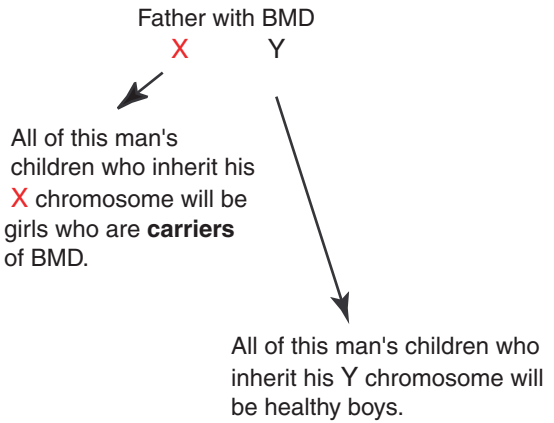
If a father has BMD

Men who have BMD have an altered copy of the dystrophin gene on their X chromosome. When a man with BMD has children, all of his daughters will become carriers of BMD. This is because a man with BMD passes his X chromosome on to all of his daughters (he only has one X chromosome). The sons of a man with BMD will be normal healthy males, because they inherit his healthy Y chromosome.



Tests during pregnancy

It may be possible to have a test during pregnancy to discover if a baby has inherited the BMD gene. If you would like to find out more about these tests, please ask to speak to a genetic counsellor or doctor.

For more information

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

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 Fax: 01224 559390
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035 Fax: 01382 645731
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)
Tel: 0131 651 1012 Fax: 0131 651 1013
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 201 0808 Fax: 0141 201 0361
(Glasgow, Argyll & Bute, Aryshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

The Muscular Dystrophy Campaign
7-11 Prescott Place
London
SW4 6BS
Helpline: 020 7720 8055
Website: www.muscular-dystrophy.org
Email: info@muscular-dystrophy.org

Seen in clinic by .....

This leaflet was written by Clinical Genetics, Guy's Hospital. Updated by Genetic Interest Group Scotland. Last updated Jan 2003.

Becker Muscular Dystrophy

CLINICAL GENETICS CLINICAL GENETICS

Patient Information Leaflet

What is Becker Muscular Dystrophy (BMD)?

BMD is a condition that affects all the muscles of the body, causing them to become progressively weaker. It is caused by an alteration in an important gene called **dystrophin**. The dystrophin gene normally makes a protein that is essential for keeping our muscles strong and healthy. If the dystrophin gene has an alteration in it, the production of this protein does not occur normally and the body may not be able to replace damaged muscle tissue or to grow new muscle cells.

BMD is sometimes confused with a condition called Duchenne Muscular Dystrophy (DMD), which is caused by an alteration in the same dystrophin gene. However DMD is a more serious condition than BMD.

Boys with BMD can sometimes be slow to learn to walk. Symptoms usually occur later in childhood, beginning with cramps during exercise. Most people with BMD are not very athletic in childhood and struggle with school sport. In early adulthood, it may become difficult to walk quickly, run, climb stairs or lift heavy objects. Some people with BMD require a wheelchair later in life, although many do not.

The symptoms of BMD vary greatly from one person to another. Some people lose their ability to walk in early adulthood. Others are able to continue walking throughout their lives. Life expectancy can be reduced, but many people with BMD live into their 70s and 80s. Some may develop heart trouble in early adulthood, others never do. Even people in the same family can have very different symptoms.

How common is it?

BMD is a rare condition. About 1 in 30,000 newborn babies are affected with this condition.

How is BMD diagnosed?

BMD is usually diagnosed after someone notices that they are having problems with muscle weakness.

A blood test will reveal an increased level of the enzyme, creatine kinase (CK, sometimes also known as CPK). A muscle biopsy may also be necessary to confirm that the diagnosis of BMD is correct.

Once the diagnosis is confirmed, genetic testing is often offered to identify the exact nature of the gene alteration. If a specific gene alteration is identified, other family members can be tested to see if they are carriers of the condition. In about one-third of cases, no alteration in the gene can be found with current testing methods.

What are genes and chromosomes?

A gene is a segment of DNA that has a particular purpose. Genes determine many of our personal characteristics, such as eye colour 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.
- 44 of these chromosomes are numbered in order of decreasing size, as 22 matching pairs.

The two remaining chromosomes are called **'sex chromosomes'** and they are given letters **(X and Y)** rather than numbers.
- 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 BMD inherited?

Most boys with BMD inherit the gene that causes it from their mother, who carries an altered copy of the dystrophin gene on one of her two X chromosomes. 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 BMD from his father (even if the father has BMD), because boys inherit a Y chromosome from their fathers, and the dystrophin gene is on the X chromosome.

As a general rule:

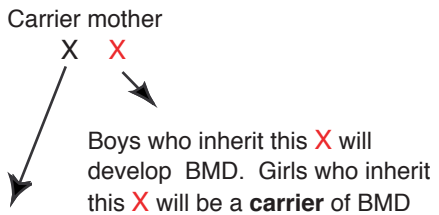
BOYS GET BMD, GIRLS DO NOT.

Will children inherit the BMD gene?

If a mother is a carrier of BMD

If a mother carries an alteration in the BMD gene on one of her X chromosomes, she is known as a carrier of BMD. Females have two X chromosomes (XX), and when they have children they pass only one of their X chromosomes to each of their children. A mother who is a **carrier** of BMD has a 50% chance of passing on either her normal X chromosome or the X chromosome that carries the altered copy of the dystrophin gene.

This means that each of her sons has a 50% chance of developing BMD, and each of her daughters has a 50% chance of being a **carrier** of BMD.



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