

BECKER MUSCULAR DYSTROPHY



What is Becker Muscular Dystrophy?

Becker muscular dystrophy (BMD) is an inherited degenerative muscle disease. It is a milder and less progressive variation of Duchenne muscular dystrophy (DMD).

Both disorders are caused by a mutation of the dystrophin gene which causes a deficiency of the protein dystrophin. This protein is an important building block which helps give muscles structure and strength.

This form of muscular dystrophy is named after the German doctor Peter Emil Becker who described the condition in 1955. While the disease usually affects boys, girls in BMD families are often carriers and can pass on the gene for the disease to the next generation.

What are the symptoms of BMD?

Babies are born with the mutated dystrophin gene, however, symptoms of BMD typically appear in the teens or early adulthood. Initial symptoms include

weakness in the leg and pelvis muscles. Another sign is enlarged calf muscles. With progression, walking becomes more difficult, leading to an increased risk of falls. Muscles lose their elasticity, leading to tightness (contractures) around joints, and difficulty with stretching leg and heel muscles. Contractures can lead to skeletal deformities such as scoliosis or a curved spine. Due to weakening of the heart muscle, people with BMD have a high risk of developing heart disease (cardiomyopathy). Heart health should be monitored closely.

How is BMD diagnosed?

Several diseases, such as DMD, limb-girdle muscular dystrophy and spinal muscular atrophy, have some of the same symptoms as BMD, and so it is important to get a proper diagnosis. In addition to a family history and physical examination, a blood test will examine the level of an enzyme called creatine kinase (CK). Elevated CK levels indicate a problem with the muscles, causing the enzyme to leak out. A genetic test that analyzes the dystrophin gene and determines the type of genetic mutation will confirm the diagnosis. In some cases, a muscle biopsy is also helpful to understand what is happening inside the muscle, and to demonstrate the reduced expression of dystrophin.

How is BMD inherited?

BMD is known as an X-linked recessive genetic defect. This means it is an inherited disease due to a defective gene on the X chromosome. Women have

two X chromosomes (one from her mother and one from her father). Men have one X chromosome (from his mother) and one Y chromosome (from his father).

With two X chromosomes, women have a lower risk of manifesting symptoms of the disease. If one X chromosome has the defective gene, the second X chromosome likely has a working copy of the gene to compensate. Women who inherit the defect in one X chromosome are unlikely to develop the disease themselves, but they can pass the defective gene on to their children.

The fact men have only one X chromosome works against them if they inherit the defective gene. Without a second X chromosome to compensate for the defective gene, they will develop symptoms.

In some cases, the genetic mutation is not inherited, but develops spontaneously within the early development of an embryo.

What is the risk of passing on the gene for BMD to offspring?

A woman who carries the gene has a 50% risk of passing the gene for BMD to each child she bears. Daughters who inherit the mutation will be carriers and may pass the defect on to their children. Sons with the mutated gene will eventually develop symptoms. People with a family history of the disease may choose to have genetic counseling to help understand the risks of passing the gene on to their children.

What treatments are available for BMD?

There is no cure for BMD or other muscular dystrophies. However, a variety of treatments help relieve symptoms. Treatments tend to be supportive

and include physiotherapy, use of supportive braces, assistive devices, occupational therapy, speech therapy, and in some cases surgery.

- Physiotherapy is useful for teaching techniques to help delay loss of function and how to conserve energy.
- Devices such as knee, leg or back braces help keep muscles flexible.
- For some people, surgery can help relieve advanced problems with tendons.
- Occupational therapy helps teach people how to use wheelchairs and other assistive devices, as well as training on new ways for doing daily tasks.
- While corticosteroids, such as deflazacort and prednisone, have proven successful at slowing the progression of DMD, research has yet to prove its effectiveness on BMD patients.

Research is ongoing in areas such as gene therapy, stem cell research, myoblast therapy and pharmacological treatments.

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