Genetics Uncoded:

Facts about GRACILE Syndrome

What Your Test Results Mean

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

GRACILE Syndrome Explained

GRACILE syndrome is an inherited condition that affects the ability of the mitochondria to convert energy from food into energy that is useful for the body. The BCS1L protein is part of an enzyme complex called Complex III that is required for the conversion process, and the complex breaks down very quickly when there is a variant in the *BCS1L* gene, resulting in decreased cellular energy and organ damage. GRACILE is an acronym for the symptoms of the disease — affected individuals have fetal growth restriction, aminoaciduria (abnormal presence of amino acids in the urine), cholestasis (impeded flow of bile from the liver), iron overload, lactic acidosis (lactic acid buildup in the bloodstream), and early death. Individuals with GRACILE syndrome typically do not survive beyond the first year of life.

How the Genetics Work

GRACILE syndrome is an autosomal recessive disorder caused by pathogenic variants in the *BCS1L* gene. In general, individuals have two copies of the *BCS1L* gene. Carriers of GRACILE syndrome have a single variant in one copy of the *BCS1L* gene while individuals with GRACILE syndrome have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with GRACILE syndrome 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.

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