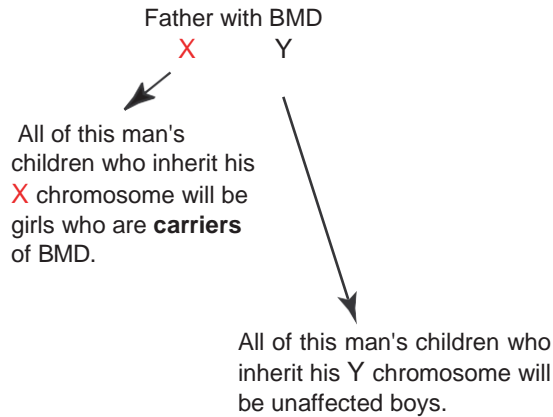


### If a father has BMD

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 be 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 unaffected because they inherit his Y chromosome.



### Having children

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

### 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

(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)

#### Muscular Dystrophy UK

61A Great Suffolk

Street

London

SE1 0BU

Helpline: 0800 652 6352 (Mon-Fri 830-6pm)

Website:

[www.muscular dystrophyuk.org](http://www.muscular dystrophyuk.org)

Email: [info@muscular dystrophyuk.org](mailto:info@muscular dystrophyuk.org)

# Becker Muscular Dystrophy



Patient Information Leaflet

Seen in clinic by .....

This leaflet was written by Clinical Genetics, Guy's Hospital.

Updated by Yvonne Robb on behalf of ScotGEN, July 2018

## What is Becker Muscular Dystrophy (BMD)?

Becker Muscular Dystrophy (BMD) is a genetic condition causing progressive muscle weakness of the arms, legs and trunk. Symptoms are usually only seen in males. It is caused by an alteration or change in an important gene called Dystrophin. The Dystrophin gene usually 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 is disrupted and as a result causes muscle weakness.

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

Symptoms of BMD can occur any time from early childhood to the fourth decade or beyond. Children may have walking difficulties, falls and cramps during exercise. Most people with BMD describe being not very athletic in childhood and struggle with school sports. In early adulthood, it may become difficult to walk quickly, run, climb stairs or lift heavy objects.

However, symptoms of BMD vary greatly from one person to another. Some people are able to continue walking throughout their lives. Others lose their ability to walk and need to use a wheelchair some time in their adult life. Some may develop heart muscle weakness in 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 17,000 newborn babies are affected with this condition.

## How is BMD diagnosed?

BMD may be suspected when someone develops symptoms of muscle weakness, as above.

A blood test will reveal an increased level of the enzyme, creatine kinase (CK). Genetic testing of a blood sample will identify an alteration in the Dystrophin gene in the majority of cases. Very occasionally, a muscle biopsy may also be necessary to confirm the diagnosis of BMD is correct.

Once a gene alteration is identified, females in the family can be offered a genetic blood test 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. Males have an **X** and a **Y** sex chromosome (**XY**) and females have two X sex chromosomes (**XX**). The Dystrophin gene is found on the **X** chromosome.

## How is BMD inherited?

Most boys with BMD inherit the altered gene 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.

It is extremely rare for female carriers of BMD 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 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. A mother who is a **carrier** of BMD 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% chance of developing BMD, and each of her daughters has a 50% chance of being a **carrier** of BMD.

