



Duchenne muscular dystrophy

Penny Southall, mum of Dan Hanson:

“My son, Dan, was diagnosed with Duchenne muscular dystrophy when he was three years old. I know the diagnosis can be devastating.

“Like we did, you may be wondering how you’ll ever come to terms with it. We found our own way of coping and you will too. What kept me going, was telling myself that he was still the same person, but now I knew something different about him. And if he felt OK, I could be OK.

“One of the first things I did after Dan’s diagnosis was to look up research on the quality of life experienced by people with Duchenne muscular dystrophy. Finding out he could have a fulfilling, satisfying life just like anyone else was a great comfort to me at the time, and as the years have gone by I have seen this first-hand.

“Dan has lasting friendships, he is doing well at school, he enjoys sports and he is looking forward to the future. There is so much help and support out there to help young people with disabilities try different activities and access the education they need.

“I asked my son, who is now 15, what he would say to families with a new diagnosis. He said, ‘It may seem bad at first, but it will turn out OK’.”

What is Duchenne muscular dystrophy?

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.

What are the causes of Duchenne muscular dystrophy?

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.

How is Duchenne muscular dystrophy diagnosed?

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.

Is there a treatment or cure?

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.

If you wish to learn more about the latest research from our team, contact the research department at: 020 7803 4813 or research@muscardystrophyuk.org

What is the prognosis?

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.

Some boys with Duchenne muscular dystrophy will also have learning and/or behavioural difficulties. These can arise from the effect Duchenne muscular dystrophy has on the brain. As learning difficulties in Duchenne muscular dystrophy are not progressive, it is important that they are promptly identified and addressed (e.g. at school) to give the child the support he needs to develop his skills and reach his full learning potential.

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

We're here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child's muscle-wasting condition, and keep you updated about progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same condition, who can tell you about their experiences
- ▶ tell you about – and help you get – the services, equipment and support you're entitled to.

Other useful publications and resources:

- ▶ 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
- ▶ wheelchair Provision for Children and Adults with Muscular Dystrophy and other neuromuscular conditions
- ▶ muscle biopsies
- ▶ steroids and Duchenne muscular dystrophy
- ▶ manifesting carriers of Duchenne muscular dystrophy
- ▶ we have an alert card available for Duchenne muscular dystrophy. Our alert cards are vital for emergencies. Order one through our helpline: 0800 652 6352

If you would like your GP or other health professional to have more information about Duchenne muscular dystrophy, we have some relevant materials. We've developed an online training module for GPs, as well as one for physiotherapists working with adults with muscle-wasting conditions. Contact our helpline or email us to find out more.



References

1. TREAT-NMD network, *The diagnosis and management of Duchenne muscular dystrophy* at www.treat-nmd.eu/diagnosis-and-management-of-DMD/
2. A. Emery, F. Muntoni, *Duchenne muscular dystrophy*, 3rd edition, 2003, Oxford University Press, New York
3. Muscular Dystrophy Campaign report, *Muscle disease: The impact* at www.muscular dystrophyuk.org/app/uploads/2015/06/MDC_incidencesreport.pdf
4. The Duchenne Muscular Dystrophy Registry is managed by *Action Duchenne*. For more news about clinical trials and the DMD Registry, contact us on: 0800 652 6352.

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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** from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.muscular dystrophyuk.org