

Limb-Girdle Muscular Dystrophy Type 2A (Calpainopathy)

What Your Results Mean

Test results indicate that you are a carrier of limb-girdle muscular dystrophy type 2A (LGMD2A). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of LGMD2A 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 limb-girdle muscular dystrophy type 2A, each of your children has a 1 in 4 (25%) chance to have the condition.

Limb-Girdle Muscular Dystrophy Type 2A Explained

What is Limb-Girdle Muscular Dystrophy Type 2A?

LGMD2A is an inherited condition that causes deterioration of the skeletal muscles, especially those around the hips, shoulders, upper arms, pelvic area, and thighs. 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. LGMD2A is a progressive disease, and muscles will continue to waste, often leading the affected individual to require a wheelchair. LGMD2A does not affect intelligence or mental function and rarely includes weakening of the heart muscle (cardiomyopathy).



Prognosis

Prognosis is considered mild, as it does not impact life expectancy. However, most affected individuals will require a wheelchair for mobility.

Treatment

There is no cure for this disorder, but physical therapy can help an individual retain their mobility for as long as possible.



Resources

LGMD-Info

<http://lgmd-info.org/>

Muscular Dystrophy Association

<https://www.mda.org/disease/limb-girdle-muscular-dystrophy>

National Society of Genetic Counselors

<https://www.nsgc.org/>