CONDITION

Becker muscular dystrophy

Learn about Becker muscular dystrophy, including muscle and heart symptoms, genetic causes, diagnosis, treatment, complications, and follow-up care.

WHAT IT IS
Inherited muscular dystrophy causing progressive weakness

BODY SYSTEM
Muscles and heart

MAIN SYMPTOMS
Proximal muscle weakness, difficulty climbing stairs, frequent falls, muscle cramps

WHAT CAUSES IT
Inherited DMD gene change

HOW IT IS DIAGNOSED
DMD genetic testing, sometimes muscle biopsy

MAIN TREATMENT
Coordinated supportive care

CAN IT BE CURED?
No, genetic change cannot be corrected

IS IT AN EMERGENCY?
Sometimes, severe trouble breathing

LONG-TERM OUTLOOK
Variable progression with serious heart or breathing complications

Overview

Becker muscular dystrophy is an inherited condition that causes muscles to weaken over time. It is one of several disorders called dystrophinopathies, which result from changes in the gene that makes dystrophin. Dystrophin is a protein that helps protect muscle fibers during movement.

In Becker muscular dystrophy, the body usually produces some dystrophin, but the protein may be shorter or may not work normally. This generally leads to a slower and more variable course than Duchenne muscular dystrophy, a related condition in which little or no working dystrophin is produced.

Becker muscular dystrophy mainly affects skeletal muscles, which control movement. It can also affect the heart muscle. Some people develop significant heart disease even when their

muscle weakness is mild. Regular heart monitoring is therefore an important part of care.

The condition most often affects males because the altered gene is on the X chromosome. Females who carry a dystrophin gene change often have no muscle symptoms, but some develop weakness, muscle pain, or heart problems.

Symptoms may begin in childhood, adolescence, or adulthood. Their severity and rate of progression vary widely. Some people continue walking independently for many years, while others need mobility support earlier. Treatment cannot currently correct the underlying genetic change, but coordinated care can protect mobility, support heart and lung health, and help people remain active and independent.

Symptoms

The first signs often involve weakness in the muscles around the hips, pelvis, thighs, or shoulders. This is called proximal muscle weakness because it affects muscles close to the center of the body.

A child may have trouble keeping up with peers, running, jumping, or climbing stairs. An adolescent or adult may notice reduced stamina, frequent falls, or difficulty rising from a low chair. Some people use their hands to push against their thighs when standing from the floor. This movement is called Gowers' sign.

Common symptoms include:

Weakness usually progresses gradually. The legs are often affected before the arms. Over time, some people need ankle braces, a cane, a scooter, or a wheelchair. The timing is different for each person.

Becker muscular dystrophy can also affect the heart. Dilated cardiomyopathy occurs when the heart's main pumping chamber becomes enlarged and weakened. Abnormal heart rhythms may also occur. Early heart disease may cause no symptoms, which is why routine testing matters. When symptoms occur, they may include shortness of breath, reduced exercise tolerance, ankle swelling, palpitations, chest discomfort, dizziness, or fainting.

Breathing muscles may weaken later in the condition. Possible signs include morning headaches, daytime sleepiness, restless sleep, weak coughing, or shortness of breath when lying down.

Learning and behavior differences are less prominent than in Duchenne muscular dystrophy, but they can occur. Some people have difficulties with attention, language, learning, or mood. These concerns deserve assessment and support rather than being assumed to be part of the muscle condition.

- Difficulty running, climbing stairs, or walking uphill
- A waddling or toe-walking gait
- Frequent falls or problems with balance
- Muscle cramps, pain, or unusual tiredness after activity
- Enlarged-looking calves caused partly by replacement of muscle with fat and connective tissue
- Tightness in the ankles or other joints
- Weakness in the shoulders and upper arms
- Difficulty getting up from the floor or a chair

Causes and risk factors

Becker muscular dystrophy is caused by a disease-causing change in the DMD gene. This gene provides instructions for making dystrophin. Without enough working dystrophin, muscle fibers are more easily damaged during normal contraction and repair.

The condition follows an X-linked inheritance pattern. Males usually have one X chromosome, so a disease-causing change in their only copy of the DMD gene can produce symptoms. Females usually have two X chromosomes. If one carries the change, the other often produces enough dystrophin to prevent major weakness, although this protection is not complete for everyone.

A carrier has a chance of passing the altered X chromosome to each child. Sons who inherit it may develop a dystrophinopathy. Daughters who inherit it are usually carriers but may have muscle or heart symptoms. A genetic counselor can explain inheritance based on a family's specific test results.

Not everyone has a known family history. A new, or de novo , genetic change may occur in an egg cell, during early development, or less commonly in a sperm cell. The absence of affected relatives does not rule out the condition.

The type and location of the DMD gene change influence how much functional dystrophin the body can make. However, genetic findings do not always predict the exact age of onset or severity. People with similar gene changes can have different symptoms.

Diagnosis

Diagnosis usually begins with a medical history, family history, and physical examination. A clinician may assess walking, balance, joint movement, muscle strength, and the ability to rise from the floor.

