Muscular Dystrophy

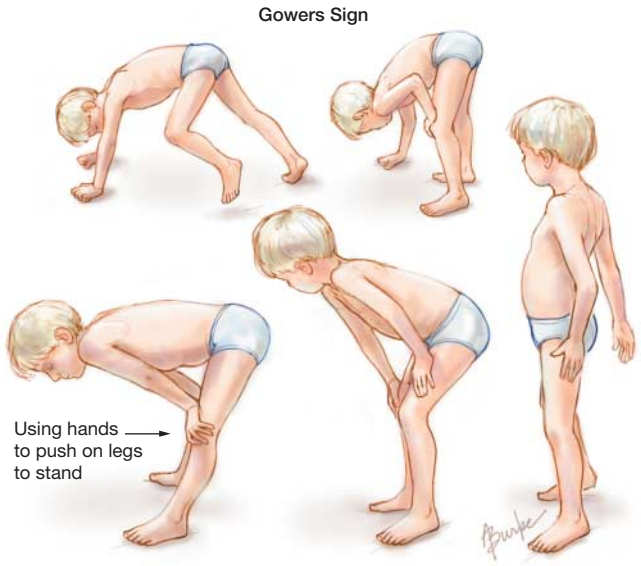
Muscular dystrophies are a group of diseases that are caused by a **genetic mutation** (a change in genetic material that can be passed on to children), affect muscles, and are **progressive** (increase in severity over time). The most common types are

- **Duchenne muscular dystrophy**, the most common childhood form of muscular dystrophy. It is an **X-linked disease**, which means that the gene is passed from mothers to their sons.
- **Becker muscular dystrophy**, with a genetic defect very similar to that in Duchenne muscular dystrophy, but not as severe. Together, Duchenne and Becker muscular dystrophies affect 1 to 2 in 10 000 males between the ages of 5 and 24 years.
- **Myotonic dystrophy**. Patients have **myotonia** (delayed relaxation of muscles after contraction), for example, after grasping a doorknob. There are several different types within this group. They occur in both men and women and affect about 1 in 8000 people. The December 14, 2011, issue of *JAMA* includes an article about myotonic muscular dystrophy.

CLINICAL FEATURES

The physical examination and personal history for each type of muscular dystrophy is different and contributes to the diagnosis.

- In Duchenne muscular dystrophy, young boys start walking very late and have difficulties arising from the floor (**Gowers sign**, using their hands to push on their legs to get up). Intellectual impairment may also be present.
- In Becker muscular dystrophy, weakness may be limited to the **quadriceps** (muscles in the front of the thigh).
- Infants with **type 1** myotonic dystrophy can have severe trouble breathing and swallowing and may not survive the **neonatal** (newborn) period. Adults with type 1 myotonic dystrophy may develop cataracts, hand and lower leg weakness and **wasting** (loss of muscle mass), and grip myotonia. Adults with later-onset **type 2** myotonic dystrophy can have stiffness and myotonia in the thighs and hands, neck weakness, and hip weakness with trouble climbing stairs.



DIAGNOSIS

- The blood level of **creatine kinase** (a protein released with muscle breakdown) is abnormally high early in Duchenne muscular dystrophy. As the disease progresses and muscle mass decreases, the level of creatine kinase decreases.
- Blood tests look for the genetic mutations that cause each type of muscular dystrophy. These allow a definitive diagnosis.

PROGNOSIS

- Muscular dystrophies are progressive diseases. Children with Duchenne muscular dystrophy often require a wheelchair by their teenage years, as do children with Becker muscular dystrophy in their late adolescence.
- Because muscular dystrophies affect muscles everywhere in the body, including the lungs and heart, patients may need specialized care, such as a pacemaker. Each of the forms has unique features that require attention. For example, myotonic dystrophy can lead to impotence and high cholesterol levels.
- Corticosteroids may improve muscle strength and function in Duchenne dystrophy. In patients with myotonic dystrophy, other medications improve myotonia. However, muscular dystrophies cannot be cured and weakness is progressive. Many people with either Duchenne or Becker muscular dystrophy may die in their early to late 20s.

Sources: Mayo Clinic, Genetics Home Reference, Centers for Disease Control and Prevention

FOR MORE INFORMATION

- Mayo Clinic
  www.mayoclinic.com/health/muscular-dystrophy/DS00200
- Genetics Home Reference
  ghr.nlm.nih.gov/condition/myotonic-dystrophy
  ghr.nlm.nih.gov/condition/duchenne-and-becker-muscular-dystrophy

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