Muscular dystrophy

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Muscular dystrophy

- Muscular dystrophy refers a group of disorders that involve a progressive loss of muscle mass and consequent loss of strength.
- The main forms of muscular dystrophy may affect up to 1 in every 5,000 males.
- The most common form is Duchenne muscular dystrophy. It typically affects young boys, but other variations can strike in adulthood.
- Muscular dystrophy is caused by genetic mutations that interfere with the production of muscle proteins that are needed to build and maintain healthy muscles.
- The causes are genetic. A family history of muscular dystrophy will increase the chance of it affecting an individual.
- There is currently no cure, but certain physical and medical treatments can improve symptoms and slow the progression.

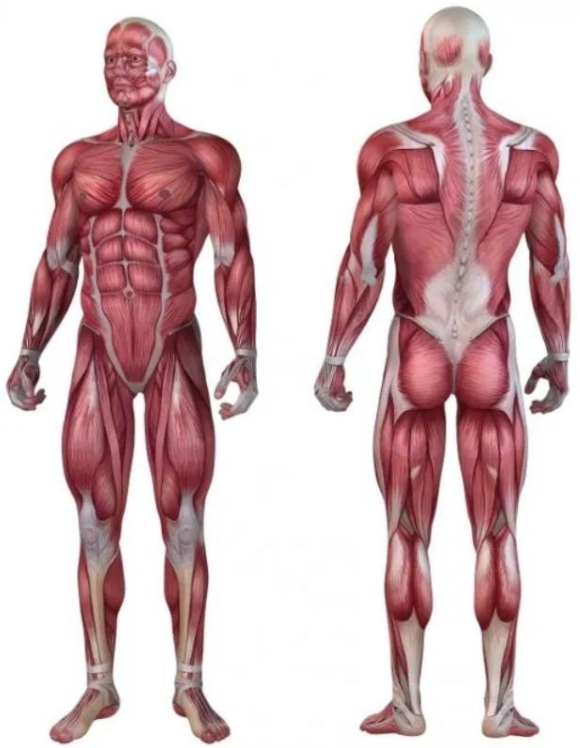
Some facts on muscular dystrophy

- Here are some key points about muscular dystrophy. More detail and supporting information is in the main article.
- Muscular dystrophy is a collection of muscle-wasting conditions
- Duchenne muscular dystrophy is the most common type
- A lack of a protein called dystrophin is the main cause of muscular dystrophy
- Gene therapies are currently being trialed to combat the disease
- There is currently no cure for muscular dystrophy

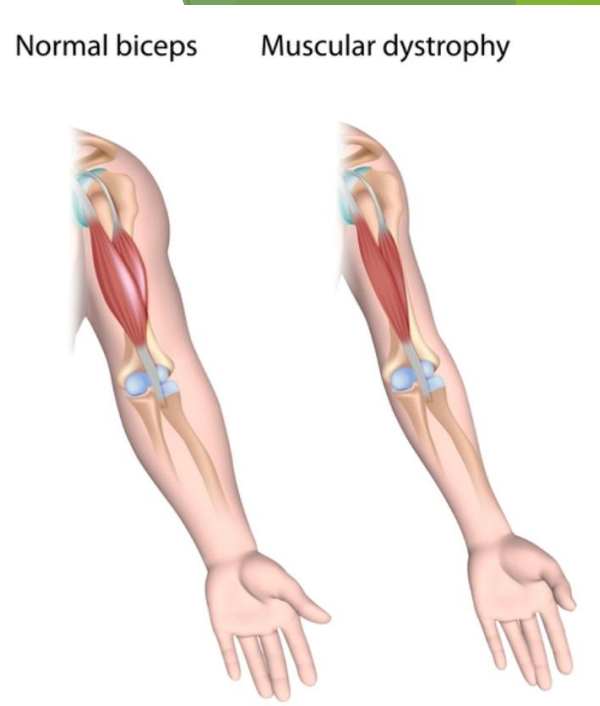
Symptoms

Below are the symptoms of muscular dystrophy, the most common form of the disease.

Early symptoms include:
•a waddling gait
•pain and stiffness in the muscles
•difficulty with running and jumping
•walking on toes
•difficulty sitting up or standing
•learning disabilities, such as developing speech later than usual



Sypmtoms continued



- As time goes on, the following become more likely:
- inability to walk
- a shortening of muscles and tendons, further limiting movement
- breathing problems can become so severe that assisted breathing is necessary
- curvature of the spine can be caused if muscles are not strong enough to support its structure
- the muscles of the heart can be weakened, leading to cardiac problems

