possible perforation. pH studies may be used to evaluate acid reflux. **Medication:** Proton pump inhibitors and histamine H2 blockers are commonly used for patients with GI symptoms and discomfort. Stool softeners, laxatives, and increased fluid intake may help manage constipation. **Surgery:** Often necessary and can be lifesaving. Interventions include partial colectomies (partial removal of colon), colostomies (creating an abdominal opening to the colon), and Hartmann's procedure (where the rectum is sealed and waste is diverted to a colostomy bag, with possible reversal after healing). Avoid routine colonoscopy as they could increase the risk of bowel perforation (holes) in patients with vEDS.



Lungs:

Spontaneous/recurrent pneumothoraxes (collapse of lung); hemothorax (blood between lung and chest wall); dyspnea (shortness of breath); sleep apnea; emphysema

Imaging: Chest x-ray, non-contrast computed tomography, CT, or MRI of the thorax can be used to image the lungs and make a diagnosis. Bronchoscopy may also be performed but careful consideration must be taken. **Surgery:** Temporary tube insertion into the chest may be needed to treat and drain pneumothorax and hemothorax. Lung transplantations may also be performed as a last resort. In case of sudden chest pain and difficulty breathing, call 911 or local emergency phone number to be transferred to hospital. In case of excessive fatigue during the day, talk to your doctor.



Eyes:

Keratoconus (thinning of the cornea); carotid cavernous fistulas (abnormal artery-vein connection near eye); bleeding in/around the eye; difficulty closing eyes

Treatment: Corrective lenses or glasses may be given to treat keratoconus. If it worsens, a cornea transplant or corneal collagen cross-linking may be necessary. Routine eye check ups are essential. Any visual disturbance, sudden onset should raise fears of a complication. Immediately call an ophthalmologist for an emergency consultation.

Other potential manifestations: periodontal disease; gingival recession; spleen rupture; liver rupture

**This is not an exhaustive list of manifestations. vEDS patients will not present with all of these manifestations, and having these manifestations does not mean a person has vEDS.*

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