Vascular Ehlers-Danlos syndrome

What is Vascular Ehlers-Danlos syndrome (vEDS)?

Vascular EDS is a distinct type of EDS caused by faulty collagen III. Vascular EDS can be very variable even within the same family. It is a rare condition and therefore many health professionals will not have seen someone with this diagnosis.

What are the main symptoms of vEDS?

Individuals with Vascular EDS may have the following features:

- Tendency to bruise very easily because the blood vessels are more fragile.
- Thin skin which makes small blood vessels visible on the upper chest and legs.
- Fragile blood vessels which can lead to major complications, including rupture of blood vessels.
- Risk of damage to hollow organs, such as bowel perforation or uterine rupture (where part of the womb tears).

What causes vEDS?

Vascular EDS is a genetic condition caused by an alteration, also known as a mutation, in a gene called COL3A1. This gene is the instruction for making collagen type III. When this gene is altered it causes a lack or deficiency of this collagen. This leads to disordered packing of collagen fibres making the connective tissue less effective, particularly in blood vessels, hollow organs and the skin.

What follow-up is recommended for people with vEDS?

Individuals should be referred to a cardiologist to discuss the available options for heart and blood vessel monitoring and whether medication is indicated.

Blood pressure should be checked regularly by the GP. Treatment can be used to lower the blood pressure, if necessary.

Any unusual signs or symptoms should be investigated thoroughly because of the possibility of internal bleeding.

If any surgery or invasive procedures are being considered, the surgeon must be made aware of the diagnosis of Vascular EDS and should be encouraged to seek specialist advice.

We encourage wearing Medic Alert bracelets and carrying a medical information card in case of emergencies.

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What is Ehlers-Danlos syndrome (EDS)?

EDS is an officially recognised, multisystemic, inherited connective tissue disorder. Mutations of certain genes cause abnormal connective tissue synthesis, resulting in abnormal fragile, hyper-extensible tissue. Main symptoms include; easy bruising, atrophic (depressed) scarring and delayed wound healing. The syndrome is named after Danish and French dermatologists, Edvard Ehlers and Henri-Alexandre Danlos. Edvard Ehlers first described the syndrome as a separate entity in 1901.

There are currently 6 main types of EDS:
- the arthrochalasia type
- the classic type
- the dermatosparaxis type
- the hypermobility type
- the kyphoscoliosis type
- the vascular type

Other forms of the condition may exist, but they have been reported only in single families or are not well characterized.

Some forms of EDS, notably the vascular and kyphoscoliosis types, can involve serious and potentially life-threatening complications. Blood vessels can tear (rupture) unpredictably, causing internal bleeding, stroke, and shock. The vascular type of EDS is also associated with an increased risk of organ rupture, including tearing of the intestine and rupture of the uterus (womb) during pregnancy. People with the kyphoscoliosis form of EDS experience severe, progressive curvature of the spine that can interfere with breathing.

How common is EDS?

<table>
<thead>
<tr>
<th>Major Type</th>
<th>Incidence</th>
<th>Inheritance</th>
<th>Causative Genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Classical</td>
<td>1/20,000</td>
<td>AD</td>
<td>COL5A1/COL5A2</td>
</tr>
<tr>
<td>Hypermobility</td>
<td>1/5,000</td>
<td>AD</td>
<td>- UNKNOWN GENE-</td>
</tr>
<tr>
<td>Vascular</td>
<td>1/50,000</td>
<td>AD</td>
<td>COL3A1</td>
</tr>
<tr>
<td>Kyphoscoliotic</td>
<td>1/100,000</td>
<td>AR</td>
<td>PLOD1</td>
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<tr>
<td>Arthrochalasia</td>
<td>30</td>
<td>AD</td>
<td>COL1A1/COL1A1</td>
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<tr>
<td>Dermatosparatic</td>
<td>8</td>
<td>AR</td>
<td>ADAMTS2</td>
</tr>
</tbody>
</table>

(AD) Autosomal Dominant inheritance — Condition develops even if one abnormal gene is inherited

(AR) Autosomal Recessive inheritance — 2 copies of the abnormal gene need to be inherited for the condition to develop