Genetics Uncoded:

Facts about Segawa Syndrome

What Your Test Results Mean

Carriers typically show no symptoms of Segawa syndrome; however, carriers are at an increased risk of having a child with Segawa syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Segawa Syndrome Explained

Segawa syndrome is an inherited condition characterized by muscle rigidity, unusual limb positioning, a lack of coordination when walking or running, unusually slow movement, tremors, and an inability to hold the body upright and balanced. These symptoms range from mild to severe depending on the case. The symptoms of the disorder are caused by a deficient level of the enzyme tyrosine hydroxylase, preventing production of the neurotransmitter dopamine. Milder cases can be very successfully treated with medications. If symptoms have gone untreated for a while, some symptoms such as uneven gait and motor and speech difficulties may not reverse with medication but improve with physical, occupational, and/or speech therapy.

How the Genetics Work

Segawa syndrome is an autosomal recessive disorder caused by variants in the *TH* gene. In general, individuals have two copies of the *TH* gene. Carriers of Segawa syndrome have a single variant in one copy of the *TH* gene while individuals with Segawa syndrome 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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