

Duchenne muscular dystrophy

Definition

Duchenne muscular dystrophy (DMD) is an inherited disorder that is characterized by rapid progression of muscle degeneration

Duchenne muscular dystrophy is the most prevalent of muscular dystrophies

Duchenne muscular dystrophy affects one in 2500 males.

It is a severe recessive x-linked form of muscular dystrophy. In general, males are only afflicted, though females can be carriers.

Causes

Duchenne muscular dystrophy is a rapidly-worsening form of muscular dystrophy. It is caused by a defective gene for dystrophin (a protein in the muscles). However, it often occurs in people without a known family history of the condition.

Signs

Duchenne muscular dystrophy is usually diagnosed when a child is 3 to 6 years of age.

Early signs include delay in walking, frequent falling, and difficulty getting up from a sitting or lying position.

Muscle deterioration continues to progress and, around the time they are 12 years of age, children with DMD become unable to walk.

The disease is usually fatal in the teens or early 20s, most commonly due to severe respiratory or heart problems, or both.

Diagnosis

A clinical diagnosis may be made and is confirmed with other tests: electromyogram analysis and muscle biopsy.

Treatment

There is no known cure for Duchenne muscular dystrophy. Treatment is aimed at control of symptoms to maximize the quality of life. It includes :

- Medical care (neurology, cardiology, respiratory and orthopedic)
- Physical therapy and occupational therapy
- Adapted positioning
- Orthopedic appliances
- Psychology care
- Orthopedic surgery

Expectations (prognosis):

Duchenne muscular dystrophy results in rapidly progressive disability.

The condition is severe enough to shorten life expectancy.