



Genetics Uncoded:

Facts about

Andermann Syndrome



What Your Test Results Mean

Carriers typically show no symptoms of Andermann syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Andermann Syndrome Explained

Andermann syndrome is an inherited disorder characterized by nerve damage, resulting in abnormal or absent reflexes, weak muscle tone, muscle wasting, severe progressive weakness and 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, people 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 usually have intellectual disability, which may be mild to severe, and some experience seizures. They may also develop psychiatric symptoms such as depression, anxiety, agitation, paranoia, and hallucinations, which usually appear in adolescence. There is no cure for Andermann syndrome; however, physical therapy may be useful to maintain movement as long as possible.

How the Genetics Work

Andermann syndrome is an autosomal recessive disorder caused by variants in the *SLC12A6* gene. In general, individuals have two copies of the *SLC12A6* gene. Carriers of Andermann syndrome have a variant in one copy of the *SLC12A6* gene while individuals with Andermann syndrome have variants in both copies of the *SLC12A6* gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.