NxGen MDx

Familial Dysautonomia

What Your Results Mean

Test results indicate that you are a carrier of familial dysautonomia. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of familial dysautonomia for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for familial dysautonomia, each of your children has a 1 in 4 (25%) chance to have the condition.

Familial Dysautonomia Explained

What is Familial Dysautonomia?

Familial dysautonomia is an inherited nerve disorder that typically presents at birth and progresses over time. This condition affects cells responsible for involuntary actions, such as digestion, breathing, production of tears, and regulating blood pressure and body temperature. The condition also affects the senses, including taste and the perception of pain, heat, and cold. Individuals with familial dysautonomia have gastrointestinal dysfunction, vomiting crises, recurrent pneumonia, cardiovascular instability, and developmental delays. Approximately one-third of affected individuals have intellectual disability. The disease is associated with a high incidence of sudden death caused by lung infections, sepsis, and unexplained causes.

Prognosis

With improved supportive treatment, lifespan has increased. Approximately 60% of individuals with familial dysautonomia survive past age 20.

Treatment

Treatment is focused on managing symptoms of the disease as there is no cure.

Resources

Familial Dysautonomia Foundation
http://www.familialdysautonomia.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/dysautonomia-familial/
National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com
© 2020 NxGen MDx
Revised: 7/2020