Limb-girdle muscular dystrophy(LGMD)

Introduction

- Limb-girdle muscular dystrophy refers to a large group of muscle disorders that causes weakness of the muscles in the limbs (legs and arms) and girdle area (hips, pelvis, abdomen) Like other muscular dystrophies, limb-girdle can affect other body systems, such as the heart in some instances.
- As the disease progresses, limb-girdle muscular dystrophy can also cause weakness of the breathing and swallowing muscles.

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Clinical manifestation /Symptoms

- Toe walking, or walking on the balls of the feet
- Walking with a waddling gait
- Inability to rise from a squatting position without using the arms for leverage
- Difficulty running
- Abnormal curvature of the spine, such as lordosis and scoliosis. Learn more about scoliosis.
- Joint stiffness

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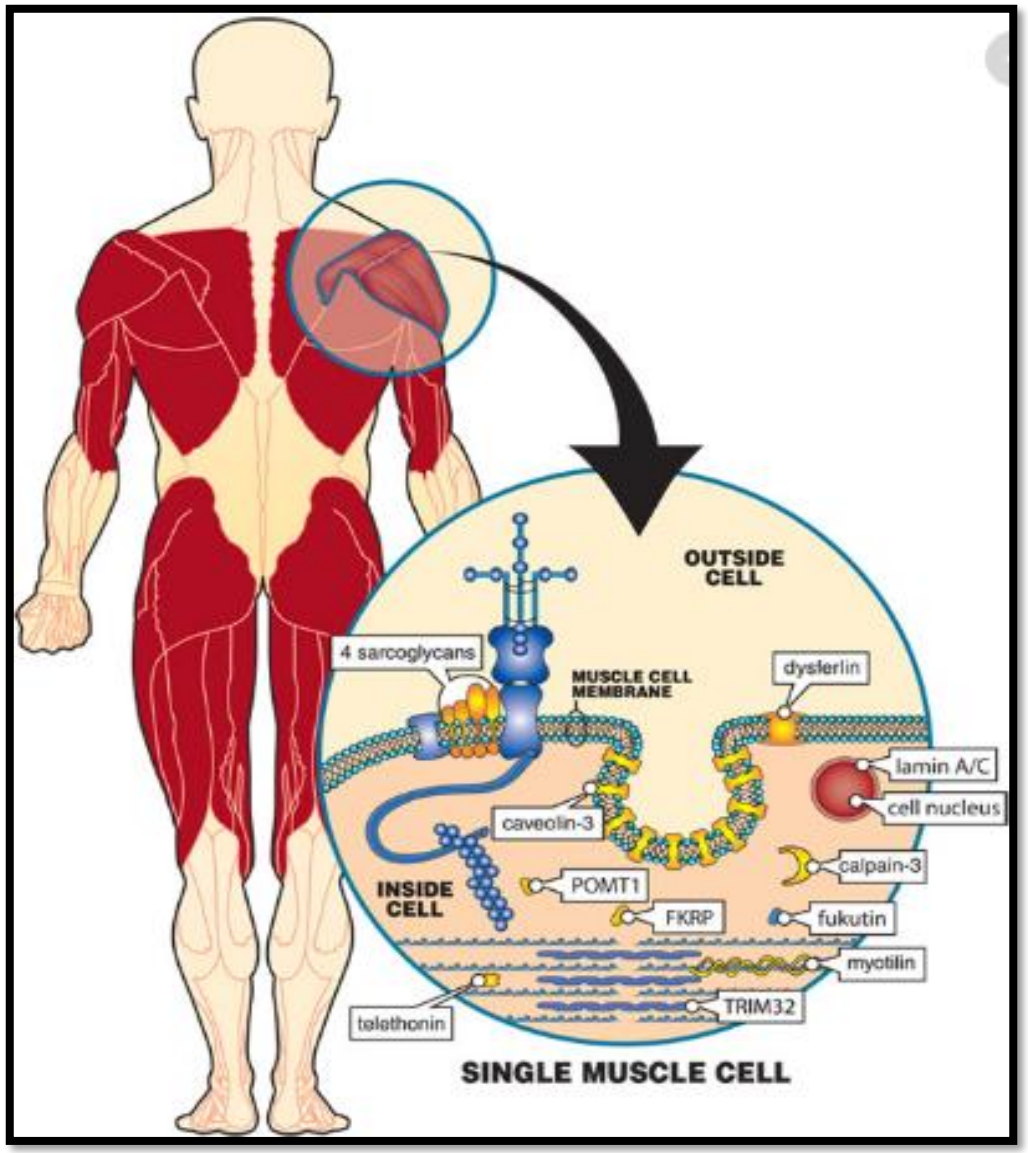
Clinical manifestation /Symptoms



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Causes

The various forms of limb-girdle muscular dystrophy are caused by mutations in many different genes. These genes provide instructions for making proteins that are involved in muscle maintenance and repair.

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Inheritance Pattern

❖Limb-girdle muscular dystrophy can have different inheritance patterns. Most forms of this condition are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

❖Several rare forms of limb-girdle muscular dystrophy are inherited in an autosomal dominant pattern

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Gene Panel

ANO5, CAPN3, CAV3, DES, DNAJB6, DYSF, FKRP, FLNC, HNRNPDL, ISPD, LMNA, MYOT, PNPLA2, SGCA, SGCB, SGCD, SGCG, TCAP, TRIM32, TTN

No of Genes : 20
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Diagnostic Tests

anderson
Clinical Genetics

Management

Reference

https://ghr.nlm.nih.gov/condition/limb-girdle-muscular-dystrophy#genes

https://www.mda.org/disease/limb-girdle-muscular-dystrophy/causes-inheritance

https://stanfordhealthcare.org/medical-conditions/brain-and-nerves/limb-girdle-muscular-dystrophy/causes.html