If a father has DMD

Men who have DMD have an altered copy of the dystrophin gene on their X chromosome. If a man with DMD has children, all of his daughters will become carriers of DMD. This is because a man with DMD passes his X chromosome on to all of his daughters (he only has one X chromosome).

The sons of a man with DMD will be unaffected, because they inherit his Y chromosome.

For more information

If you need any more advice about any aspect of DMD, you are welcome to contact:

Clinical Genetics Department
Northern Scotland (main base Aberdeen) Tel: 01224 552120
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh) Tel: 0131 537 1116
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 354 9201
(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

The DMD Family Support Group
78 York Street, London, W1H 1DP
Helpline: 0800 121 4518
Website: www.dfsg.org.uk
Email: info@dfsg.org.uk

The Muscular Dystrophy Campaign
61A Great Suffolk Street
London
Helpline: 0800 652 6352 (Mon-Fri 830-6pm)
Website: www.musculardystrophyuk.org
Email: info@musculardystrophyuk.org

If you have a family history of Becker Muscular Dystrophy or know you are either affected with the condition or are a carrier and planning to start a family, you would be welcome to contact your genetic department for further advice. Where at all possible, having this discussion in advance of a

Seen in clinic by ..................................................
Duchenne muscular dystrophy (DMD) is a genetic condition that affects all the muscles of the body, causing them to become progressively weak. It is caused by an alteration in an important gene called dystrophin. The dystrophin gene normally makes a protein that is essential for keeping the muscles strong and healthy. If the dystrophin gene has an alteration in it, the production of this protein does not occur and the body does not have a mechanism to replace damaged muscle tissue or to grow new muscle cells.

DMD is a serious condition that worsens over time. Boys with DMD may be slow at learning to walk, fall more easily and are unable to run and jump like their peers. They often struggle to climb stairs and to rise from the floor.

Some boys also have a mild learning disability and delayed speech or behavioral problems.

As muscles progressively weaken mobility is affected and boys will inevitably lose the ability to walk necessitating the use of a wheelchair. Improved knowledge, management and treatment is increasing the age that independent mobility is lost in the majority of cases. This has also improved the management of other systems that tend to cause problems in the late teens including respiratory muscles, the heart muscle and muscles of the gastrointestinal tract.

Although no cure for DMD has yet been discovered, intensive research is continuing to find a cure and effective treatments for DMD.

How is DMD diagnosed?

A doctor usually diagnoses this condition after parents notice their child is having problems with walking or other physical activities.

A blood test will reveal an increased level of the enzyme, creatine kinase (CK) and enzyme released by the muscles. If this is high, genetic testing of a blood sample will identify an alteration in the Dystrophin gene in the majority of cases. Very rarely, a muscle biopsy may also be necessary to confirm the diagnosis of DMD is correct.

If a specific gene alteration is identified, female family members can be tested to see if they are carriers of the condition.

What are genes and chromosomes?

A gene is a tiny segment of genetic material found in all cells in our bodies which are like instructions or recipes. Genes determine many of our personal characteristics, such as eye and hair colour. Genes lie on tiny structures called chromosomes, rather like beads (the genes) threaded onto a string (the chromosomes). Each chromosome contains thousands of genes.

Most of our body cells have 46 chromosomes, arranged in 23 pairs. We inherit one of each pair from our mother and the other of each pair from our father.

One of these 23 pairs determines whether we are male or female. These are the sex chromosomes. Females have two X chromosomes, one inherited from their mother and the other from their father. (XX)

Males have one X chromosome (inherited from their mother) and a Y chromosome inherited from their father. (XY)

How is DMD inherited?

Most boys with DMD inherit the gene alteration from their mother. In a small percentage (about one-third), the gene alteration is not inherited but happens for the first time when the boy is conceived.

It is not possible for a boy to inherit DMD from his father (even if the father has DMD), because boys inherit a Y chromosome from their fathers, and the dystrophin gene is on the X chromosome.

It is extremely rare for female carriers of DMD to develop the muscle symptoms seen in males with the condition. However, rarely the heart muscle can be affected in later life and monitoring is recommended. (Further information is available from the genetic clinic)

Will children inherit the DMD gene?

If a mother is a carrier of DMD

If a mother carries an alteration in the DMD gene on one of her X chromosomes, she is known as a carrier of DMD. A mother who is a carrier of DMD has a 50% (1 in 2) chance of passing on her normal X chromosome and a 50% (1 in 2) chance of passing on her altered X chromosome in each pregnancy.

This means that each of her sons has a 50% (1 in 2) chance of developing DMD, and each of her daughters has a 50% (1 in 2) chance of being a carrier of DMD.

### Carrier mother

**X**  
**X**

- Boys who inherit this X will develop DMD. Girls who inherit this X will be a carrier of DMD.

Both boys and girls who inherit this X will be healthy children.