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CARRIER SCREEN

Ehlers-Danlos syndrome type VIIC

What is Ehlers-Danlos syndrome type VIIC?

Ehlers-Danlos syndrome type VIIC is an inherited connective-tissue disorder characterized by skin fragility, easy bruising, and distinctive facial features [1, 2]. Individuals with Ehlers-Danlos syndrome type VIIC have reduced production or activity of procollagen I N-proteinase, an enzyme that processes different procollagen molecules. Procollagens develop into collagens, which help make the skin strong and elastic[1, 2]. Abnormalities in the enzyme lead to improper formation of collagen and weakening of the connective tissues[2]. Ehlers-Danlos syndrome type VIIC is also known as dermatosparaxis type Ehlers-Danlos syndrome [2].

What are the symptoms of Ehlers-Danlos syndrome type VIIC and what treatment is available?

Ehlers-Danlos syndrome type VIIC is a disease with variable severity and age of onset. Symptoms may include [1, 2]:

- Extreme skin fragility
- Bruising
- Distinctive facial features
- Joint instability
- Umbilical hernia
- Blue sclera (white of the eye)

There is no cure for Ehlers-Danlos syndrome type VIIC. Treatment is supportive.

How is Ehlers-Danlos syndrome type VIIC inherited?

Ehlers-Danlos syndrome type VIIC is an autosomal recessive disease caused by mutations in the *ADAMTS2* gene[2]. An individual who inherits one copy of an *ADAMTS2* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *ADAMTS2* mutations, one from each parent, is expected to be affected with Ehlers-Danlos syndrome Type VIIC.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Ehlers-Danlos syndrome type VIIC?

Ehlers-Danlos syndrome type VIIC is a rare condition that can occur in individuals of all races and ethnicities.

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/ehlers-danlos-syndrome

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/ehlers-danlos-syndrome/

The Ehlers-Danlos National Foundation: http://www.ednf.org/

Ehlers-Danlos Syndrome Network C.A.R.E.S., Inc.: http://www.ehlersdanlosnetwork.org/typesofehlersdanlos.html

The Marfan Foundation: http://www.marfan.org/ehlers-danlos

References

1. Colige A. *et al*. Human Ehlers-Danlos syndrome type VII C and bovine dermatosparaxis are caused by mutations in the procollagen I N-proteinase gene. *Am J Hum Genet*. 1999 Aug;65(2):308-17.
2. ADAMTS2. *Genetics Home Reference*. Available at http://ghr.nlm.nih.gov/gene/ADAMTS2. Accessed on February 15, 2016