CONDITION

Duchenne muscular dystrophy

Duchenne muscular dystrophy is an inherited condition that causes worsening muscle weakness from childhood.

WHAT IT IS
Inherited condition causing progressive muscle weakness

BODY SYSTEM
Muscles, heart, lungs, bones, and nervous system

MAIN SYMPTOMS
Delayed movement skills, frequent falls, difficulty climbing, muscle weakness

WHAT CAUSES IT
Inherited or new DMD gene change

HOW IT IS DIAGNOSED
Genetic testing, sometimes muscle biopsy

MAIN TREATMENT
Steroids, rehabilitation, heart and breathing care

CAN IT BE CURED?
No - treatments can slow progression

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or collapse

LONG-TERM OUTLOOK
Progressive disability, with survival improving through coordinated care

Overview

Duchenne muscular dystrophy, or DMD, is a genetic condition that causes muscles to weaken over time. It begins in childhood and mainly affects muscles used for movement, but it can also affect the heart and breathing muscles.

DMD occurs when the body cannot make enough working dystrophin, a protein that protects muscle cells during movement. Without it, muscle cells are easily damaged and gradually replaced by fat and scar tissue.

The condition mainly affects boys because of how the responsible gene is inherited. Girls and women who carry a DMD gene change can sometimes develop muscle weakness or heart problems. Rarely, a girl has the full condition.

There is no cure, but coordinated care can protect movement, breathing, heart health, bones, comfort, and independence. Some treatments may slow muscle decline or target particular genetic changes.

Symptoms

Symptoms usually appear in early childhood. Weakness often starts around the hips, thighs, and shoulders.

Early signs can include:

Using the hands to rise from the floor is called Gowers' sign. It suggests hip-area weakness but is not specific to DMD.

Weakness progresses over time. Muscles and tendons may shorten, causing contractures that limit joint movement. Most people eventually use a wheelchair. Wheeled mobility can reduce falls, conserve energy, and support independence.

DMD can also affect learning, speech, attention, emotions, or social communication, although intellectual ability varies widely.

Heart changes may not cause early symptoms. Later signs can include unusual tiredness, shortness of breath, palpitations, dizziness, or swelling. Weak breathing muscles may cause restless sleep, morning headaches, daytime sleepiness, a weak cough, or repeated chest infections.

- Sitting, standing, or walking later than expected
- Frequent falls or difficulty keeping up with other children
- Trouble running, jumping, or climbing stairs
- Toe walking or a waddling walk
- Using the hands to "climb up" the legs when standing from the floor
- Enlarged-looking calves caused by muscle being replaced by fat and scar tissue

Causes and risk factors

DMD is caused by a harmful change in the DMD gene, which contains instructions for making dystrophin.

The gene is on the X chromosome. Boys usually have one X chromosome, so a harmful change in their only copy can cause DMD. Girls usually have two X chromosomes, and the second copy

often provides enough working dystrophin to prevent the full condition.

A child may inherit the change from a mother who carries it, or it may arise for the first time in the child. DMD can therefore occur without a known family history.

Some carriers develop muscle weakness or heart muscle damage. Genetic testing and heart monitoring may be offered to relatives after a diagnosis. Genetic counselling can explain inheritance, testing choices, and possible implications for family members, although it cannot predict exact symptoms or progression.

Diagnosis

A clinician may suspect DMD after reviewing a child's movement, development, family history, and physical examination.

A blood test for creatine kinase, or CK, is usually an early step. CK leaks into the blood when muscle cells are damaged and is often very high in DMD, but this does not confirm the condition.

Genetic testing identifies harmful DMD gene changes and confirms the diagnosis in most people. Finding the precise change may also show whether a gene-targeted treatment could be considered.

A muscle biopsy is needed less often. It involves removing a small muscle sample to examine dystrophin and muscle damage and may be considered when genetic testing is inconclusive.

Other assessments may include heart imaging, an electrocardiogram, breathing tests, bone health checks, and reviews of movement, nutrition, learning, and emotional wellbeing.

Treatment and management

DMD care usually involves a specialist neuromuscular team working with rehabilitation, education, social care, and local health services.

Corticosteroids like prednisone or hydrocortisone are commonly used to preserve muscle strength and function for longer. Monitoring is needed for effects including weight gain, slowed growth, mood or behaviour changes, high blood pressure, eye problems, weakened bones, and reduced natural steroid production.

Steroids should not be stopped suddenly unless directed by the medical team. During serious illness, surgery, or repeated vomiting, someone taking long-term steroids may need an emergency plan because their body may not produce enough stress hormones.

Physical and occupational therapy can support movement and independence through gentle activity, stretching, positioning, splints, mobility equipment, and changes at home or school. Exhausting exercise and heavy resistance work can damage vulnerable muscles, while prolonged inactivity can worsen stiffness and fitness. A specialist therapist can recommend a suitable balance.

Heart care starts before symptoms develop. Regular checks can identify heart muscle weakness or rhythm changes, and medicines may be used to protect the heart or treat cardiomyopathy.

