

Duchenne muscular dystrophy

Some questions answered for parents who have just learned about the diagnosis

What is Duchenne muscular dystrophy?

It is one of more than 20 types of muscular dystrophy. All the muscular dystrophies are caused by faults in genes (the units of inheritance that parents pass on to their children) and they cause progressive muscle weakness because muscle cells break down and are gradually lost. The Duchenne type affects only boys (with extremely rare exceptions) and a problem in this gene is known to result in a defect in a single important protein in muscle fibres called dystrophin. It is named after Dr Duchenne de Boulogne who worked in Paris in the mid-19th century who was one of the first people to study the muscular dystrophies.

How serious is it?

This is a very serious condition. Most affected boys develop the first signs of difficulty in walking at the age of 1 to 3 years and are usually unable to run or jump like their peers, they often struggle to climb stairs and need to use a banister for support. Rising from the floor can also prove difficult.

As the condition progresses boys with DMD are unable to walk as far or as fast as other children and may occasionally fall down.

Some boys also have learning and or behavioural difficulties, which may begin to manifest at this stage.

By about 8 to 11 years (rarely earlier or a little later) boys become unable to walk and by their late teens or twenties the condition is severe enough to shorten life expectancy. There are however many forms of management which are now available, which have changed the outlook and which we believe in most cases can help with the complications of the condition.

How common is it?

About 100 boys with Duchenne muscular dystrophy are born in the United Kingdom each year. There are about 1500 known boys with the disorder living in the UK at any one time. For the general population the risk of having an affected child is about 1 in every 3500 male births.

Is there any treatment?

Unfortunately no cure has yet been discovered. We do have ways to manage the condition, which help with its complications. These have had a very important impact on the quality and length of life that can be expected with this condition. A great deal can be done to help limit the effects of the muscular dystrophy and this includes treatments which are now definitely known to help some of the problems which may be life threatening, but no treatment is known which affects the actual loss of muscle cells. Intensive research to find a cure is carrying on in many

centres around the world. You can find updates on progress in this area in the MDC magazine Target md and on the MDC website.

How is Duchenne muscular dystrophy diagnosed?

Reliable tests are available once somebody has recognised that a child's problems might be due to this relatively rare condition. All affected boys have very abnormally high levels of an enzyme called creatine kinase in their blood. Most hospital laboratories can do this test. But there are other even rarer causes of a positive creatine test, so for a specific diagnosis in families with no previous affected member other tests are needed. These may involve genetic testing (looking for the actual fault in the dystrophin gene) and a muscle biopsy which can be studied to look at the dystrophin protein.

Can we be sure there is no mistake in the diagnosis?

With the most reliable genetic and protein tests, the diagnosis is very clear indeed. There are only two conditions, which are at all likely to cause any confusion in diagnosis to a doctor experienced in Duchenne dystrophy - and both of these are other types of muscular dystrophy.

Autosomal recessive types are about 20 times rarer than the Duchenne type in boys and can be somewhat similar, but the specialised tests can pick out the differences.

The Becker type of muscular dystrophy is a milder variant of dystrophin deficiency but there may be some overlap in severity with the Duchenne type. It may be difficult in very young children to gauge severity at first but in the great majority of cases the position is clear. Protein and genetic tests can make this distinction clearer as well.