Treatments and drugs for treatment

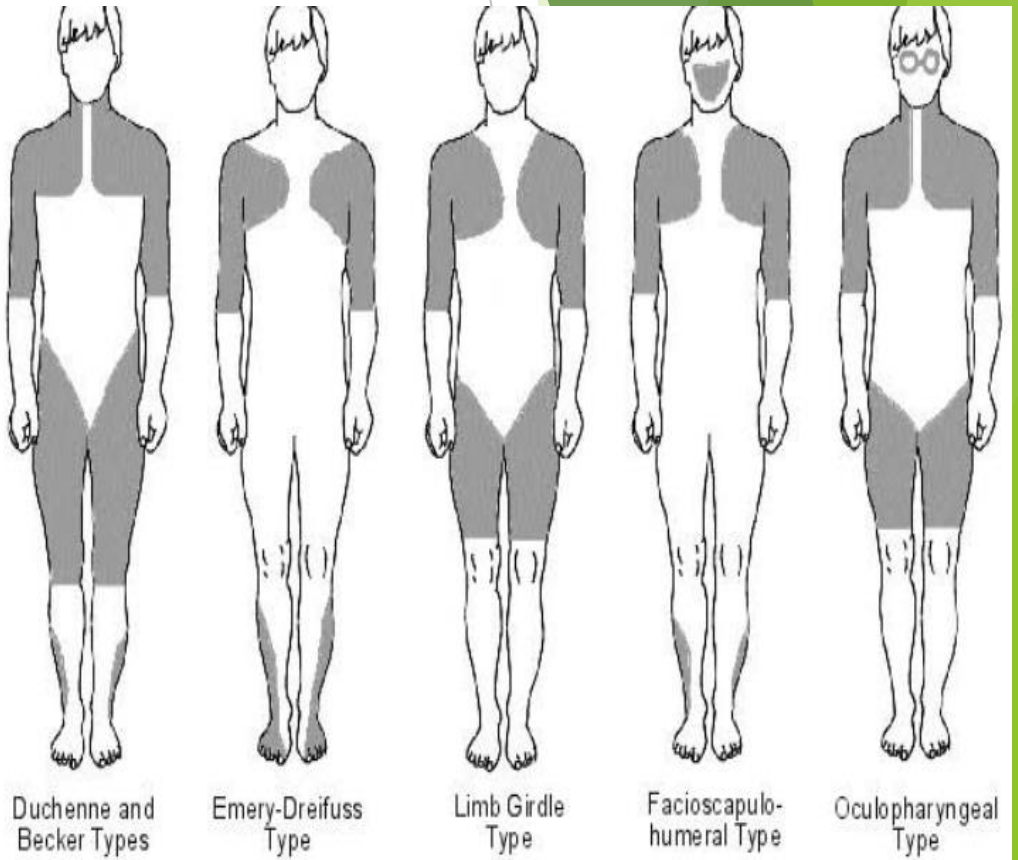
- **Treatment**
- Currently, there is no cure for muscular dystrophy. Medications and various therapies help slow the progression of the disease and keep the patient mobile for the longest possible time.
- **Drugs**
- The two most commonly prescribed drugs for muscular dystrophy are:
- **Corticosteroids:** This type of medication can help increase muscle strength and slow progression, but long-term use can weaken bones and increase weight gain.
- **Heart medications:** If the condition impacts the heart, beta blockers and angiotensin-converting enzyme (ACE) inhibitors may help.

Treatments continued…

- **Physical therapy**
- **General exercises:** A range of motion and stretching exercises can help combat the inevitable inward movement of the limbs as muscles and tendons shorten.
- **Breathing assistance:** As the muscles used for breathing become weaker, it may be necessary to use devices to help improve oxygen delivery through the night.
- **Mobility aids:** Canes, wheelchairs, and walkers can help the person stay mobile.
- **Braces:** These keep muscles and tendons stretched and help slow their shortening. They also give added support to the user when moving.