Breathing care may include lung-function tests, assisted coughing, prompt treatment of chest infections, and non-invasive ventilation through a mask, often first used during sleep.

Some treatments target specific DMD gene changes. Gene-transfer treatment may also be available to some people in certain countries. Eligibility, evidence, risks, monitoring, and availability vary. These treatments do not restore normal muscle or replace ongoing heart, breathing, and rehabilitation care.

Nutrition, swallowing, bone health, pain, puberty, mental health, and educational support are also important. Surgery may sometimes be considered for fractures, severe contractures, or spinal curvature.

Complications

Progressive weakness can cause reduced mobility, falls, contractures, and scoliosis. Reduced movement and steroid treatment can weaken bones and increase fracture risk. A fracture may cause a sudden and lasting loss of mobility.

Weak breathing muscles can make coughing less effective, increasing the risk of mucus retention and serious chest infections. Breathing may also become too shallow during sleep.

Most people with DMD develop some cardiomyopathy, and abnormal heart rhythms can occur. Regular monitoring is important because heart changes may develop without noticeable symptoms.

Swallowing weakness can cause long meals, weight loss, dehydration, or food entering the airway. Some people instead gain excess weight as mobility decreases or during steroid treatment.

Anaesthesia requires special planning. Certain anaesthetic medicines can cause dangerous muscle breakdown or high potassium levels in people with DMD. Always tell the anaesthesia team about DMD before surgery or sedation.

Prevention

DMD cannot be prevented through exercise, diet, pregnancy behaviour, or routine medical care. It results from a genetic change that may be inherited or arise unexpectedly.

When a DMD gene change is known, genetic counselling can explain carrier testing and reproductive options, including testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices and do not change the condition in someone who already has DMD.

Planned heart and breathing checks, safe activity, bone protection, vaccinations, prompt infection treatment, and careful steroid management can reduce or identify some complications earlier.

Living with it and what to expect

DMD is progressive, but its exact pace cannot be predicted. Genetic differences, treatment response, heart and breathing health, injuries, and access to coordinated care can affect the course.

During childhood, running, climbing, and rising from the floor usually become harder. Braces, scooters, standing devices, powered wheelchairs, and transfer aids can improve comfort, safety, participation, and independence.

When walking is no longer practical, care focuses on preserving arm and hand function, comfortable seating, access to technology, skin care, and preventing contractures. Heart and breathing monitoring becomes increasingly important through adolescence and adulthood.

Many people with DMD attend mainstream education, form relationships, work, pursue interests, and participate in decisions about their lives. Support may be needed for personal care, transport, housing, education, employment, and finances.

Psychological support can help people and families manage changing abilities, uncertainty, anxiety, grief, or low mood. Discussions about future care should be honest, paced appropriately, and guided by the person's preferences and communication needs.

Modern heart and respiratory care has improved survival into adulthood. DMD still shortens life, and serious heart or breathing complications remain possible. Regular follow-up allows care to change as needs evolve.

When to get medical help

Arrange routine or prompt follow-up for noticeable changes in walking, falls, pain, sleep, swallowing, weight, mood, or daily activities. Report new palpitations, increasing tiredness, morning headaches, a weaker cough, or more frequent chest infections.

Contact the specialist team promptly after a fall or injury, especially if there is pain, swelling, or refusal to use a limb. Fractures can occur after relatively minor injuries.

Seek emergency medical help for:

Bring the person's DMD and steroid emergency information to urgent appointments whenever possible.

- Severe or rapidly worsening difficulty breathing
- Blue, grey, or unusually pale lips or skin
- Collapse, fainting, severe chest pain, or a sustained racing heartbeat
- Choking with ongoing breathing difficulty
- Sudden weakness with dark urine after anaesthesia or a major illness
- Severe illness or repeated vomiting in someone taking long-term steroids, especially if an emergency steroid plan cannot be followed

Common questions

Is Duchenne muscular dystrophy always inherited from the mother?

No. A mother may carry and pass on the change, but it can also occur for the first time in the child. Genetic testing can help clarify the likely inheritance pattern.

Can girls have Duchenne muscular dystrophy?

Yes, but it is uncommon. Girls who carry a DMD gene change may also develop milder muscle symptoms or heart problems and should be assessed individually.

Does a high CK blood test mean my child has DMD?

Not by itself. A high CK level shows muscle damage and occurs in several conditions. Genetic testing is usually needed to confirm DMD.

Will every person with DMD need a wheelchair?

Most eventually use wheeled mobility because leg weakness progresses, although timing differs. A powered wheelchair can prevent falls, conserve energy, and support participation.

Can exercise make DMD worse?

Very strenuous or high-resistance exercise may damage muscles. Gentle, low-impact activity and guided stretching can support comfort and general health. A neuromuscular physiotherapist can recommend suitable activity.

Are new genetic treatments a cure?

No. Some gene-targeted treatments may slow decline or improve certain measures in eligible people, but they do not fully restore dystrophin or prevent every complication.

Sources

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