Andermann Syndrome

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Andermann syndrome. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Andermann syndrome 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 are carriers for Andermann syndrome, each of your children has a 1 in 4 (25%) chance to have the condition

Andermann Syndrome Explained

What is Andermann Syndrome?

Andermann syndrome is a progressive inherited disorder characterized by nerve damage resulting in abnormal or absent reflexes, weak muscle tone, muscle wasting, severe progressive weakness, loss of sensation in the limbs, and tremors. Individuals with Andermann syndrome typically walk late and lose the ability to walk in their teenage years. As they get older, individuals with this disorder frequently develop joint deformities, which restrict the movement of certain joints, as well as scoliosis, which may require surgery. Andermann syndrome also results in abnormal function of certain cranial nerves, which emerge directly from the brain and extend to various areas of the head and neck. Cranial nerve problems may result in facial muscle weakness, drooping eyelids, and difficulty following movements with the eyes. Individuals with Andermann syndrome typically have an intellectual disability, which may be mild to severe, and some can additionally experience seizures. They may also develop psychiatric symptoms such as depression, anxiety, agitation, paranoia, and hallucinations, which usually appear in adolescence.

Prognosis

Prognosis for Andermann syndrome is unfavorable as it is associated with a shortened lifespan. Many individuals only live to their third or fourth decade of life

Treatment

Currently, there is no cure for Andermann syndrome; treatment is typically symptomatic and may include physical therapy to aid in movement or the use of a walking device. Individuals may additionally benefit from a psychiatric evaluation.

Resources

National Organization of Disorders of the Corpus Callosum
http://nodcc.org/

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/andermann-syndrome

National Society of Genetic Counselors
https://www.nsgc.org/

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