Types of Muscular dystrophy

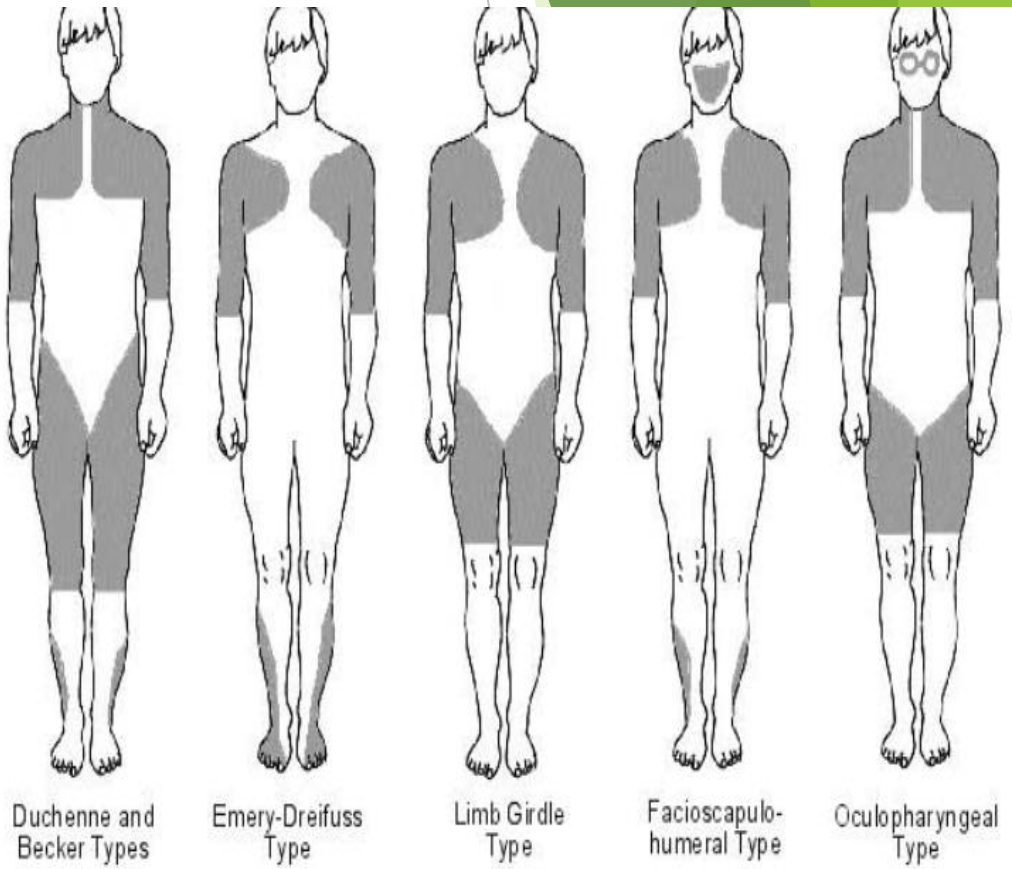
- **Types**
- There are different types of muscular dystrophy, including the following:
- **Duchenne muscular dystrophy:** Symptoms normally start before a child's third birthday; they are generally wheelchair-bound by 12 years and die of respiratory failure by their early-to-mid-twenties.
- **Becker muscular dystrophy:** Similar symptoms to Duchenne but with a later onset and slower progression
- **Myotonic (Steinert's disease):** The myotonic form is the most common adult-onset form. It is characterized by an inability to relax a muscle once it has contracted.



Main areas of muscle weakness in different types of dystrophy

Types continued…

- **Congenital:** This type can be obvious from birth or before the age of 2 years. Some forms progress slowly whereas others can move swiftly and cause significant impairment.
- **Facioscapulohumeral (FSHD):** Onset can be at almost any age but is most commonly seen during teenage years. The muscular weakness often begins in the face and shoulders.
- **Limb-girdle:** Individuals with the limb-girdle muscular dystrophy might have trouble raising the front part of the foot, making tripping a common problem.
- **Oculopharyngeal muscular dystrophy:** Onset is between the ages of 40 and 70 years. Eyelids, throat, and face are first affected, followed by the shoulder and pelvis



Main areas of muscle weakness in different types of dystrophy

Causes of muscular dystrophy

- Muscular dystrophy is caused by mutations on the X chromosome. Each version of muscular dystrophy is due to a different set of mutations, but all prevent the body from producing dystrophin. Dystrophin is a protein essential for building and repairing muscles.
- Duchenne muscular dystrophy is caused by specific mutations in the gene that encodes the cytoskeletal proteindystrophin. Dystrophin makes up just 0.002 percent of the total proteins in striated muscle, but it is an essential molecule for the general functioning of muscles.

Diagnosis

- There are a variety of techniques used to definitively diagnose muscular dystrophy:
- Share on PinterestThe genetic mutations involved in muscular dystrophy are well known and can be used to make a diagnosis.
- **Enzyme assay:** Damaged muscles produce creatine kinase (CK). Elevated levels of CK in the absence of other types of muscle damage could suggest muscular dystrophy.
- **Genetic testing:** As genetic mutations are known to occur in muscular dystrophy, these changes can be screened for.
- **Heart monitoring:**Electrocardiography and echocardiograms can detect changes in the musculature of the heart. This is especially useful for the diagnosis of myotonic muscular dystrophy.
- **Lung monitoring:** Checking lung function can give additional evidence.
- **Electromyography:** A needle is placed into the muscle to measure the electrical activity. The results can show signs of muscle disease.
- **Biopsy:** Removing a portion of muscle and examining it under a microscope can show the tell-tale signs of muscular dystrophy.

Outlook

- The outlook will depend on the type of muscular dystrophy and how severe the symptoms are.
- Duchenne muscular dystrophy can lead to life-threatening complications, such as breathing difficulties and heart problems.
- In the past, people with this condition did not usually survive beyond their 20s, but progress is improving the outlook.
- Currently, the average life expectancy for people with Duchenne is 27 years, and it may improve in time, as treatment progresses.
- A person with muscular dystrophy is likely to need lifelong assistance.