

Limb-Girdle Muscular Dystrophy Type 2E

What Your Results Mean

Test results indicate that you are a carrier of limb-girdle muscular dystrophy type 2E (LGMD2E). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of LGMD2E 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 2E, each of your children has a 1 in 4 (25%) chance to have the condition.

Limb-Girdle Muscular Dystrophy Type 2E Explained

What is Limb-Girdle Muscular Dystrophy Type 2E?

LGMD2E 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. LGMD2E is a progressive disease, and muscles will continue to waste, often leading the patient to require a wheelchair. In addition, patients may experience a weakening of the heart muscle, which may shorten life expectancy. LGMD2E does not affect intelligence or mental function.



Prognosis

Prognosis is dependent on whether or not the heart is impacted. Most affected individuals will be wheelchair bound for mobility. Those with cardiac involvement may have a shortened lifespan.

Treatment

There is no cure for this disorder, and treatment is based on managing symptoms. 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/>