Syndrome/Diseases Name

- Introduction
- Clinical manifestation /Symptoms
- Causes
- Gene Panel
- Inheritance Pattern
- Diagnostics
- Mangement
- Future Options
- Reference

Duchenne muscular dystrophy (DMD)

Introduction

- Duchenne muscular dystrophy is an inherited disorder. It involves muscle weakness, which quickly gets worse.
- DMD causes progressive weakness and loss (atrophy) of skeletal and heart muscles.
- DMD is a progressive form of muscular dystrophy that occurs primarily in males, though in rare cases may affect females.
- Many children with DMD develop normally during infancy and early childhood.

Duchenne muscular dystrophy

Clinical manifestation /Symptoms

- difficulty walking
- a loss of ability to walk
- enlarged calves
- learning disabilities, which occurs in about one-third of affected individuals
- a lack of motor skills development
- fatigue
- rapidly worsening weakness in the legs, pelvis, arms, and neck

Duchenne muscular dystrophy

Causes

❖Duchenne muscular dystrophy (DMD) is caused by mutations in the DMD gene. The DMD gene provides instructions for making a protein called dystrophin.

❖Dystophin is primarily made in the muscle cells of the heart and skeletal muscle.

❖The main job of dystrophin in muscle cells is to help stabilize and protect muscle fibers.

Duchenne muscular dystrophy

Inheritance Pattern

❖DMD gene is located on the X chromosome, one of the two types of sex chromosomes.

❖Males have an X and a Y chromosome; whereas females have two X chromosomes. Since males only have one X chromosome, they also only have one copy of the DMD gene. If this copy has a genetic change that causes DMD, the male will have DMD. Males get their X chromosome from their mother and the Y chromosome from their father

Fragile X syndrome (FXS)

Gene Panel

DMD

No of Genes : 1
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Diagnostic Tests

anderson
Clinical Genetics

Management

Duchenne muscular dystrophy

Reference

https://rarediseases.info.nih.gov/diseases/6291/duchenne-muscular-dystrophy

https://ghr.nlm.nih.gov/gene/DMD

https://www.healthline.com/health/duchenne-muscular-dystrophy#treatment