Annabelle’s Challenge is a UK registered charity raising awareness of Vascular Ehlers-Danlos Syndrome (vascular EDS) to help aid early diagnosis and prevent misdiagnosis of this life threatening genetic condition. We provide help, support and advice to families affected by vascular EDS including 12 months free MedicAlert UK membership for vascular EDS patients when they join Annabelle’s Challenge.

Our aims
To advance the education of the general public and medical profession in all areas relating to vascular EDS.

The relief of sickness and preservation and protection of good health by the provision of funding for the development of research and early diagnosis of vascular EDS.

To promote and protect the physical and good mental health of sufferers of vascular EDS, their families and carers through the provision of financial assistance, support, education and practical advice.

Vascular Ehlers-Danlos Syndrome
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What is vascular EDS?

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. Collagen is a tough, fiber-like protein that makes up about a third of our body protein. When this gene is altered it causes a lack or deficiency of this collagen making the connective tissue less effective, particularly in blood vessels, hollow organs and the skin.

- Estimated to be present in 1 in 250,000 of the UK population.
- Very hard to diagnose.

The vascular type accounts for less than 5% of all Ehlers-Danlos cases and is considered the most severe due to risk of life threatening vascular ruptures. It is a rare genetic condition and therefore many health professionals will not have seen someone with this diagnosis. When the diagnosis is known, management is more successful and survival is higher.

Possible Signs and Symptoms?

The signs & symptoms associated with vascular EDS can be different in each case and are not always present to warrant a diagnosis.

- Thin and translucent skin with visible underlying vessels.
- Easy bruising.
- Clubfoot, apparent at birth.
- Positive family history, sudden deaths in close relatives.

Facial features

Some individuals have the typical facial appearance which is characterised by a thin pinched nose, thin lips, prominent eyes, hollow cheeks, small earlobes and fine hair. Some of these features can be rather subtle, particularly in children.

Easy bruising

This can be seen at sites not prone to trauma and is often the presenting sign in childhood. Child protection agencies may have been or could be involved due to this prominent feature.

Skin involvement

The skin is thinner than normal resulting in a translucent appearance. This results in the underlying blood vessels being clearly visible, especially over the chest and extremities.

Vascular EDS in children

In children, the features most commonly identified as the reason for testing are family history, easy bruising, thin skin and club foot.