Duchenne muscular dystrophy (DMD)

Duchenne muscular dystrophy (DMD) is an X-linked rare disease. DMD occurs as a result of mutations in the dystrophin gene. It leads to an absence of or defects in the protein dystrophin and is manifested by progressive muscle degradation.

Typically the disorder is diagnosed only around 4-5 years of age. Duchenne patients are often wheelchair bound between the ages of seven and 13 years old. Eventually, increasing difficulty in breathing due to respiratory muscle dysfunction requires ventilation support, and cardiac dysfunction can lead to heart failure. The condition is universally fatal, and without intervention the mean age at death is around 19 years.

Although DMD is an X-linked recessive disease and females are typically asymptomatic carriers of mutations, some female carriers may manifest symptoms varying from mild muscle weakness to a more severe clinical course and are classified as manifesting or symptomatic carriers. The manifesting phenotype is based on inactivation of the normal X chromosome or on translocation between X chromosome and an autosome. Female carriers having elevated CK-MM concentrations due to muscle degeneration at birth may result as screen positive (depending on the cut-off used) in newborn screening for DMD. The incidence of DMD is 1 in 3000-6000 live male births worldwide.

Treatment

Although no cure exists for the progressive muscle weakness of DMD, corticosteroid treatment improves muscle strength and function, and in combination with supportive medical care is associated with delayed loss of ambulation and markedly improved survival. In addition, novel molecular therapies are under development and therefore interest towards newborn screening is increasing.