



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.

Changing needs

This page is designed to help you understand the way Duchenne muscular dystrophy progresses and the expected timing of its complications. It is important to note, however, that all age-related timings are approximate and can vary from person to person.

With the introduction of standards of care in Duchenne muscular dystrophy, the natural course of the condition has significantly changed. There are standards of care for corticosteroid treatment, respiratory support and heart drugs, orthopaedic and physiotherapy management. Mobility and quality of life have improved considerably, and children with Duchenne muscular dystrophy now live into adulthood.

Duchenne muscular dystrophy is a severe and progressive muscle-wasting condition. Treatments and proactive interventions are available to help delay the complications of the condition, even though there is currently no cure. There are also several clinical trials currently in progress for potential new drug treatments for the condition.

As a mother of a son with Duchenne muscular dystrophy, I can't over-emphasise the importance of correct information. Once I had a good idea of Vivek's condition and how it would affect him, I was able to do some soul searching and find other ways of doing things.

It helped me to think and plan ahead for Vivek's changing needs. We can't control the future, but Vivek and I have been able to control his environment. That's been empowering for both of us.

Manjula Gohil, whose now adult son, Vivek, was diagnosed with Duchenne muscular dystrophy as a child.

Ages 7-11

During the junior school years, the child with Duchenne muscular dystrophy will usually experience deterioration of muscle strength and function, especially in the legs. Walking and climbing stairs become more difficult with time: the child tires more easily and tends to fall frequently. They need help to get up from the floor and will likely need a wheelchair for long distances.

Over this period, they might lose the ability to walk independently but the timing of loss of ambulation varies from child to child, and might depend on whether they have been on steroids and the response they have had to them. Walking or standing ability may be prolonged through the treatment with corticosteroids. Physiotherapy and orthotics (splints) are also used to help delay contractures and maintain mobility.

Main priority areas

- Regular stretching to preserve joint mobility and muscle tone.
- Medical/orthopaedic surveillance of contractures and use of orthoses (ankle splints, callipers, etc.).
- Support at school in lessons requiring physical activity.
- Attention to behavioural problems.
- Early consideration of needs for secondary education. These may include school adaptations, adapted toilet, and hoisting.
- Maintenance of independent mobility through provision of a wheelchair, with appropriate seating, to maintain good posture.
- Housing needs addressed so adaptations completed before stair-climbing becomes impossible, to ensure independent mobility in the home.

Ages 12-14

This can be a particularly difficult time for a child with Duchenne muscular dystrophy, as further deterioration of muscle strength can have an impact on mobility and independence. Home and school should have been fully adapted by now to allow maximum independence.

Walking may still be possible, although a wheelchair is usually required at this stage. An electric wheelchair is preferable, as it will enhance independence.

At this stage, the arms may be noticeably weaker. It may be tiring to write for a long time, so support at school is important.

Significant heart and breathing problems are unexpected at this age, although careful medical surveillance is important to ensure early changes are promptly treated. The number of hospital appointments might increase in this period.

Some children, especially those who have not been treated with steroids, may develop curvature of the spine (scoliosis). It is important that this is checked regularly, particularly if they use a wheelchair full-time.

Steroids have many positive effects, however side-effects – such as delayed puberty and growth, both of which can cause frustration – need to be addressed. As muscle function often deteriorates during adolescence, it's extremely important to ensure the young person's psychological wellbeing. Sports or social activities with other young people in similar situations can often be helpful.

Main priority areas

- Full access to school in mainstream or special education as chosen by the family. This includes full access to toilets, with appropriate hoisting arrangements as necessary.
- Medical surveillance, especially for spine, heart and lungs.
- Continued regular stretching to control joint contractures and to keep mobility.
- Psychological support.
- Address increasing need for physical support at school and at home.
- Respite arrangements for the child and their family.

Ages 14-16

At this stage, most of the adaptations at home should already be in place to enable maximum independence. Using an electric wheelchair will provide mobility both indoors and outdoors. IT and classroom support can be helpful and can compensate for weakness in the arms.

Heart function and breathing surveillance is essential and, if necessary, start treatment promptly.

The major medical complication at this stage is the development of spinal curvature, for which regular follow-up is needed. It is important to adapt the wheelchair to ensure good posture. Spinal surgery might be discussed, however it is only occasionally required in young people in this age group, who may have been treated with steroids since childhood.

There will generally be an increasing number of hospital appointments in this period.

Main priority areas

- Attention to post-school education or employment.
- Medical surveillance for heart and breathing function, as well as spinal curvature.
- Adequate support in the wheelchair to ensure good posture.
- Respite care.

Ages 16+

The availability of surgical management for scoliosis and the medical management of cardiac and respiratory failure have allowed some young people with Duchenne muscular dystrophy to live into adulthood. As the young person approaches their late teens, they are susceptible to chest infections and require closer medical surveillance. Treatment with corticosteroids has led to more young people retaining adequate respiratory function and some upper limb function into their late teens, as well as reducing the need for spinal surgery. Respiratory, heart and orthopaedic surveillance remain essential.

Encouraging independence is vital at this stage. Some young people may wish to attend college or university, and choose to live away from home. Care support, for example reviewing a care package, looking at employing more carers (who are not family members) or personal assistants (PAs) to enable this, need to be put in place to fully support their needs.

Growing into adulthood, the young person will have significant weakness in their muscles. It is therefore important that in these teenage years they develop skills and hobbies that can be fulfilling as an adult. Technological advances ensure that accessing the internet and computer use are possible, even for those with very weak muscles.

[The Joseph Patrick Trust \(JPT\)](#), the welfare fund within Muscular Dystrophy UK, provides grants towards the costs of essential powered mobility equipment for children and adults with muscular dystrophy or a related neuromuscular condition.

Most young people with Duchenne muscular dystrophy will, in their 20s and 30s (and sometimes younger), use a breathing machine (ventilator) at night because of weakness in the breathing muscles. This is a very effective treatment that improves quality of life and survival.

Cough assist machines are recommended in the NICE (National Institute for Health and Care Excellence) Accredited Guidelines for Duchenne muscular dystrophy and in NHS England's neuromuscular service specifications. This specialist equipment can help keep respiratory function as strong as possible and prevent potentially fatal respiratory problems.

Main priority areas

- Independence and provision of care support.
- Attention to tertiary education or employment.
- Constant medical surveillance.
- Respite care.

If you wish to learn more about the latest research and standards of care, contact our research team on research@muscular dystrophyuk.org.

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Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on 0800 652 6352 or info@muscular dystrophyuk.org.

Version: 04 / Date published: 1 March 2002 / Original author: Prof. KMD Bushby, Professor of Neuromuscular Genetics, University of Newcastle upon Tyne / Updated: 1 November 2015 / Updated by: Senior Clinical Associate, Michela Guglieri / Date of review: 1 November 2016