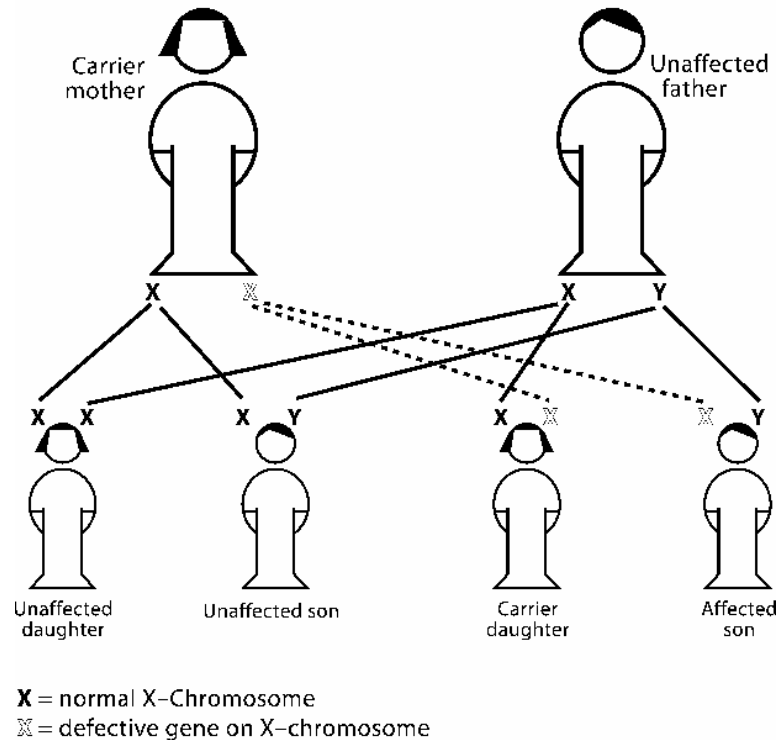
How is it inherited?

Duchenne muscular dystrophy is caused by an X-linked gene (that is, the gene is on the X chromosome; girls have two of these and boys only one). This means that only boys are affected but that their mothers may be carriers. Actually in almost half of all affected boys nowadays it turns out that the faulty gene has arisen by a change in the gene or 'mutation' in the boy himself and no other member of the family carries it. But this may be difficult to prove and can be decided only after careful and expert assessment of the family.

In just over half of all cases the mother carries the gene 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'.

One of the most important things that needs to be done soon after the diagnosis of a boy with Duchenne muscular dystrophy is to seek genetic advice and appropriate tests for those members of the family who are at risk of being carriers.

X-LINKED INHERITANCE



How can we be sure that no other boy in the family has it?

If a boy of any age has a normal creatine kinase blood test you can be sure that he is not affected and will never develop the condition.

Can any carriers in the family be identified?

This may be less easy but geneticists can identify from the family tree which women are at risk of being carriers. A combination of creatine kinase and DNA blood tests allows the great majority of such women to be either identified as carriers or given a strong reassurance that their risk is very low. Specialised genetic advice is now available to all families.

Can DMD be diagnosed before birth?

Once a child with DMD has been born in a family, it is often possible to offer prenatal diagnosis in future pregnancies, either for the mother or for other women who are found to be at risk of being carriers. This is normally possible when DNA studies give precise information that allows the status of the foetus to be identified. This is achieved by studying the foetus's own DNA in a chorion villus biopsy. This test is performed on a tiny piece of the developing placenta usually at about the 11th-12th week of pregnancy.

Further information is available in the Organisation's leaflet on *Carrier detection and pre-natal diagnosis*.

What medical help will he need now?

Very little in the early days after diagnosis. Active exercise is important but not necessarily with medical supervision though you may find it helpful to be in touch with a physiotherapist to keep an eye on things. The most important way in which a doctor can help at this stage is to help you, as parents, to learn as much as you can about Duchenne muscular dystrophy and to provide or arrange genetic advice. It should also be possible at this stage to set up an arrangement for long term follow up for continuing discussion and help.

What medical help will he need later?

Regular supervision from a clinic used to dealing with this condition will become increasingly important as the years go by. Knowing the kind of problems that can develop means that much can be done to prevent them or manage them appropriately. In the early stages, your son will be monitored carefully for signs of increasing problems with his mobility, and in particular for the development of shortening of his muscles (contractures). Physiotherapy plays an important role in helping here.

As getting about becomes more difficult, extra options to help will be discussed and a plan of action decided later on, attention will be paid to management of any spinal curvature and surveillance of heart and breathing muscles. Picking up problems in any of these areas means that they can be managed properly, reducing the long-term effects. Management of heart and breathing problem has improved dramatically over the last few years and this improvement is likely to continue.

What can we do to help our son?

Being a parent is a challenge even without having a child with DMD. This news will have made you reevaluate some of the hopes and expectations you had for your son. The new challenge is to move forward in the light of this information, to continue to grow as a family and value each member and ensure that he or she has an equal share of family time.

