Genetics Uncoded:

NxGen MDx

Facts about

Ehlers-Danlos Syndrome Type VIIC

What Your Test Results Mean

Carriers show no symptoms of Ehlers-Danlos Syndrome Type VIIC (EDS-VIIC) and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Ehlers-Danlos Syndrome Type VIIC Explained

EDS-VIIC is an inherited condition that impairs the body's ability to properly make collagen, an important component of connective tissue. The result is called dermatosparaxis; affected individuals have soft, velvety, fragile skin that stretches and tears easily, and severe joint hyperextensibility. People with EDS-VIIC bruise and scar very easily, and their fragile connective tissue can lead to health complications such as spontaneous rupture of the bladder or diaphragm. EDS-VIIC does not affect intelligence or mental function and management is supportive.

How the Genetics Work

EDS-VIIC is an autosomal recessive disorder caused by variants in the *ADAMTS2* gene. In general, individuals have two copies of the *ADAMTS2* gene. Carriers of EDS-VIIC have a single variant in one copy of the *ADAMTS2* gene while individuals with EDS-VIIC have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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Revised 12/15/16