



Duchenne muscular dystrophy (DMD)

Overview

Duchenne muscular dystrophy is a muscle-wasting condition caused by the lack of a protein called dystrophin. It usually affects only boys.

About 100 boys with Duchenne muscular dystrophy are born in the UK each year and there are about 2,500 boys and young men known to be living with the condition in the UK at any one time. For the general population, the risk of having a child with Duchenne muscular dystrophy is about one in every 3,500-5,000 male births.

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

Symptoms

In the early stages, boys with Duchenne muscular dystrophy show signs of muscle weakness, such as difficulties running, jumping, climbing stairs and getting up from the floor.

They can show a Gower's manoeuvre (needing to support themselves with hands on thighs as they get up from the floor), and a waddling gait (walking on their toes with arched lower back).

With the progression of the muscle weakness, boys become unable to walk as far or as fast as other children, and may fall down. 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, boys may experience more problems climbing stairs and getting up from the floor.

Steroids have significantly changed the natural course of Duchenne muscular dystrophy. They help to maintain the muscle strength and function over a certain period of time, and can delay the time when the boys may require a wheelchair. It is therefore difficult to define when boys will start using a wheelchair, as this might be different from one boy to another.

However, boys will usually need a wheelchair by the age of about eight to 11 years (sometimes a little earlier or later). At first, they will probably use the wheelchair only for long distances. Later, they are likely to need to use the wheelchair full-time. At this stage, they may experience difficulties raising their arms above shoulder level.

With further progression of muscle weakness, the maintenance of good posture is increasingly difficult and complications are more likely. The condition is severe enough to shorten life-expectancy but nowadays, with high standards of medical care, most young men with Duchenne muscular dystrophy reach adulthood.

Family support is essential and specialists may be needed to address specific issues of learning and behaviour.

Causes

Duchenne muscular dystrophy is a genetic condition – it is caused by a mistake or mutation in the genetic code (DNA).

In Duchenne muscular dystrophy, the mutation occurs in a gene called dystrophin, which is located on the X chromosome or sex-chromosome (girls have two X chromosomes and boys have only one). In just over half of 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.

If a woman carries the mutation, then she is known as a 'carrier'. Usually female carriers are not affected because they have a second X chromosome, from which the dystrophin protein can be produced. A small number of female carriers have a degree of muscle weakness themselves, and they are known as 'manifesting carriers'.

Each son of a carrier has a 50:50 chance of being affected, and each daughter has a 50:50 chance of being a carrier.

Genetic advice (counselling), and testing for other family members at risk of being carriers, should be provided as soon as possible following the diagnosis of a boy with Duchenne muscular dystrophy. Your clinician or GP can arrange this for you.

Diagnosis

Most boys with Duchenne muscular dystrophy are not diagnosed until they start displaying symptoms, unless there is someone else in the family with the condition.

The first signs of Duchenne muscular dystrophy usually appear between the ages of one and three years and usually consist of problems with muscle function. Boys might start walking later than their peers, can fall more often or show difficulty running, jumping or getting up from the floor. They might have enlarged calf muscles.

Some boys with Duchenne muscular dystrophy have delayed speech development and this can be the first sign of the condition. If a blood test is done, high levels of a protein called creatine kinase (CK) are seen. CK is normally found in muscle but when muscles are damaged, such as in Duchenne muscular dystrophy, it leaks into the bloodstream.

The liver enzymes (aminotransferases, ALT and AST) are also often found to be high, as a consequence of muscle damage and not of a liver problem.

Duchenne muscular dystrophy has to be confirmed by genetic testing usually on a blood sample. Different types of genetic tests can provide specific and more detailed information about the DNA mutation.

Genetic confirmation is crucial. It enables families to make decisions about prenatal diagnosis in future pregnancies and for genetic testing to be available to other family members at risk of carrying the mutation in the dystrophin gene. Moreover, the genetic diagnosis will assist in determining if the boy qualifies for a number of clinical trials, which are currently running or are planned.

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 also help in some cases to distinguish between Duchenne muscular dystrophy and a milder form of the condition, known as Becker muscular dystrophy. However, the clinical signs and the genetic test can usually distinguish between the two forms, without the need for a muscle biopsy.

Treatment

No cure has yet been discovered, but there is promising research into the condition. A multi-disciplinary approach, with the input of specialists such as physiotherapists and occupational therapists, is the best way to manage Duchenne muscular dystrophy.

Having access to a multi-disciplinary team is vital to ensure someone with Duchenne muscular dystrophy receives a holistic approach to their care.

This means that in a single visit to your specialist neuromuscular centre, you can get important input from each health professional involved in your care. This includes respiratory, cardiac and physiotherapy professionals who are able to provide better support when working within a multi-disciplinary team.

Regular check-ups with a specialist doctor are important in order to make decisions about new treatments at the most appropriate time and, if possible, to foresee and prevent problems. It is recommended that you visit your doctor every six months, and the specialist physiotherapist about every three to four months.

The specialist physiotherapist will advise you on any interventions (such as stretching exercises), which might be required. It is important to allow your son to be as active as possible, and your specialist physiotherapist can guide you.

Steroids (prednisone or deflazacort) are often routinely prescribed for Duchenne muscular dystrophy, as they slow the decline in muscle strength and mobility over a certain period of time and prevent or postpone the development of complications. However, there are many possible side-effects which must be carefully managed.

Other drugs are beginning to become available for Duchenne muscular dystrophy, including Translarna (ataluren), which is currently available in some European countries to slow down the progression of symptoms in boys with Duchenne muscular dystrophy.

The drug works for only a small group of boys who carry a particular mutation in the dystrophin gene ('nonsense' mutation – where a single letter change in the DNA code results in a premature

stop codon). Your clinician will be able to tell you whether or not your son could benefit from this medicine. Other drugs targeting specific mutations may be approved in the coming years.

Intense research is continuing, in trying to find treatments for Duchenne muscular dystrophy. Some medicines are currently being tested in clinical trials.

It can also be useful to ask for a copy of the genetic report (with the type and the location of the mutation in the dystrophin gene identified in your child). This will help in understanding which medicine and trials might be suitable for your child.

The Duchenne Muscular Dystrophy Registry provides updated information on ongoing clinical trials for Duchenne muscular dystrophy and can help identify which children are potentially eligible for specific clinical studies. Your clinicians will be able to tell you how to register your child on this registry.

The North Star Adult Network, made up of neuromuscular expert consultants, allied health professionals, individuals living with Duchenne muscular dystrophy, and Muscular Dystrophy UK, is working together to improve the standards of care and support available to adults across the UK. This network mirrors the paediatric version – the North Star Project – which works to optimise the care of children with Duchenne muscular dystrophy.

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