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. 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. Abnormalities in the enzyme lead to improper formation of collagen and weakening of the connective tissues. Ehlers-Danlos syndrome type VIIC is also known as dermatosparaxis type Ehlers-Danlos syndrome.

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:

- 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. 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.
Ehlers-Danlos syndrome type VIIC

Where can I get more information?

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/ehlers-danlos-syndrome/
The Ehlers-Danlos National Foundation: http://www.ednf.org/
The Marfan Foundation: http://www.marfan.org/ehlers-danlos

References