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My46 Trait Profile

Ehlers-Danlos syndrome - Vascular type

Other Names: Ehlers-Danlos syndrome (EDS) type IV

Ehlers-Danlos syndrome - vascular type is a genetic condition characterized by significant joint hypermobility (i.e., stretchy skin and easy bruising), as well as an increased chance for blood vessel, bowel, and organ rupture. It is caused by mutations in the COL3A1 gene which makes the collagen type 3 protein.

Characteristics of Ehlers-Danlos syndrome - Vascular type

Ehlers-Danlos syndromes are named after the physicians who first recognized significant joint hypermobility (i.e., stretchy skin and easy bruising) as a disorder. Several different forms (designated by roman numerals) have been recognized since then. EDS type IV is distinguished from other forms of EDS by the presence of large blood vessel rupture, thus earning the name Vascular Type, as well as bowel and organ rupture.

Most individuals with EDS type IV are identified by laboratory testing after experiencing a medical complication most often being a ruptured blood vessel or a tear or perforation of the bowel. Many individuals with EDS type IV will also have thin, fragile, translucent skin; easy bruising and small joint (finger) hypermobility. Fewer may have a characteristic facial appearance with deep-set eyes, a thin nose and delicate facial features. Because the clinical presentation can vary between affected individuals, it is not unheard of for very few of these features to be present. In infancy, clubfoot and/or dislocation of the hips at birth can be seen. In childhood, hernia, collapsed lung and repeated joint dislocations are commonly seen. A first major complication, either a ruptured artery or bowel rupture typically occurs in the late 20's or early 30's on average. Expansion or ballooning of an artery (also called an arterial aneurysm) may grow and result in rupture of the artery. For this reason, a fair number of individuals identified to have an arterial aneurysm at a young age will be tested for EDS type IV. Life expectancy, overall, is diminished as a result of the occurrence of these complications.

Diagnosis/Testing

To date, all individuals with EDS type IV have a change or mutation in a gene called COL3A1 which makes a collagen protein that is responsible for the strength and structure of connective tissue: blood vessels, bowel, skin and other organs. Mutations in the COL3A1 gene interfere with the amount of and the shape of the type III collagen protein in these tissues. This alters the strength and the integrity of these tissues.

Management/Surveillance

Management of EDS type IV often involves imaging (usually done by magnetic resonance imaging (MRI)) to look at the medium and large arteries of the body for evidence of aneurysm. If an aneurysm is found, periodic repeat MRI may be done to monitor the size and extent of the aneurysm. Surgical repair can be undertaken if necessary and must be carefully planned. It is important for individuals with EDS type IV to be closely followed by a medical care team familiar with their condition. Medical attention should be sought if the person experiences unexplained acute or immediate pain as it could indicate the onset of a complication.

Women with EDS type IV who become pregnant should seek the care of a fetal medicine specialist as there are known risks of artery or uterus rupture to be considered near the time of delivery.

Mode of inheritance

EDS type IV is inherited in an autosomal dominant pattern. This means that one COL3A1 mutation is enough for an individual to be affected. The mutation can be inherited from an affected parent (40-50% of the time) or can occur brand new (de novo) in an affected individual.

Risk to family members

The risk to family members depends on whether or not the individual with EDS type IV also has a parent affected with the condition. If a parent also has the EDS type IV, the risk of having a child with condition is 50% with each pregnancy. If a parent does not have EDS type IV, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Ehlers-Danlos National Foundation
http://www.ednf.org
EDS Cares Network
http://www.ehlersdanlosnetwork.org
Genetics Home Reference: Ehlers-Danlos syndrome
http://ghr.nlm.nih.gov/condition/ehlers-danlos-syndrome

References

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