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## Duchenne muscular dystrophy

Duchenne muscular dystrophy is an inherited disorder. It involves muscle weakness, which quickly gets worse.

### Causes

Duchenne muscular dystrophy is a form of muscular dystrophy. It worsens quickly. Other muscular dystrophies (including Becker muscular dystrophy) get worse much more slowly.

Duchenne muscular dystrophy 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.

The condition most often affects boys due to the way the disease is inherited. The sons of women who are carriers of the disease (women with a defective gene, but no symptoms themselves) each have a 50% chance of having the disease. The daughters each have a 50% chance of being carriers. Very rarely, a female can be affected by the disease.

Duchenne muscular dystrophy occurs in about 1 out of every 3,600 male infants. Because this is an inherited disorder, risks include a family history of Duchenne muscular dystrophy.

### Symptoms

Symptoms most often appear before age 6. They may start as early as infancy. Most boys show no symptoms in the first few years of life.

Symptoms may include:

- Fatigue
- Learning difficulties (the IQ can be below 75).
- Intellectual disability (possible, but does not get worse over time).

Muscle weakness:

- Begins in the legs and pelvis, but also occurs less severely in the arms, neck, and other areas of the body.
- Problems with motor skills (running, hopping, jumping).
- Frequent falls.
- Trouble getting up from a lying position or climbing stairs.

- Shortness of breath, fatigue, and swelling of the feet due to a weakening of the heart muscle.
- Problem breathing due to a weakening of the respiratory muscles.
- Gradual worsening of muscle weakness.

Progressive difficulty walking:

- Ability to walk may be lost by age 12, and the child will have to use a wheelchair.
- Breathing difficulties and heart disease usually start by age 20.

## **Exams and Tests**

A complete nervous system (neurological), heart, lung, and muscle exam may show:

- Abnormal, weak heart muscle (cardiomyopathy) that becomes evident by age 10
- Congestive heart failure or irregular heart rhythm (arrhythmias) that is present in all people with Duchenne muscular dystrophy by the age of 18
- Deformities of the chest and back (scoliosis)
- Enlarged muscles of the calves, buttocks, and shoulders (around age 4 or 5). These muscles are eventually replaced by fat and connective tissue (pseudohypertrophy).
- Loss of muscle mass (wasting)
- Muscle contractures in the heels, legs
- Muscle deformities
- Respiratory disorders, including pneumonia and swallowing with food or fluid passing into the lungs (in late stages of the disease)

Tests may include:

- Blood creatine kinase levels
- Electrocardiogram (ECG)
- Cardiac testing
- X-rays of the spine to determine if there is a scoliosis
- Pulmonary function tests
- Electromyography (EMG)
- Genetic tests
- Muscle biopsy

## **Treatment**

There is no known cure for Duchenne muscular dystrophy. Treatment aims to control symptoms to improve quality of life.

Steroid drugs can slow the loss of muscle strength. They may be started when the child is diagnosed or when muscle strength begins to decline.

Other treatments may include:

- Albuterol (to help with muscle strength)
- Amino acids
- Carnitine
- Coenzyme Q10
- Creatine
- Fish oil
- Green tea extracts
- Vitamin E

However, the effects of these treatments have not been proven. Stem cells and gene therapy may be used in the future.

Genetic therapies may be helpful to certain patients, depending on the genetic cause. Each therapy only works on a small number of patients, depending on their specific genetic change (mutation). While these treatments have been shown to increase dystrophin production, they have not yet been shown to have significant clinical benefits. Treatments include:

- Eteplirsen
- Golodirsen
- Vitolarsen
- Casimersen
- Ataluren
- Delandistrogene moxeparvovec

The use of steroids and the lack of physical activity can lead to excessive weight gain. Activity is encouraged. Inactivity (such as bedrest) can make the muscle disease worse. Physical therapy may help to maintain muscle strength and function. Speech therapy is often needed.

Other treatments may include:

- Assisted ventilation (used during the day or night)
- Drugs to help heart function, such as angiotensin-converting-enzyme inhibitors, beta-blockers, and diuretics
- Orthopedic appliances (such as braces and wheelchairs) to improve mobility
- Spine surgery to treat progressive scoliosis for some people
- Proton pump inhibitors (for people with gastroesophageal reflux)

Several new treatments are being studied in trials.

## Support Groups

You can ease the stress of illness by joining a support group where members share common experiences and problems. The Muscular Dystrophy Association is an excellent source of information on this disease.

# **Outlook (Prognosis)**

Duchenne muscular dystrophy leads to progressively worsening disability. Death often occurs from lung disorders, although advances in supportive care have resulted in many men living to age 35 years.

## **Possible Complications**

Complications may include:

- Cardiomyopathy
- Congestive heart failure (rare)
- Deformities
- Heart arrhythmias (rare)
- Mental impairment (varies, usually minimal)
- Permanent, progressive disability, including decreased mobility and decreased ability to care for self
- Pneumonia or other respiratory infections
- Respiratory failure

## **When to Contact a Medical Professional**

Contact your health care provider if:

- Your child has symptoms of Duchenne muscular dystrophy
- Symptoms get worse, or new symptoms develop, particularly fever with cough or breathing problems

## **Prevention**

People with a family history of the disease may want to seek genetic counseling. Genetic studies done during pregnancy are very accurate in detecting Duchenne muscular dystrophy.

## **Alternative Names**

Pseudohypertrophic muscular dystrophy; Muscular dystrophy - Duchenne type

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