NxGen MDx

Tyrosine Hydroxylase Deficiency

What Your Results Mean

Test results indicate that you are a carrier of tyrosine hydroxylase deficiency. Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for 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 and their own personal clinical management.

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 or donor are carriers for tyrosine hydroxylase deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Tyrosine Hydroxylase Deficiency Explained

What is Tyrosine Hydroxylase Deficiency?

Tyrosine hydroxylase deficiency 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. There are three types of tyrosine hydroxylase deficiency: TH-deficient dopa-responsive dystonia (DRD), infantile parkinsonism (IP), and progressive infantile encephalopathy (PIE). TH-deficient DRD is the mildest form of tyrosine hydroxylase deficiency, with symptoms typically appearing during childhood and mostly affecting movement. Children with TH-deficient DRD typically lack coordination when walking or running and have unusual limb positioning due to abnormal muscle contractions. These children can also have a postural tremor (involuntary shaking when trying to hold a position) or involuntary eye movements.

IP is a more severe form of tyrosine hydroxylase deficiency and individuals typically show symptoms of the condition within six months after birth. Symptoms include those seen in individuals with TH-deficient DRD as well as delayed movement milestones (such as sitting up) due to low muscle tone in the trunk of the body and stiff muscles (especially the limbs). Some children will also experience gastroesophageal reflux and difficulty regulating blood sugar (hypoglycemia), body temperature, and blood pressure. Some children also have attention deficits, learning disabilities, speech difficulties, and psychiatric symptoms such as depression, anxiety, or obsessive-compulsive tendencies. Some children who are more severely affected have an intellectual disability.

Individuals with the PIE form of tyrosine hydroxylase deficiency have the most severe symptoms and typically have profound physical and intellectual disabilities.

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Prognosis

With early diagnosis and treatment, the prognosis for individuals with the mild form of tyrosine hydroxylase deficiency (TH-deficient DRD) is good. If treatment of the condition is delayed, individuals may have some lifelong walking difficulties. Those children with the more severe forms of the condition (IP and PIE) can also see improvement of motor symptoms (such as walking) if they are given the appropriate treatments, but it often takes a longer period of time before results are seen. Improvement of motor function may not be seen in those individuals with the most severe symptoms. Aside from motor symptoms, the prognosis for individuals with IP and PIE can be poor depending on the severity of their non-motor symptoms, such as profound intellectual disability.

Treatment

The most effective treatment for individuals with tyrosine hydroxylase deficiency is levodopa in conjunction with carbidopa, which helps minimize side effects of treatment. Individuals with the mild form of tyrosine hydroxylase deficiency respond well to treatment with supplements of L-dopa and carbidopa. If taken before symptoms appear, the symptoms may be avoided completely. Even if symptoms have already begun, children with the disease often respond extremely well to the medication. If the disease has gone untreated for some time, certain symptoms may remain, including an irregular gait and other mild movement and speech difficulties.

Treatment with L-dopa and carbidopa supplements has been less beneficial for individuals with severe tyrosine hydroxylase deficiency, but this treatment may improve motor skills over time.

If symptoms have gone untreated, physical, occupational, and/or speech therapists may prove helpful.

Resources

Dystonia Medical Research Foundation
https://www.dystonia-foundation.org/
Pediatric Neurotransmitter Disease Association
http://www.pndassoc.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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