Genetics Uncoded:

NxGen MDx

Facts about Familial Dysautonomia

What Your Test Results Mean

Carriers typically show no symptoms of Familial Dysautonomia 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.

Familial Dysautonomia Explained

Familial Dysautonomia is an inherited nerve disorder that typically presents at birth and progresses with time. Individuals with Familial Dysautonomia have gastrointestinal dysfunction, vomiting crises, recurrent pneumonia, altered sensitivity to pain and temperature, and cardiovascular instability. The disease has been associated with a high incidence of sudden death associated with lung infections and sepsis, as well as unexplained causes.

Treatment is focused on managing symptoms of the disease. With improved supportive treatment, lifespan has increased with approximately 60% of individuals with Familial Dysautonomia surviving beyond age 20.

How the Genetics Work

The clinical features of Familial Dysautonomia can be explained by variants in the *IKBKAP* gene. In general, individuals have two copies of the *IKBKAP* gene. Carriers of Familial Dysautonomia have a single variant in one copy of the *IKBKAP* gene while individuals with Familial Dysautonomia 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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

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