

About Duchenne muscular dystrophy:

- Duchenne muscular dystrophy (DMD) is a serious genetic condition that causes progressive muscle weakness. With a few rare exceptions, DMD affects only boys.
- Boys with DMD first show signs of difficulty in walking between the ages of one to three years, and later cannot run, jump or manage stairs. By about 8-11 years, patients are unable to walk, and the condition is life-threatening by their early twenties.
- There is no cure for DMD at the moment, but there are treatments that can help with some of the complications of the disorder, especially with heart and breathing problems.
- DMD is caused by a genetic change, called a mutation. In around half of all affected boys, DMD appears 'out of the blue', but in the remaining cases the mother is an unaffected carrier of the condition. Any boys born to a carrier mother have a 50 per cent chance of being affected by DMD, while any girls have a 50 per cent chance of being a carrier themselves.
- DMD can be diagnosed by looking for unusually high levels of a substance called creatine kinase in a blood sample from the patient. Further tests may include a muscle biopsy or genetic test. A genetic test can also be used to identify female family members who may be unaffected carriers of the condition.

Find out more

To find out more about Duchenne muscular dystrophy and other types of muscular dystrophy, and for information and support contact:

Muscular Dystrophy Campaign
020 7720 8055
www.muscular-dystrophy.org

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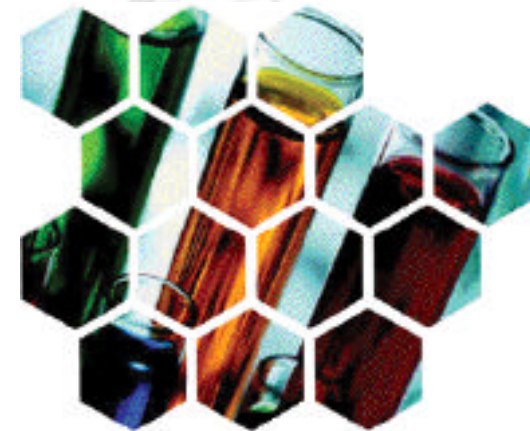
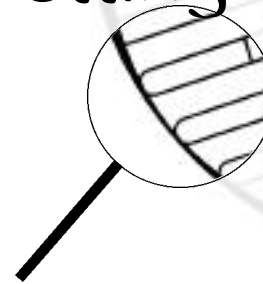
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Getting a diagnosis of...



Duchenne/Becker muscular dystrophies



Duchenne/Becker Muscular Dystrophies (D/BMD)

Who should have a genetic test for Duchenne/Becker Muscular Dystrophies (D/BMD)?

Duchenne and Becker muscular dystrophies (D/BMD) are two forms of an X-linked disease, caused by an absent or abnormal protein – called dystrophin – in muscle, resulting in progressive muscle weakness in affected boys. These abnormal proteins are caused by mutations in the dystrophin gene – the largest known human gene – located on the X chromosome. DMD affects 1 in 3500 boy births, whereas the milder BMD affects only 1 in 18500. The conditions can occur either spontaneously or by transmission of the mutation from an unaffected carrier mother.

Approximately 60-65% of the cases of D/BMD are caused by the loss of a significant portion of the dystrophin gene (a ‘deletion’), while 5-10% result from duplications of the DNA in the gene. The majority of the remainder result from individual small changes to the DNA code of the gene.

When a boy is diagnosed with D/BMD or is suspected of having D/BMD, a sample is taken for DNA analysis. Firstly, the DNA is examined for a deletion. Identification of a deletion as the disease-causing mutation enables carrier detection for women in the family. If a woman is found to be a carrier, prenatal diagnosis can be offered. Similarly, when a duplication is identified as the cause of D/BMD, women in the family can be offered carrier detection and prenatal diagnosis, if requested and appropriate.

Occasionally, no mutation is identified in a boy affected with D/BMD; then it is possible to identify the X-chromosome most likely to carry the mutation and to trace it through the family to identify carriers. This technique is known as gene tracking. Deletion/duplication analysis and tracking of the high risk X chromosome can both be used for prenatal testing, usually in cells obtained by chorionic villus sampling (CVS) at about 10-12 weeks gestation. There are still major problems associated with the laborious nature of most techniques used to recognise small mutations in D/BMD and so this analysis is, as yet, not widely available diagnostically.



Sue and Dave live in Leicestershire, with their children Eliot and Katy. Eliot, now aged three, was diagnosed with Duchenne muscular dystrophy (DMD) two years ago.

Sue first became concerned about Eliot when he hadn't started to walk by the time he was 18 months old:

Sue: *'I took him over to the doctor...he just said we'll do a blood test and some X-rays and things but he didn't actually say why'.*

Two days later, the doctor came to visit Sue and Dave, to tell them that the blood tests had showed that Eliot had Duchenne muscular dystrophy (DMD), a serious condition that causes progressive muscle weakness. He said that Eliot would probably need a wheelchair by the time he was 10 or 11, and that the condition would be life-threatening by his early twenties. Sue says that at first she was ‘devastated’ by the diagnosis, as was her husband when he found out:

Dave: *'I was driving, so I came straight off the road and rang the doctor...he said he'll be disabled so I just burst into tears and came straight home to Sue'.*

The couple then saw a specialist doctor, who told Sue and Dave more about DMD and put them in touch with the Muscular Dystrophy Campaign, a

support group for families affected by all types of muscular dystrophy.

'She was really good, she gave us all the information...we knew that she couldn't make us feel brilliant, but she helped us feel better'.

Both Sue and Dave are glad that Eliot's condition was picked up so early, and feel that knowing what's in store for him is helping them to plan ahead:

Sue: *'It's probably better for us to know right from the start, so when we have to tell Eliot we'll have known for a long while, and probably can cope better'.*

Dave: *'It saves us saying we wish we hadn't done this, or we wish we'd done that - we'll have done everything we can for him'.*

Sue: *'Yes - there's certain things like we said we'd like to take him to Disneyland when he's older...obviously now we probably want to take him when he's seven or eight, while he's still walking and he'll be able to enjoy it - I'm definitely glad for the diagnosis'.*

DMD is caused by a genetic change, called a mutation, on the X-chromosome, which means that only boys are usually affected. Often the condition appears ‘out of the blue’, but sometimes the boy's mother may be a carrier. So when Sue found out she was pregnant again, the family went to see a geneticist:

Sue: *'We found out about Eliot in the February and I found out I was pregnant in the April...I was tested to see if I was a carrier or not, before we decided whether I'd need any tests or not with the pregnancy'.*

Sue found out she was carrying the DMD mutation, and so had a prenatal test when she was 12 weeks pregnant. The results showed the baby was a girl, and therefore could not be affected by DMD. Katy, now 16 months old, will be able to decide if she wants to know her carrier status or not when she is older.