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 1-855-776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.