Biotinidase Deficiency

NxGen MDx

What Your Results Mean

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

Biotinidase Deficiency Explained

What is Biotinidase Deficiency?

Biotinidase deficiency is an inherited metabolic disorder in which the body cannot reuse biotin normally. Signs and symptoms for biotinidase deficiency vary in severity based on the type. If not properly treated, the signs and symptoms of profound biotinidase deficiency include developmental delays, vision and hearing loss, issues with movement and balance, skin rashes, muscle weakness, and alopecia. Affected individuals first experience signs and symptoms around the first few weeks or months of life.

Partial biotinidase deficiency is a milder form of the disease. In periods of stress or illness, individuals may experience symptoms such as skin rashes, alopecia, or muscle weakness. Some affected individuals with partial biotinidase deficiency may not experience any signs or symptoms at all.

Prognosis

Prognosis is generally favorable as long as biotinidase deficiency is managed. Early diagnosis and treatment with biotin supplements prevent most signs and symptoms and individuals can live a healthy life. When treated, life expectancy is not expected to be affected. Some signs and symptoms can also be reversible with treatment; however, vision loss, hearing loss, and intellectual disability are typically not reversible.

Treatment

Treatment involves lifelong management with biotin supplements, which can lessen and prevent many of the complications from biotinidase deficiency. If untreated, individuals can benefit from physical and occupational therapy. Vision and hearing aids can also be considered. Individuals with partial biotinidase deficiency often do not require any treatment.

Resources

Biotinidase Deficiency Family Support Group
http://biotinidasedeficiency.20m.com/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/biotinidase-deficiency/
National Society of Genetic Counselors
https://www.nsgc.org/

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