Genetics Uncoded:

Facts about

Limb-Girdle Muscular Dystrophy Type 2D

What Your Test Results Mean

Carriers typically show no symptoms of limb-girdle muscular dystrophy type 2D (LGMD2D); however, carriers are at an increased risk of having a child with LGMD2D. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Limb-Girdle Muscular Dystrophy Type 2D Explained

LGMD2D is an inherited condition that causes deterioration of the skeletal muscles, especially those around the hips and shoulders. Most of the time this disease is diagnosed in childhood, when the affected individual begins to have trouble with tasks like walking, climbing the stairs, and rising from a sitting position. However, mild cases may not be manifest until adulthood. LGMD2D is a progressive disease, and muscles will continue to waste, often leading the patient to require a wheelchair. There is no cure for this disorder, but physical therapy can help an individual retain their mobility for as long as possible. LGMD2D does not affect intelligence or mental function and rarely includes weakening of the heart muscle (cardiomyopathy).

How the Genetics Work

LGMD2D is an autosomal recessive muscle disorder caused by pathogenic variants in the *SGCA* gene. In general, individuals have two copies of the *SGCA* gene. Carriers of LGMD2D have a single variant in one copy of the *SGCA* gene while individuals with LGMD2D have variants in both copies of the gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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