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My46 Trait Profile

Ehlers-Danlos syndrome - Hypermobility type

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

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 III is mainly characterized by joint hypermobility.

Characteristics of Ehlers-Danlos syndrome - Hypermobility type

EDS type III is a highly complex connective tissue disorder – a condition that affects the joints, skin, and blood vessels. The most consistent feature is joint hypermobility. Individuals with EDS type III have joint instability that results in chronic pain. There is also a risk of recurrent joint dislocations or subluxations, especially of the shoulders and kneecaps. Other skeletal problems include an increased risk for curving of the spine (scoliosis) and an increased risk for low bone density. Soft velvety skin is a common feature, but typically the skin is not hyperextensible or as fragile as with other forms of EDS. Symptoms involving the gastrointestinal tract, such as heartburn (reflux), chronic diarrhea, and constipation are also common. Many female individuals also report difficult menses, with heavy bleeding, cramping, and feelings of pelvic congestion. Dizziness upon changing positions has been reported, and some individuals with EDS type III have been diagnosed with postural orthostatic tachycardia syndrome (POTS) – a condition where the heart rate increases dramatically upon standing. Many individuals with EDS type III also complain of intolerance to temperature changes resulting in changes in color of the skin of the lower legs. This results from a compromise of the involuntary (autonomic) nervous system which is known as autonomic dysfunction. Some individuals also report recurring headaches/migraines and chronic fatigue. High-arched palates, dental crowding, and a high frequency of bleeding gums have been reported. The severity and type of symptoms can vary among individuals even within the same family.

Diagnosis/Testing

Most individuals with EDS type III are diagnosed based on their symptoms. Since the genetic cause of EDS type III is still unknown, diagnosis is based entirely on clinical evaluation. The major diagnostic clinical criteria include joint hypermobility, soft skin with normal or only slightly increased extensibility, and an absence of fragility or other significant skin or soft tissue abnormalities. Other criteria which can be suggestive, though not diagnostic, include a family history of EDS type III, recurrent joint dislocations, joint/limb/back pain, easy bruising, bowel problems (constipation, diarrhea, irritable bowel syndrome), POTS, and a high, narrow palate.

Management/Surveillance

Management of EDS type III is largely dependent on symptoms. Physical therapy to build up global strength as well as the use of ergonomic tools (e.g. ergonomically designed pens, back supports) and braces/splints may help decrease pain and improve quality of life. Topical gels or oral pain medications may also be used. Bone density (DEXA) scans and vitamin D levels should also be monitored due to the increased risk for low bone density associated with joint hypermobility. Also, as there is an increased risk for aortic root enlargement and other cardiac problems such as mitral valve prolapse, a baseline echocardiogram should be performed. Affected individuals with POTS or dizziness upon

changing position may benefit from drinking electrolyte rich drinks or consuming salty foods or salt tablets. In some cases, medications are also needed to improve low blood pressure symptoms.

Mode of inheritance

EDS type III is inherited in an autosomal dominant pattern. This means that an individual with the condition has a 1 in 2 (or 50%) chance to pass the condition to each of his/her children. It is also possible that the condition can occur brand new (de novo) in an affected child.

Risk to family members

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

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