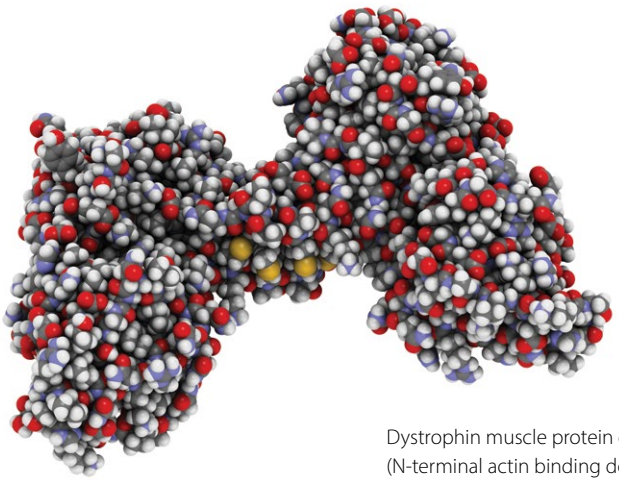
Becker muscular dystrophy

Becker muscular dystrophy is a genetic condition where complications can be minimised by adhering to a management programme specially designed by medical professionals.

Becker muscular dystrophy (BMD) is named after German doctor Peter Emil Becker, who first distinguished the symptoms from other muscular dystrophies in the mid-1950s. BMD is considered to be a milder form of Duchenne muscular dystrophy (DMD), as both are caused by mutations in the same gene, and thus have similar symptoms. BMD occurs once in approximately 30,000 live male births.



Dystrophin muscle protein domain (N-terminal actin binding domain).

Features of BMD

BMD is variable in severity, depending on the type of mutation in the dystrophin gene. It is less severe than DMD and usually has a much later onset. Some people with BMD are able to walk well into adulthood. In some cases, BMD may not be diagnosed until after adolescence.

The following features may be displayed:

- Muscle weakness.
- Muscle cramps.
- Fatigue.
- Breathing distress.
- Skeletal deformities.
- Unusual walking gait; waddling.
- Difficulties in hopping, running, jumping.
- Muscle deformities – pseudo hypertrophy of calf; contractures. (Contractures are muscles or tendons that have remained too tight for too long, thus becoming shorter. Once they occur they cannot be stretched or exercised away.)

Some children with BMD will experience further complications:

- A minority of BMD children will display intellectual problems or learning difficulties.
- Behavioural difficulties will on occasion arise with BMD. Such problems are usually mild, and can be managed.
- Rarely, heart disease such as cardiomyopathy will occur as the disease progresses.
- Contractures occur as scar tissue replaces normal elastic tissue. This prevents normal movement in that area, first in the ankles, then knees, hips and joints of the upper body.

The causes

BMD is a genetic condition caused by a defect in the dystrophin gene located on the X chromosome. The faulty gene results in a deficiency of the protein dystrophin, causing muscles to deteriorate and break down in males. The dystrophin gene is located on the X chromosome. Females have two copies of the X chromosome. A woman who has one correct dystrophin gene and one faulty dystrophin gene can nearly always produce enough dystrophin to have normal muscle function.

She is therefore a "carrier" of the mutation or a "genetic carrier" of DMD or BMD. Males have only one X chromosome and therefore one dystrophin gene copy. So if a male has a faulty dystrophin gene he will be affected with DMD or BMD since

he cannot produce the correct amount or type of the dystrophin protein.

The pattern of inheritance of BMD is called X-linked recessive. If a woman is a carrier of the mutation (she has one copy of the faulty dystrophin gene), and she has a daughter, there is one chance in two that the daughter will be a carrier of the mutation and one chance in two that she will not.

For each son of a genetic carrier, there is one chance in two, i.e. a 50 percent probability, of being affected and an equal chance of not being affected.

Since women have two X chromosomes, if one X chromosome has the defective gene, the other X chromosome functions to produce enough dystrophin for normal muscle function. Males on the other hand, have one X and one Y chromosome; thus they do not have a compensatory X chromosome, and will develop symptoms.

Spontaneous mutations are responsible for approximately one-third of BMD cases, with the genetic fault arising in the affected boy himself. This happens when the mutation in the dystrophin gene happens by chance in the formation of the egg or sperm. With a spontaneous mutation, the boy will be the first in his family to have BMD.

Managing BMD

As yet, there is no treatment that can overcome the progressive muscle weakness of BMD. It is possible, however, to minimise complications by adhering to a management programme designed by a team of medical professionals. The team will usually be headed by a paediatric specialist, and include a physiotherapist, occupational therapist, with specialists as required.

- **Exercise:** Both passive and active exercises play an important role in BMD management. Walking is easily achieved in the early stages, but can become more difficult as strength declines. Walking sticks and aids can be valuable in prolonging mobility. Swimming is good for ensuring all muscles are exercised, and the joints mobilised.

  Passive exercises, or assisted stretching, should be established as early as possible. A physiotherapist is invaluable in developing an exercise programme to delay the shortening of muscles (contractures). These exercises should be undertaken on a daily basis, and will often require assistance. Moderate exercise rather than heavy strenuous exercise is important. People who have MD disorders are more likely to tire quickly and overdoing it may cause irreparable muscle damage.
- **Supportive Equipment:** If, and when, contractures develop in the ankle joints, a type of orthotic may be offered to be worn at night. These ankle splints will help maintain the joint in a normal position, and may help reduce pain from muscle cramping.

  Standing frames may prolong standing and walking, although it is likely that a wheelchair will eventually be needed.

  The suitability of the home environment is important to consider at an early stage, so that future adjustments can be made over time.
- **Medical Treatment:** Many medicines and dietary supplements have been tried over the years to treat the symptoms of BMD. So far there is only one group of drugs – the catabolic steroids – that have shown any significant benefit. Prednisone and Deflazacort have been shown to slow the loss of muscle function, or even to increase strength. This option needs to be discussed with the child's doctor as there are possible side effects.
- **Nutrition:** Excessive weight gain can occur from reduced physical activity produced by the muscle weakness. It is more important for an individual with BMD than the average person that weight is monitored and that a well-balanced diet is followed.
- **Surgery:** If contractures develop at the ankle joints, these can be surgically treated by release of the Achilles tendon. This procedure is usually done once the child is wheelchair dependent, and helps improve their foot position. Having a comfortable foot position may help prolong mobility for some BMD boys. Spinal fusion surgery is performed to correct scoliosis. Becker boys who undergo this 'spinal fusion' are usually very pleased with the outcome.
- **Respiration:** The normal defences people use to rid themselves of excessive secretions do not function effectively in BMD boys, and the early treatment of sniffles and sore throats, and the prevention of chest infections are important. Family and caregivers must watch carefully for signs of disrupted sleep due to respiratory problems. ®