Congenital muscular dystrophy (CMD)

Introduction

- A congenital muscular dystrophy (CMD) is a muscle disorder that is present at birth.
- The word "congenital" means present at birth. Muscular dystrophy is a term used to describe many different types of skeletal muscle disorders that cause muscle weakness and wasting (loss of muscle mass).
- Congenital muscular dystrophies are inherited disorders that are passed from parent to child through a faulty gene. Many different gene abnormalities can cause a baby to be born with a congenital muscular dystrophy.

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Clinical manifestation /Symptoms

- Babies that appear to be floppy when held
- Low level of spontaneous movements like kicking the legs or waving the arms
- Difficulty meeting developmental milestones like raising the head or rolling over
- Learning disabilities as your child ages.

Common features include hypotonia; progressive muscle weakness and degeneration (atrophy); joint contractures; and delayed motor milestones

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Causes

CMD is a genetic disorder. It occurs when a faulty gene results in abnormal muscle function.

❖The gene can be passed from parent to child, even if the parent does not have symptoms. Or, the child can be the first in their family to have muscular dystrophy.

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Inheritance Pattern

❖Congenital muscular dystrophies (CMD) are a heterogeneous group of autosomal recessively inherited diseases, presenting at birth or within the first 6 months of life.

Most forms are inherited in an autosomal recessive manner.

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Gene Panel

FKTN, LARGE1, POMGNT1, POMT1, POMT2

No of Genes : 5
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Diagnostic Tests

anderson
Clinical Genetics

Management

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Reference

https://www.dmd.nl/CMD.html

https://rarediseases.info.nih.gov/diseases/9138/congenital-muscular-dystrophy

https://stanfordhealthcare.org/medical-conditions/bones-joints-and-muscles/congenital-muscular-dystrophy/causes.html