



**The Leeds
Teaching Hospitals**
NHS Trust

Yorkshire Regional Genetics Service

Becker Muscular Dystrophy (BMD)

Information for
patients



Yorkshire Regional
Genetics Service

What is Becker Muscular Dystrophy?

Becker Muscular Dystrophy (BMD) is a condition that affects all the muscles of the body, causing them to become progressively weaker.

Boys with BMD can sometimes be slow to learn to walk but symptoms usually occur later in childhood, beginning with cramps during exercise. Most people with BMD are not very athletic in childhood and may struggle with sport at school. 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 or breathing problems early in adulthood. People with BMD should have heart and breathing checks every few years to look out for this. The severity of symptoms can vary even within families.

In some people, BMD can also affect the brain to cause mild learning difficulties; however, most people with BMD will not notice any problems with learning.

BMD is sometimes confused with a condition called Duchenne Muscular Dystrophy (DMD), which is caused by an alteration in the same gene called dystrophin; however, BMD is less severe and has a later onset than DMD.

BMD is rare and affects only about 1 in 30,000 people.

What causes BMD?

BMD is caused by an alteration (*mutation*) in a gene called dystrophin, which is found on the X chromosome. The dystrophin gene normally makes a protein that is essential for keeping our muscles healthy and strong. If the dystrophin gene has an alteration in it, then the protein is not produced correctly and the body may not be able to replace damaged muscle tissue or grow new muscle cells.

How is BMD diagnosed?

BMD is usually diagnosed after problems with muscle weakness are noticed. A blood test will reveal an increased level of the enzyme creatine kinase. A genetic test and / or muscle biopsy may be necessary to confirm the diagnosis of BMD. In a small number of cases, scientists are unable to find an alteration in the dystrophin gene.

How is BMD treated?

The main result of BMD is muscle weakness. People may need help from physiotherapy and occupational therapy to help them stay as mobile as possible. They might use orthotics, walking aids and in some cases, a wheelchair.

Medications can be used to help any heart problems. There are not currently any treatments to stop the muscle weakness, although some are being testing in clinical trials.

What does this mean for other family members?

If a BMD gene alteration is identified, other family members can be tested to see if they are carriers of the condition, if they wish. This can give them information about their own health and whether they need any extra health checks. This can also give them information on their chance of having a child with BMD.

What are genes and chromosomes?

Genes are the unique set of instructions inside our bodies which makes each of us individual. There are many thousands of different genes, each carrying a different instruction. As well as determining how we look, our genes control the way each cell or building block of the body works.

Genes lie on tiny structures called chromosomes. Each chromosome contains thousands of genes.

Most of our body cells have 46 chromosomes, arranged in 23 pairs numbered 1 - 22. The two remaining chromosomes are the sex chromosomes as they determine gender. Females have two X chromosomes, one inherited from each parent. Males have one X chromosome and one Y chromosome.

We inherit one of each chromosome pair from our mother and the other from our father. Men always inherit their X chromosome from their mother and their Y chromosome from their father.

How is BMD inherited?

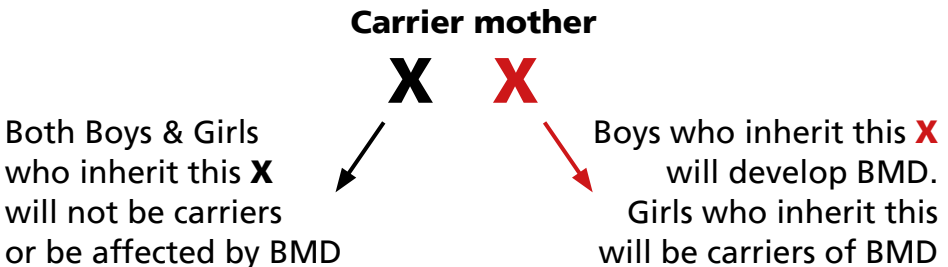
Most boys with BMD inherit the gene that causes the condition from their mother, who carries an altered copy of the dystrophin gene on one of her two X chromosomes. In a small percentage, the gene alteration is not inherited but happens for the first time when a boy is conceived. It is not possible for 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 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.



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). He would not pass BMD onto a son because he passes his Y chromosome on to sons.

Tests before / during pregnancy

It may be possible to have a test during pregnancy to discover if a baby has inherited the BMD gene alteration. It may also be possible to undergo a form of IVF (called Pre-implantation genetic testing) to reduce the chance of having a child with BMD. If you would like to find out more about these tests, please ask to speak to a genetic counsellor or doctor.

For more information:

There are several useful websites for people with BMD:

- Muscular dystrophy UK

This has lots more useful information about BMD and it is possible to download an alert card, which explains the condition.

- The DMD Registry

This allows you to register to stay up to date with news about treatments or clinical trials. It is for BMD and DMD patients.

If you need more information, please contact your local Genetics Department. If you live in the Yorkshire region, please contact:

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