Becker muscular dystrophy (BMD) is a genetic disorder characterized by progressive weakness and degeneration of the skeletal muscles that control movement. Heart muscle also is commonly affected, making cardiac problems a prominent feature of the disease.

The incidence (rate of occurrence of new cases) of BMD has been estimated to be between 1 in 18,000 and 1 in 30,000 live male births.

BMD is classified as a dystrophinopathy, a muscle disease that results from the deficiency of a protein called dystrophin. Because the DMD gene is located on the X chromosome, BMD primarily affects males, while females typically are carriers. Some females can experience varying ranges of physical symptoms, including muscle weakness and cramps, and are therefore called manifesting carriers.

BMD typically is inherited through the mother; however, in some of cases, it occurs spontaneously in people who do not have a family history of the disease.

In both Duchenne muscular dystrophy (DMD) and BMD, a mutation in the DMD gene interferes with production of the dystrophin protein, a critical component in the formation and maintenance of healthy muscle. A BMD-causing mutation typically leads to an abnormal version of the dystrophin protein that is only partially functional. However, lack of fully functional dystrophin protein in muscle cells causes them to be fragile and become damaged with use.

There is no cure for BMD, but medications, assistive devices, and therapy can help manage some symptoms and improve quality of life.
What are the signs and symptoms of BMD?

BMD is a multi-systemic condition, affecting many parts of the body and resulting in atrophy of the skeletal, cardiac (heart), and pulmonary (lung) muscles.

**Nervous system**
- Developmental delay
- Motor delay

**Cognition**
- Learning disability
- Behavioral deficits

**Gastrointestinal**
- Dysphagia
- Constipation
- Reflux
- Gastroparesis

**Lungs**
- Breathing difficulties
- Respiratory infections
- Sleep apnea

**Skeleton and muscle**
- Muscle weakness
- Fatigue
- Loss of balance
- Impaired coordination
- Muscle cramps
- Difficulty walking
- Difficulty climbing stairs
- Exercise intolerance

**Heart**
- Cardiomyopathy
- Congestive heart failure
- Arrhythmias
- Swelling in the feet and lower legs

**Sleep**
- Apnea
- Nocturnal cough
- Excessive daytime sleepiness

**Nephrology**
- Proteinuria

**Neurology**
- Hypoesthesia
- Sensory loss
- Motor weakness

**Endocrinology**
- Hypothyroidism
- Hypogonadism
- Diabetes mellitus

**Skin**
- Skin atrophy
- Increased skin laxity

**Musculoskeletal**
- Short stature
- Low bone density

**Cervical spine**
- Kyphoscoliosis
- Lordosis
- Contractures
- Scoliosis

**Pulmonary**
- Breathlessness
- Cyanosis
- Fontanels

**Pharyngeal**
- Glottal incompetence

**Symptoms of BMD**

- In some cases, the first sign of disease may be a heart condition called dilated cardiomyopathy. This form of heart disease enlarges and weakens the heart muscle, preventing it from pumping blood efficiently. Dilated cardiomyopathy progresses rapidly and can be life-threatening.

- Skeletal muscle weakness often begins in the legs and pelvic area and slowly progresses to the muscles of the shoulders, neck, arms, and respiratory system. Over time, affected individuals begin to have difficulty walking, running, hopping, and jumping, and they may have frequent falls. Although loss of muscle mass is common, the calves may become enlarged.

- Some individuals with BMD will have problems getting up from the floor and may use a distinctive method known as Gower’s maneuver or Gower’s sign to “walk” their hands up their thighs in order to stand up.

- In other symptoms of BMD, children may have problems getting up from the floor and using a method known as Gower’s maneuver or Gower’s sign to “walk” their hands up their thighs in order to stand up.

- The diaphragm and other muscles involved in breathing typically weaken, making the lungs less effective at moving air in and out. Signs of poor respiratory function can include headaches, difficulty concentrating or staying awake, and nightmares. Weakened respiratory muscles also make it difficult to cough, leading to increased risk of serious respiratory infection.

- Medical and scientific advances are helping to improve quality of life, and people with BMD often survive into their 40s and beyond.

How is BMD treated?

Over-the-counter pain relievers may be used to treat muscle pain or cramps.

Wheelchairs or scooters may be used to help maintain mobility.

Braces, also called orthoses, support the ankle and foot and may extend up over the knee. Ankle-foot orthoses (AFOs) are sometimes prescribed for night wear to keep the foot from pointing downward and keep the Achilles tendon stretched while a child is sleeping.

Physical therapy through exercise helps to restore and maintain muscle strength and function. Stretching helps to maintain range of motion.

Speech therapy can help with swallowing problems.

Assisted ventilation can help treat respiratory muscle weakness.

Treatment with angiotensin converting enzyme (ACE) inhibitors and beta blockers may be used to slow the course of cardiac muscle deterioration in BMD. Cardiac evaluations are recommended beginning at around 10 years old, or when symptoms first begin, and be repeated at least every two years.

Ediducational and psychological interventions can help with learning disabilities.

Ocational therapy can help improve daily living and work skills.

Corticosteroids (such as prednisone and Emflaza) may be prescribed in BMD to help preserve muscle strength and function, to prevent scoliosis, and to prolong the time that people with BMD can walk. It’s thought that they work, at least in part, by reducing inflammation. However, corticosteroids also cause unwanted side effects such as increased appetite, weight gain, loss of bone mass, and cataracts.

Please talk to your medical provider to obtain more information on these treatments.
MDA Glossary

**Atrophy**
A decrease in the size and mass of muscle tissue

**Cardiomyopathy**
A condition in which the heart muscle is weakened, making it harder for the heart to pump blood to the body

**Contracture**
A shortening of muscles or tendons around joints that can limit mobility

**Corticosteroid**
One of a group of steroid hormones that has been shown to dampen the inflammatory response in damaged muscle

**Dysphagia**
Difficulty swallowing

**Gastroparesis**
A condition that affects the ability of the stomach to empty its contents into the small intestine, even though there is no blockage; also known as delayed gastric emptying

**Gower’s maneuver**
A person’s use of their hands and arms to “walk” up their own body in order to rise from a squatting position; indicates weakness of the muscles in the hips and legs

**Lordosis**
Posture characterized by an inward curving of the lower back

**Muscular dystrophy**
A term that refers to a number of diseases that cause progressive loss of muscle mass, resulting in weakness and sometimes, loss of mobility

**Mutation**
A flaw in the DNA code

**Pseudohypertrophy**
A condition in which muscles become enlarged with deposits of fat and fibrous tissue

**Scoliosis**
An abnormal sideways curvature in the spine that occurs when weakened muscles are unable to hold the spine straight

To learn more about BMD, visit mda.org or contact the MDA National Resource Center at 833-ASK-MDA1 (275-6321).