



## Types of muscle disease



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### Duchenne muscular dystrophy

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy is a neuromuscular condition caused by the lack of a protein called dystrophin. About 100 boys with Duchenne muscular dystrophy are born in the United Kingdom each year. There are about 2500 known boys with the disorder living in the UK at any one time. For the general population the risk of having a child with Duchenne muscular dystrophy is about 1 in every 3500 male births.

Duchenne muscular dystrophy is a serious condition that causes progressive muscle weakness. In Duchenne muscular dystrophy muscle fibres break down and are replaced by fibrous and or fatty tissue causing the muscle to gradually weaken.

What are the causes of Duchenne muscular dystrophy?

Duchenne muscular dystrophy is a genetic disease – it is caused by a mistake in the genetic code, which is often called a mutation. In Duchenne muscular dystrophy the mutation occurs in a gene called dystrophin which is located on the X-chromosome (girls have two X chromosomes and boys only one). In some cases, the condition is inherited from the mother who is a carrier but it can also be caused by a new mutation in the child's genes.

In just over half of all cases the mother carries the gene mutation but is usually not herself affected by it. Such women are known as 'carriers'. Each subsequent son of a carrier has a 50:50 chance of being affected and each daughter has a 50:50 chance of being a carrier herself. A small number of female carriers of the gene have a mild degree of muscle weakness themselves and are then known as **manifesting carriers**.

How is Duchenne muscular dystrophy diagnosed?

Most boys with Duchenne muscular dystrophy are not diagnosed until they start displaying symptoms unless there is a family history of the condition. The first signs of Duchenne muscular dystrophy are usually problems with muscle function for example difficulty running and jumping, enlarged calf muscles and/or delayed speech development. If a blood test is

done high levels a protein called creatine kinase are seen. Creatine kinase is normally found in muscle but when muscles are damaged, it leaks into the bloodstream.

Duchenne muscular dystrophy has to be confirmed by genetic testing usually on a blood sample. Different types of **genetic tests** are able to provide specific and more detailed information about the DNA mutation. Genetic confirmation is crucial as it will assist in determining if your son qualifies for a number of clinical trials and will assist the family with decisions related to prenatal diagnosis and future pregnancies.

Your doctor may also recommend a **muscle biopsy** which is the process of taking a small sample of muscle for analysis. Tests on the muscle biopsy can provide information on the amount of dystrophin protein present in the muscle cells. These tests can help distinguish Duchenne muscular dystrophy from a milder form of the condition known as Becker muscular dystrophy.

Is there a treatment or cure?

Unfortunately no cure has yet been discovered. Despite promising research into the condition, at the present time, the only treatment that experts recommend to slow the decline in muscle strength and mobility is **steroids**. However, there are many possible side-effects which must be carefully managed.

A multidisciplinary approach, with the input of specialists in many different areas including physiotherapists and occupational therapists, is the best way to manage Duchenne muscular dystrophy. Regular checkups with a specialist doctor are important in order to make decisions about new treatments at the most appropriate time and to foresee and prevent problems if possible. It is recommended that your doctor is visited every six months and the specialist physiotherapist and/or occupational therapist about every four months if possible.

What is the prognosis?

In the early stages, when boys are still able to walk, they will be showing several signs associated with Duchenne muscular dystrophy, a Gower's manoeuvre (they need to support themselves with hands on thighs as they get up from the floor), waddling type walking and walking on their toes. They are still able to climb stairs, but typically bring the second foot up to join the first rather than going foot over foot. Later, when walking becomes increasingly difficult, there are more problems with climbing stairs and getting up from the floor.

By about 8 to 11 years (sometimes earlier or a little later), a wheelchair will be required. At first, boys may be able to propel themselves and his posture is typically good. Later, upper limb function and maintenance of good posture is increasingly difficult and complications are more likely. The condition is severe enough to shorten life expectancy but with high standards of medical care young men with Duchenne muscular dystrophy are often living well into their 30s.

Some boys with Duchenne muscular dystrophy will also develop learning and or behavioural difficulties. These difficulties can arise from the effect Duchenne muscular dystrophy has on the brain, others to physical limitations. Family support is essential and specialists maybe needed to address specific issues of learning and behaviour.

Other related publications

This fact-sheet is to be used alongside the following publications:

- The Diagnosis and Management of Duchenne muscular dystrophy- A guide for families
- An introductory guide for families with a child newly diagnosed with Duchenne muscular dystrophy
- Duchenne muscular dystrophy: A guide for families with a child aged 5-12 years

- Duchenne muscular dystrophy: The teenage years
- Physiotherapy Management for Duchenne muscular dystrophy
- Inclusive Education Guide
- Wheelchair Provision for Children and Adults with Muscular Dystrophy and other Neuromuscular conditions
- Muscle Biopsies factsheet
- Steroids and Duchenne muscular dystrophy factsheet
- Manifesting carriers of Duchenne muscular dystrophy factsheet

## References

1. TREAT-NMD network *The diagnosis and management of Duchenne muscular dystrophy* at <http://www.treat-nmd.eu/diagnosis-and-management-of-DMD/>
2. A, Emery, F, Muntoni, *Duchenne muscular dystrophy*, 3<sup>rd</sup> edition, 2003, Oxford University Press, New York
3. Muscular Dystrophy Campaign report, *Muscle disease: The impact* at [http://www.muscular-dystrophy.org/assets/0001/4711/MDC\\_incidencesreport\\_www.pdf](http://www.muscular-dystrophy.org/assets/0001/4711/MDC_incidencesreport_www.pdf)

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