NxGen MDx

X-Linked Adrenoleukodystrophy

What Your Results Mean

Test results indicate that you are a carrier of X-linked adrenoleukodystrophy. Some female carriers can show symptoms in the form of gradual weakness with muscle spasms in the legs, known as spastic paraparesis, while others show no symptoms at all. Female carriers also have an increased chance to have a child with X-linked adrenoleukodystrophy. The chance to have a child with X-linked adrenoleukodystrophy depends on the sex of the child. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

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

Male children have a 50% chance to have X-linked adrenoleukodystrophy, and female children have a 50% chance to be carriers. Some female carriers can show symptoms of gradual weakness with muscle spasms in the legs, known as spastic paraparesis, while others show no symptoms at all. Prenatal testing for X-linked adrenoleukodystrophy is available, as well as testing shortly after birth to rule out a diagnosis in a child, particularly if the child is a male. In the absence of symptoms, reflexive testing of your male partner or donor for X-linked adrenoleukodystrophy is not recommended due to the X-linked inheritance pattern of the condition.

X-Linked Adrenoleukodystrophy Explained

What is X-Linked Adrenoleukodystrophy?

X-linked adrenoleukodystrophy is an inherited disorder that is more common in boys and primarily affects the nervous system and the adrenal glands of the kidneys. The severity of symptoms and age of onset vary greatly from person to person. Children with the cerebral form of X-linked adrenoleukodystrophy experience learning and behavioral problems that usually begin between the ages of four and 10. Symptoms include aggressive behavior, vision problems, difficulty swallowing, poor coordination, and impaired adrenal gland function. Milder forms of X-linked adrenoleukodystrophy can present later in life and may or may not be life-threatening. Symptoms include progressive stiffness and weakness in the legs (paraparesis), urinary and genital tract disorders, changes in behavior and thinking ability, and adrenocortical insufficiency.

Prognosis

Individuals with the cerebral form of X-linked adrenoleukodystrophy usually survive only a few years after symptoms begin, but they may survive longer with intensive medical support. Individuals with milder forms of X-linked adrenoleukodystrophy typically survive into adulthood.

Treatment

There is no cure for X-linked adrenoleukodystrophy. Individuals with cerebral disease benefit from supportive care. Stem cell transplantation may stop disease progression if performed during the initial stages of the disease. Individuals with milder forms of X-linked adrenoleukodystrophy may benefit from physical therapy, management of urologic complications, and family and vocational counseling. Adrenocortical insufficiency is treated with corticosteroids.

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Revised: 7/2020

Resources

Adrenoleukodystrophy Foundation
https://www.aldfoundation.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/x-linked-adrenoleukodystrophy
National Society of Genetic Counselors
https://www.nsgc.org/
Stop ALD Foundation
http://www.stopald.org/

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