

## ESSENTIAL VASCULAR DEFINITIONS

The following is condensed from *Understanding Vascular Complications: A Primer of Essential Definitions*, written by James H. Black III, MD, FACS, with illustrations by Jennifer Fairman. Their complete primer is available at [www.ednf.org](http://www.ednf.org); click on the red "Urgent Information on Vascular Complications" button for access.

### **Blood vessels consist of three layers:**

- the innermost *intima* is only a few cell layers thick;
- the *media* ("middle") layer is mostly special muscle cells that provide elasticity; and
- the outermost layer, the *adventitia*, is primarily connective tissue.

### **Aneurysm**

True aneurysms occur in arteries and are defined as a dilatation of the blood vessel wall, but with all three layers intact. In EDS aneurysm rupture is unpredictable and may occur at any diameter. While aneurysm rupture is a life-threatening condition, fortunately true aneurysm formation is relatively rare in EDS, occurring in approximately 15% of patients.

### **Dissection**

Arterial dissection refers to a tear in the intima; the tear leads to a breach in the three layers of the blood vessel wall, causing two passageways for the flow of the blood. The true lumen is the normal passageway with all three layers intact; the passageway outside the tear is missing the intima.

Aortic dissections are occasionally asymptomatic but more commonly cause an array of symptoms depending on the location and extent of the tear. A dissection may cause pain and may compromise blood flow to the extremities or internal organs. With time the weakened wall may expand to become a dissection with an aneurysm component as well.

### **Pseudoaneurysm**

Pseudoaneurysms ("false" aneurysms) are a contained rupture of a blood vessel. All three vessel layers are disrupted, so blood pulses into the space outside the vessel. Surrounding hematoma and tissue typically contain the blood flow; but the most dreaded complication remains free rupture of the artery with life-threatening bleeding.

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## EHLERS-DANLOS SYNDROME

### **Collagen is the most abundant protein in the human body.**

It provides the structural strength in most human tissue, including the heart and blood vessels, eyes and skin, cartilage and bone.

### **What happens when this basic building block is flawed?**

When muscles, ligaments, tendons and even large organs are built with structurally defective collagen there is systemic weakness and instability evident throughout the body.

### **There is Ehlers-Danlos syndrome.**

At least six types of EDS have been identified; clinical manifestations vary according to type. Each type is thought to involve a unique defect in connective tissue, although not all of the genes responsible for causing EDS have been found.

### **Vascular Type EDS is particularly destructive because of arterial or organ rupture.**

VEDS is caused by structural defects in the  $\alpha 1(\text{III})$  chain of collagen type III, encoded by the COL3A1 gene; it is inherited in an autosomal dominant manner. Within each family the EDS type runs true, but family members may vary in clinical severity and manifestations. As a group of genetic connective tissue disorders, the current estimated incidence of EDS is 1 in 2,500 to 5,000. It is known to affect men and women of all racial and ethnic backgrounds.

*Vascular Type EDS Medical Resource Guide is supported by*



Since 1889, millions of people from all walks of life have found answers at Mayo Clinic. Patients have access to a team of specialists from over 100 medical and surgical departments who take the time to listen and thoroughly understand their health issues and concerns.

Mayo Clinic treats Vascular Ehlers-Danlos and other aneurysmal diseases. To learn more visit <http://www.mayoclinic.com/health/ehlers-danlos-syndrome/DS00706> or visit the links at [ednf.org](http://ednf.org).

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# DIAGNOSIS of VASCULAR EDS

## CLINICAL SUSPICION

Generally there are three contexts for VEDS diagnoses:

1. an individual at 50% (sometimes less) risk in a family with good documentation of a history of VEDS;
2. an individual that presents with the characteristic major complications of the disorder in the absence of a family history; or
3. an individual with enough clinical features (history of bruising, thin appearing skin, etc.) to raise concern even with no family history.

AND

## TWO MAJOR CRITERIA

Major criteria:

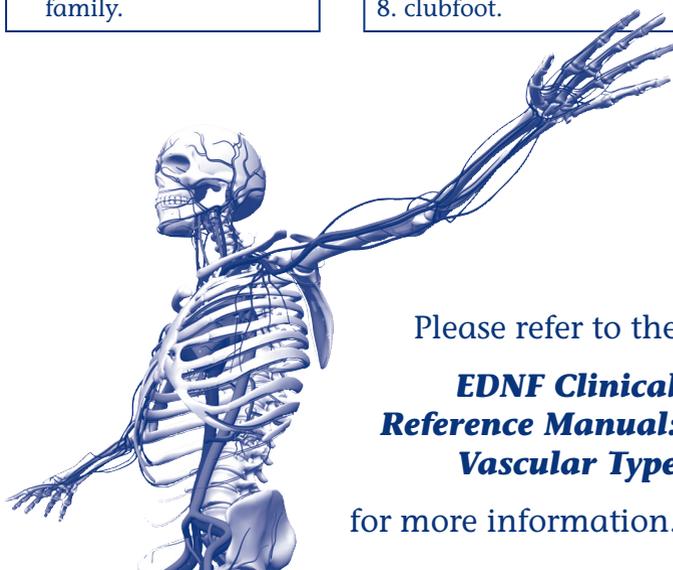
1. arterial, intestinal, and/or uterine rupture;
2. family history of VEDS, critical in diagnosis:
  - it determines if there is a history of VEDS-congruent complications in the family;
  - it aids identification of individuals who should also be studied; and
  - it provides some perspective on the natural history in the family.

OR

## ONE MAJOR CRITERION AND TWO MINOR CRITERIA

Minor criteria:

1. thin & translucent skin;
2. characteristic facial appearance (*large eyes, thin/narrow nose, thin lips, & small chin*);
3. extensive bruising;
4. arterial, intestinal, or uterine rupture;
5. aged-appearance of the hands (*acrogeria*);
6. small joint (finger) hypermobility;
7. congenital dislocation of the hip;
8. clubfoot.



Please refer to the  
**EDNF Clinical Reference Manual: Vascular Type**  
for more information.

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# TREATMENT GUIDANCE

Confirm diagnosis (*not practical in emergencies!*) with:

- biochemical testing to demonstrate abnormal type III collagen, and
- genetic testing to confirm a mutation in the COL3A1 gene which encodes type III collagen.

Additional clinical features of VEDS may include large joint hypermobility and very visible venous patterning. Prominent varicose veins in young individuals are common features. Abnormal scar formation after trauma or surgery is often present.

After diagnosis, the first priority is assembling an integrated care team to provide a clinical and social support network for the individual and family.

There is no settled care plan. Some have head-to-toe MRA or CTA scans to search for asymptomatic dissections, aneurysms, or vascular malformations; these results can be compared to future scans.

Counseling should include a frank discussion of the potential for catastrophic complications during pregnancy and other pregnancy risks. Also address the possibility of major depression precipitated by lifestyle modifications and lifespan concerns.

- Regular anti-hypertensive medications should be monitored for a goal of <130mmHg systolic.
- Patients with VEDS should see an ophthalmologist annually even if apparently free of disease, more frequently if they show evidence of ophthalmologic sequelae.
- Avoid intramuscular and subcutaneous injections (particularly of heparin or heparin substitutes) as they can cause massive hematoma and bruising.
- Avoid invasive diagnostic tests and unnecessary diagnostic angiography; perform central vein catheterizations under ultrasound guidance.
- VEDS patients should engage in low-impact activities and avoid contact sports (those involving collision or moderate-to-high static or dynamic force).
- An emergency medical alert bracelet or system is highly recommended.

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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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