Sometimes unaffected brothers and sisters miss out on the attention and affection they need because their parents are preoccupied or sad. Time devoted to them and also, to each other will make your family life a more secure and stimulating base for your son. There will not be time for everything, and it is important to recognise that and not feel guilty, but recognising everyone's needs - including your own - is very important for you all.

You may well feel very alone. Everyone's experience is different, but in time you may find it helpful to speak to another family who have had a similar diagnosis. Your consultant or family care officer should be able to help with this.

There is no doubt that news like this changes a family in very fundamental ways. You are going to have to become an expert in this new and unwelcome subject to make sure you have the confidence to foresee and prevent problems through the years. However bad it seems to start with though, families do find a way to live with DMD. Your son **will** develop and mature and it is important that he is encouraged to achieve all the independence he possibly can.

What should I tell my children?

This is a very difficult question and one which people often worry about. As time goes by, your son, and other children if you have them, will inevitably ask questions, which you need to be prepared to answer openly and honestly as they come up. Knowing that they can ask you questions, which will be answered truthfully, will enhance your relationship. That is not to say that you need to tell them 'everything' at the first opportunity. Listen to what your children are asking and answer exactly that. You may find it helpful to rehearse how you will say things - people often say that this helps and the reality is then not nearly as bad as they had feared. It is also true that often your child will know more than you think. Allowing the opportunity to talk openly is very brave. It can be a lot more satisfactory in the long run than keeping up a silence wherein everyone is trying to protect each other. It is important to reassure your other children that they are not going to develop the same problem - this can sometimes be a hidden worry and that is nobody's fault.

What plans will we have to make for the future?

The previous paragraphs may give you some ideas about planning to help your son. One practical problem that needs to be planned for in advance is how he is going to get about in your house and remain as independent as possible when he can no longer climb stairs or when he uses a wheelchair. Will he be able to go without help to the toilet or his bedroom? Will he be able to use the bath etc? Solutions can be found to these problems but special equipment may be needed and sometimes the family home needs adaptations or even an extension.

The ideal house for a person in a wheelchair is sometimes a bungalow and you should consider whether you might try to find one, but for other people different solutions may be better. Local authority grants, building permission and the building work itself can all take an unbelievably long time to arrange and it can be a struggle to get what you want. It is vital to start thinking about these decisions in advance. It is also crucial to seek skilled advice before making an expensive decision about your home. The Muscular Dystrophy Campaigns Adaptations Manual, local social services and local Occupational Therapy (OT) services and perhaps your local Family Care Officers, may be able to help you get this advice.

What about school?

Most children with muscular dystrophy cope pretty well in their local infant's school and in their first few years at junior school. Talk to your school and give them as much information as you can. Your local clinic should be able to help you keep the school fully informed. If walking becomes too precarious or access to classrooms, toilets or dining room becomes too difficult some schools will make special arrangements or even structural alterations. Local authority

transport to and from school can be arranged if necessary as can help in the classroom. If the school is too unsuitable an alternative mainstream school in the district or a special school will provide the answer.

Alternatively when he leaves school your son will naturally find it difficult to do a job requiring any muscle strength, and his leisure activities will be restricted in the same way. From the earliest days at school his education is therefore going to be doubly important so that his

talents, whether as an artist, organiser, writer or whatever can be fully developed. Computers offer a real route to help children with DMD develop their talent to the best of their abilities. Higher or further education often offers good opportunities for school leavers.

Some boys with Duchenne muscular dystrophy are found also to have learning difficulties. This problem is rarely severe and when it does occur, unlike the muscle weakness, it never becomes worse as time goes by. In those boys who have the problem, language and communication skills are often the main difficulty. Manual skills, design sense and imaginativeness are often excellent which is perhaps why many young men with Duchenne muscular dystrophy become very good artists and model makers.

As parents you can work with the school to discover and develop your son's best talents as well as helping him to learn to cope with tasks he finds difficult.

The Duchenne Family Support Group

The Duchenne Family Support Group is run by parents who have children with Duchenne muscular dystrophy and offers informal support and friendship. For further details please contact:

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MC08

Published: 03/02

Updated: 04/08

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