GRACILE Syndrome/ *BCS1L*-Related Disorders

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of GRACILE (Growth Restriction, Aminoaciduria, Cholestasis, Iron Overload, Lactic Acidosis, and Early Death) syndrome/*BCS1L*-related disorders. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of GRACILE syndrome/*BCS1L*-related disorders 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 or donor are carriers for GRACILE syndrome/BCS1L-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

GRACILE Syndrome/*BCS1L*-Related Disorders Explained

What is GRACILE Syndrome/*BCS1L*-Related Disorders?

GRACILE syndrome is an inherited condition that affects the ability of the body to convert food into energy. Affected infants must find energy through other means, and this causes toxic levels of lactic acid to accumulate in the blood during the first few days of life. GRACILE is an acronym for the symptoms of the disease: **G**rowth **R**estriction, **A**minoaciduria (abnormal presence of amino acids in the urine), **C**holestasis (impeded flow of bile from the liver), **I**ron Overload, **L**actic Acidosis (lactic acid buildup in the bloodstream), and **E**arly Death. Individuals with GRACILE syndrome usually do not survive beyond the first few months of life.

BCS1L-related disorders consist of Bjrnstad syndrome, mitochondrial complex III deficiency, and Leigh syndrome.

Björnstad syndrome is a rare disorder characterized by abnormal hair and hearing problems. Affected individuals have a condition known as pili torti, which means “twisted hair,” because the strands appear twisted when viewed under a microscope. Affected individuals also have hearing problems that become evident in early childhood. The hearing loss, which is caused by changes in the inner ear (sensorineural deafness), can range from mild to severe.

Mitochondrial complex III deficiency is a genetic condition that can affect several parts of the body, including the brain, kidneys, liver, heart, and the muscles used for movement (skeletal muscles). Signs and symptoms of mitochondrial complex III deficiency usually begin in infancy but can appear later.

Leigh syndrome is a severe neurological disorder that usually becomes apparent in the first year of life. This condition is characterized by progressive loss of mental and movement abilities (psychomotor regression) and typically results in death within two to three years, usually due to respiratory failure. A small number of individuals do not develop symptoms until adulthood or have symptoms that worsen more slowly.

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Prognosis

Prognosis for GRACILE syndrome is poor. Affected individuals usually do not survive past five months of life. Prognosis for Leigh syndrome and mitochondrial complex III deficiency is also typically poor, as affected individuals typically do not survive past early childhood. However, individuals with mitochondrial complex III deficiency with mild signs and symptoms can survive into adolescence or adulthood. Prognosis for Björnstad syndrome is considered good as it does not affect life expectancy; however, affected individuals will have some degree of hearing loss and hair loss beginning within the first two years of life.

Treatment

There is no cure for GRACILE syndrome/*BCS1L*-related disorders. Treatment is supportive and symptomatic. Affected individuals may require the skills of a team of specialists to systematically and comprehensively plan an affected child's treatment.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/gracile-syndrome
https://ghr.nlm.nih.gov/condition/bjornstad-syndrome
https://ghr.nlm.nih.gov/condition/mitochondrial-complex-iii-deficiency#resources
https://ghr.nlm.nih.gov/condition/leigh-syndrome

National Society of Genetic Counselors
https://www.nsgc.org/

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