

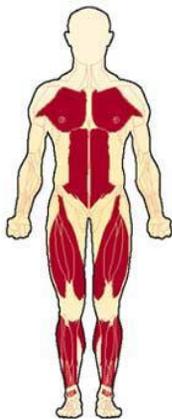
# Duchenne Muscular Dystrophy & Becker Muscular Dystrophy

## What are Muscular Dystrophies?

The muscular dystrophies are a group of genetic disorders that are characterised by progressive muscle weakness. There are differences in the various types of muscular dystrophy in the rate at which the muscle weakness progresses and the muscle groups which are most severely affected. The age of onset at which symptoms first appear in the different types varies from childhood through adult life.

This fact sheet focuses on two particular types of muscular dystrophy: **Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD)**.

Both DMD and BMD affect males predominantly, affected females are very rare. Figure 1 shows the muscles that are affected during the early stage of Duchenne and Becker muscular dystrophy.



**Figure 1:** In the early stages, Duchenne and Becker muscular dystrophy affect the pectoral muscles - which draw back the shoulders, the trunk and the upper and lower legs. These weaknesses lead to difficulty in rising, climbing stairs and maintaining balance. (Picture from the Muscular Dystrophy Association website: <http://www.mdausa.org/>)

## What are the features of Duchenne muscular dystrophy (DMD)?

Duchenne muscular dystrophy usually presents in early childhood with delayed milestones, including delays in standing independently. The first thing parents usually notice is that the calf muscles are enlarged. By approximately 8 to 11 years of age affected boys are unable to walk and most are wheelchair bound by 12 years of age. Heart disease occurs in all patients after age 18. Shortening of some muscles leads to a typical pattern of restriction of joint movement called contractures.

Most affected adolescents develop a sideways curvature of the spine called scoliosis, which may impair breathing and adds to discomfort. Few survive beyond the third decade, with respiratory complications and heart disease being common causes of death.

Intellectual ability is usually normal in people with DMD, although intellectual disability is found to occur in these individuals more often than in the general population. Unlike the physical disability, the mental handicap, if present, is not progressive.

A very small number (5-10%) of female carriers of the “faulty” gene have a mild degree of muscle weakness themselves and are then known as ‘manifesting carriers’. However, it is extremely rare for a female to show symptoms.

## What are the features of Becker muscular dystrophy (BMD)?

BMD is characterised by the same pattern of muscle weakness but is less severe and disabling than DMD. BMD usually has a later onset than DMD and is sometimes only diagnosed during adolescence or adulthood. The effects of BMD on the joints, spine, heart and lungs are generally milder. BMD is, however, very variable in severity, even between affected family members. Some people with BMD are able to walk only until early adulthood, others to an advanced age. Survival in some affected people is to middle age, but others have survived more than 80 years. Some develop heart problems in early adulthood; others never do.

## GENETICS:

As this is a genetic condition, genetic counselling is strongly recommended. Genetic counselling provides information on the condition, its inheritance pattern, risks to other family members and the prognosis (probable outcome). Psychosocial support and information about testing, including diagnostic testing, carrier testing, presymptomatic and prenatal testing (when appropriate and available) is offered.



Division of Human Genetics  
University of Cape Town

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## How common is DMD and BMD?

DMD is one of the most frequent forms of muscular dystrophy, affecting approximately 22 per 100 000 male births, while BMD affects 3 per 100 000 male births. Of DMD patients, 60% have inherited the “faulty” gene (mutation) from one of their parents (familial mutations). In 30% of DMD patients the genetic change occurs for the first time in the formation of the egg or the sperm; this is called a spontaneous mutation. In these cases the affected person would be the first in the family to have the mutation. Spontaneous mutations occur less frequently in BMD patients (approximately 10%).

## What genes are related to DMD and BMD?

DMD is caused by mutations in the dystrophin gene. The dystrophin gene is located on the X chromosome and when a fault (mutation) occurs it leads to the formation of a faulty protein in muscle fibres. The dystrophin protein is absent or non-functional in DMD. Whereas in BMD the type of fault (mutation) makes the dystrophin molecule abnormal. It is still able to function, but to a lesser degree than normal. Having some dystrophin protects the muscles of those with BMD from degenerating as badly or as quickly as those of people with DMD. The function of dystrophin in the muscle fibre is complex, but we know that when it is absent or abnormal the muscles slowly become weaker.

## How do people inherit DMD or BMD?

The dystrophin gene is carried on the X chromosome and inherited in an **X-linked recessive** manner. (Refer to Fact sheet 11.)

## When is Genetic Testing appropriate?

Genetic testing can be done to confirm a diagnosis (a diagnostic test), determine whether a person is a carrier for DMD or BMD (carrier testing) or to predict if a person or an unborn child will develop DMD or BMD (a predictive or a prenatal test).

Genetic services should be rendered with comprehensive genetic counselling. The Division of Human Genetics at the University of Cape Town can be contacted in this regard.

## How soon will I have the results?

Results will be available within approximately 4 weeks of the test. The results will be communicated to you personally via your general practitioner, neurologist or by the staff of the Division of Human Genetics at the University of Cape Town.

## Is there treatment available for DMD and BMD?

Currently there is no known treatment that alters the actual loss of muscle cells. Much can be done to help limit the effects of the muscular dystrophy. Steroids may help to delay disease progression, but should only be used under strict medical supervision. Prevention of scoliosis is one of the main challenges in the treatment of affected individuals. Back support and surgery is used to combat this. The discovery of the dystrophin gene and knowledge of the function of the protein has raised hope that rational therapeutic strategies will be developed.



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## Where can I read more about DMD and BMD?

You may find the following recourses about Muscular Dystrophies helpful.

1. Genetics Reviews:

<http://www.genetest.org/>

2. Muscular Dystrophy Association website:

<http://www.mdausa.org/>

3. Muscular Dystrophy Foundation (MDF) of South Africa:

<http://www.mdsa.org.za/>

## Are there DMD and BMD support groups?

The DMD and BMD support groups in South Africa falls under the Muscular Dystrophy Foundation of South Africa. Please contact your local MDF office for information..

### • National Office:

P.O. Box 1535,

Pinegowrie, 2123

**Tel:** (011) 789-7634, **Fax:** (011) 789-7634

**Email:** national@mdsa.org.za

### • Cape Branch:

P.O. Box 752,

Goodwood, 7459

**Tel:** (021)592-7306, **Fax:** (021)592-7306

**Email:** cape@mdsa.org.za

## Who do I contact for more information regarding testing?

### Division of Human Genetics Molecular Laboratory:

Prof. Jacquie Greenberg

(021) 406-6299

### Genetic Nurses:

(021) 406-6304