

Introduction

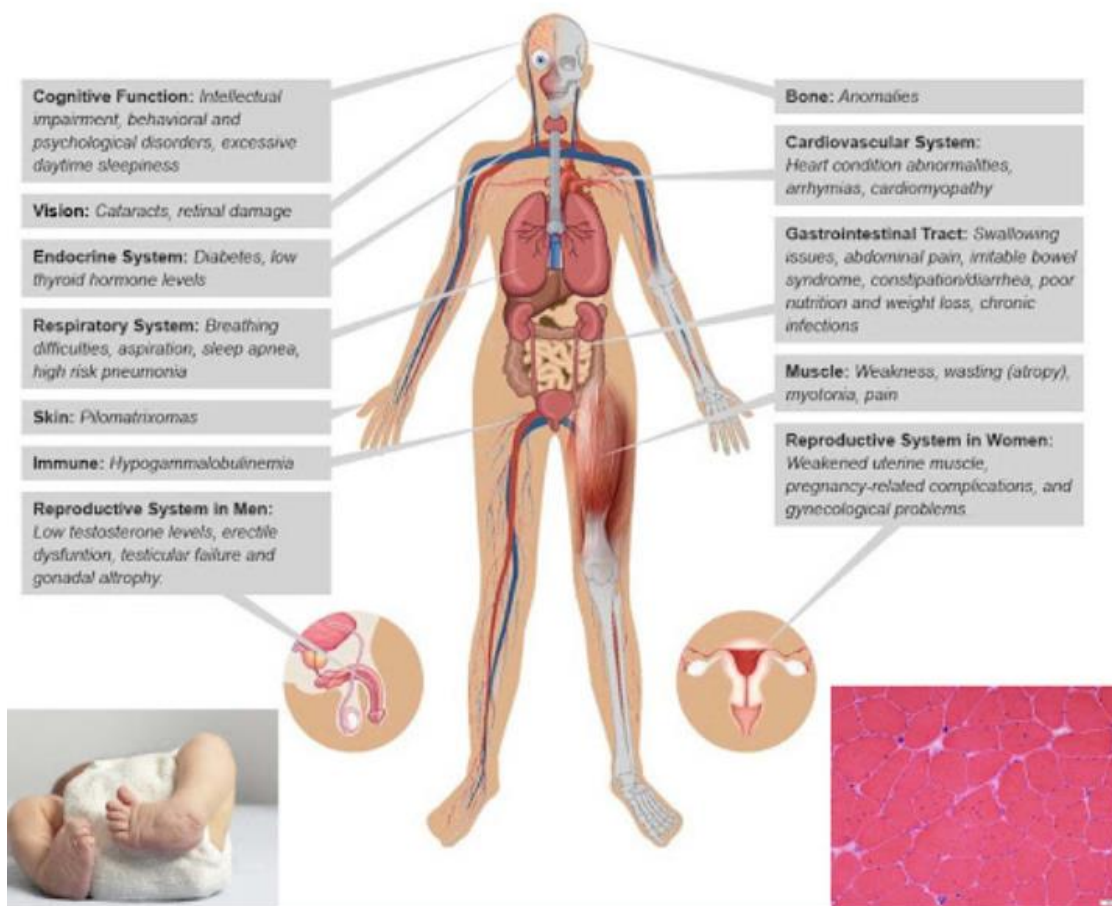
- Myotonic dystrophy is part of a group of inherited disorders called muscular dystrophies. It is the most common form of muscular dystrophy that begins in adulthood.
- The muscular dystrophies are characterized by weakness and degeneration of various voluntary muscles of the body
- There are two major types of myotonic dystrophy: type 1 and type 2. Their signs and symptoms overlap, although type 2 tends to be milder than type 1.

Clinical manifestation /Symptoms

- Myotonic dystrophy is characterized by progressive muscle wasting and weakness
- Difficulty releasing their grip on a doorknob or handle
- slurred speech or temporary locking of their jaw
- The muscle weakness associated with type 1 particularly affects the lower legs, hands, neck, and face.
- Muscle weakness in type 2 primarily involves the muscles of the neck, shoulders, elbows, and hips

Clinical manifestation /Symptoms

Myotonic dystrophy type 1



Normal biceps



Muscular dystrophy



Causes

- ❖ The two types of myotonic dystrophy are caused by mutations in different genes.
- ❖ Myotonic dystrophy type 1 is caused by mutations in the *DMPK* gene, while type 2 results from mutations in the *CNBP* gene.
- ❖ The protein produced from the *DMPK* gene may play a role in communication within cells. It appears to be important for the correct functioning of cells in the heart, brain, and skeletal muscles (which are used for movement).
- ❖ The protein produced from the *CNBP* gene is found primarily in the heart and in skeletal muscles, where it probably helps regulate the function of other genes.

Inheritance Pattern

- ❖ Both types of myotonic dystrophy are inherited in an autosomal dominant pattern

Gene Panel

DMPK, CNBP

No of Genes :2

Sample Type : EDTA-blood sample - 4 ml

TAT : 6 Weeks

Methodology : NGS

Diagnostic Tests

Management

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>