A blood test for creatine kinase, often called CK, is commonly used. CK is an enzyme that leaks from damaged muscle cells. Levels are usually much higher than normal in Becker muscular dystrophy, even before obvious weakness develops. A high CK level shows muscle injury but does not identify the cause by itself.

Genetic testing is used to look for a disease-causing change in the DMD gene. Testing often begins by checking for missing or duplicated sections of the gene. If those tests do not find the cause, sequencing may be used to look for smaller changes. Confirming the genetic result can distinguish Becker muscular dystrophy from other muscle conditions and clarify testing options for relatives.

A muscle biopsy is now needed less often. During a biopsy, a small piece of muscle is removed and examined for dystrophin and other signs of muscle disease. It may be considered when genetic testing is inconclusive or when the diagnosis remains uncertain.

The evaluation may also include:

Testing relatives should be voluntary and guided by genetic counseling. Female carriers may need heart monitoring even if they have no muscle symptoms.

- An electrocardiogram to check the heart's electrical activity
- An echocardiogram or cardiac MRI to examine heart structure and pumping function
- Breathing tests to measure lung function
- Assessment by physical, occupational, or rehabilitation specialists
- Genetic counseling for the affected person and relatives

Treatment and management

Care is usually coordinated through a neuromuscular clinic or a team familiar with dystrophinopathies. The plan depends on age, symptoms, heart and lung findings, mobility, and personal goals.

Physical therapy helps preserve comfortable movement and reduce joint tightness. A therapist can teach gentle stretching and recommend safe ways to stay active. Swimming, cycling, and other low-impact aerobic activities may support fitness without placing excessive stress on weak muscles. Exhaustive exercise or activity that causes prolonged pain and weakness may be harmful.

Occupational therapy can help with daily tasks, school, work, and home adaptations. Ankle-foot braces may maintain ankle position or make walking safer. Mobility aids can reduce falls, conserve energy, and support participation. Using a device does not mean that exercise or independence has failed.

Corticosteroids like prednisone or hydrocortisone are widely used in Duchenne muscular dystrophy, but evidence for their benefits and risks in Becker muscular dystrophy is more limited. A neuromuscular specialist may discuss them in selected situations. Treatment decisions should account for possible effects on weight, bones, blood pressure, mood, blood sugar, and infection risk.

Heart treatment may be recommended when tests show cardiomyopathy or reduced pumping function, sometimes before symptoms develop. Depending on the findings, a cardiologist may use medicines that lower stress on the heart, help control blood pressure, remove excess fluid, or manage abnormal rhythms. Some people may need an implanted device to treat serious rhythm problems or support the heart's pumping pattern.

Breathing care may include regular lung-function testing, help with clearing mucus, treatment of sleep-related breathing problems, or noninvasive ventilation. Noninvasive ventilation uses a mask to support breathing, often during sleep.

Maintaining a healthy weight and adequate nutrition can make movement and breathing easier. If swallowing becomes difficult, a speech-language pathologist or dietitian can assess food texture, safety, and nutrition.

Before surgery or sedation, the anesthesia team should know about the dystrophinopathy and any heart or breathing problems. Certain anesthetic exposures can increase the risk of severe muscle breakdown and dangerously high potassium in people with dystrophin disorders. Planning with an experienced team reduces this risk.

Complications

Cardiomyopathy is one of the most important complications. It may progress without noticeable symptoms and can lead to heart failure or dangerous heart rhythms. Regular cardiac surveillance can detect changes before they cause major problems.

Progressive weakness can lead to falls, fractures, joint contractures, and loss of mobility. A contracture is a joint that becomes fixed because muscles or tendons have shortened. Stretching, positioning, braces, and timely mobility support may reduce their effect.

Weak breathing muscles can cause an ineffective cough, repeated chest infections, or low oxygen and high carbon dioxide during sleep. Spinal curvature may add to breathing difficulty

in some people, although severe scoliosis is less common than in Duchenne muscular dystrophy.

Muscle breakdown can sometimes cause dark brown or cola-colored urine, especially after unusual exertion, illness, or certain anesthetic exposures. This can injure the kidneys and needs prompt medical assessment.

Emotional strain may affect the person with Becker muscular dystrophy and their family. Changing physical abilities can influence school, work, relationships, and mental health. Counseling, peer support, and practical accommodations can be part of medical care.

Follow up

Routine follow-up should include review of strength, mobility, falls, pain, joint movement, equipment needs, and participation in daily activities. Heart evaluations are needed regularly, including for many female carriers. The care team may also monitor breathing, sleep, bone health, nutrition, and emotional well-being.

Contact the usual care team soon if there is a clear decline in walking, new swelling in the legs, increasing shortness of breath, repeated falls, morning headaches, sleep-related breathing symptoms, or new difficulty swallowing. Dark urine after exercise or illness needs prompt same-day medical advice.

Seek emergency care for severe trouble breathing, fainting, new chest pressure or pain, a rapid or irregular heartbeat with weakness or breathlessness, or sudden inability to move with dark urine. These symptoms do not always indicate a complication of Becker muscular dystrophy, but they require urgent evaluation.

A personal emergency plan can list the diagnosis, medicines, heart and lung findings, anesthesia precautions, and specialist contact details. Keeping a copy available may help during unexpected care.

Sources

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