CONDITION

Muscular dystrophies

Muscular dystrophies are inherited conditions that cause muscles to weaken over time.

WHAT IT IS
Inherited conditions causing progressive muscle weakness

BODY SYSTEM
Muscles, movement, breathing, and sometimes heart

MAIN SYMPTOMS
Muscle weakness, falls, walking difficulty, muscle stiffness

WHAT CAUSES IT
Inherited gene changes affecting muscle function

HOW IT IS DIAGNOSED
Genetic testing, blood tests, sometimes muscle biopsy

MAIN TREATMENT
Rehabilitation, supportive care, and subtype-specific medicines

CAN IT BE CURED?
No - treatments manage symptoms or slow progression

IS IT AN EMERGENCY?
Sometimes - sudden breathing difficulty or severe chest symptoms

LONG-TERM OUTLOOK
Progression and life expectancy differ greatly by subtype

Overview

Muscular dystrophies are a group of inherited conditions that cause muscle weakness and loss of muscle tissue over time. They result from changes in genes needed to build, protect, or repair muscles.

There are many types. These include Duchenne, Becker, limb-girdle, facioscapulohumeral, myotonic, congenital, Emery-Dreifuss, and oculopharyngeal muscular dystrophy. Each affects the body differently. Some begin in infancy or childhood, while others do not become noticeable until adulthood.

Certain types mainly affect the muscles used for movement. Others can also affect the heart, breathing muscles, swallowing, eyes, hormone systems, or other organs. Myotonic dystrophy may cause myotonia , meaning muscles have trouble relaxing after they tighten.

Muscular dystrophy is different from muscle weakness caused by inactivity, injury, medication, or another medical condition. Identifying the exact type matters because monitoring, treatment choices, inheritance, and long-term outlook are not the same for every subtype.

Symptoms

Symptoms depend on which muscles and organs are affected. Weakness is usually progressive, meaning it increases over time, but the rate of change can be slow or uneven.

Possible signs include:

Some children reach movement milestones later than expected. Others develop normally at first and then have increasing trouble with activities they previously managed.

Muscle appearance can be misleading. For example, the calves may look enlarged in Duchenne or Becker muscular dystrophy even though the muscle is weak. This enlargement can partly come from fat and connective tissue replacing muscle.

Not everyone has every symptom. A person may have mild weakness for many years, while another subtype may affect movement, breathing, or the heart earlier.

- Frequent falls, difficulty running, or trouble climbing stairs
- Needing the hands to push up from the floor or a chair
- A waddling walk, walking on the toes, or reduced balance
- Weakness around the hips, shoulders, face, hands, or lower legs
- Tight joints or shortened muscles, called contractures
- Muscle stiffness, cramps, or difficulty relaxing a tightened muscle
- Drooping eyelids or difficulty moving the eyes in some types
- Problems swallowing, speaking clearly, coughing, or breathing
- Tiredness that is out of proportion to the activity
- A heartbeat that feels unusually fast, irregular, or forceful

Causes and risk factors

Muscular dystrophies are caused by changes in genes involved in muscle function. These changes may prevent the body from making an important muscle protein or cause it to make a protein that does not work properly.

The inheritance pattern differs by subtype. A gene change may be:

A person can therefore have muscular dystrophy without a known family history. Relatives may carry a gene change without obvious symptoms, or symptoms may not have been recognised in earlier generations.

Biological sex can affect some X-linked types. Duchenne muscular dystrophy mainly affects boys, but girls and women with a dystrophin gene change can sometimes develop muscle or heart problems. Other muscular dystrophies affect people of any sex.

These inherited gene changes are not caused by exercise, diet, parenting, or ordinary daily activities.

- X-linked , as in many cases of Duchenne and Becker muscular dystrophy
- Autosomal dominant , meaning one changed copy can cause the condition
- Autosomal recessive , meaning both gene copies usually need to be changed
- Newly occurring, meaning the change began in that person rather than being inherited

Diagnosis

Diagnosis begins with a medical and family history and an examination of muscle strength, movement, reflexes, joints, and walking. Because many conditions can cause weakness, tests are used to identify the cause and subtype.

A blood test may measure creatine kinase , or CK. This enzyme leaks into the blood when muscle is damaged. A high result suggests muscle injury but does not identify the exact muscular dystrophy.

Genetic testing is often the main way to confirm a subtype. It looks for gene changes known to cause muscular dystrophy. If testing does not provide a clear answer, further analysis or a muscle biopsy may be considered. A biopsy removes a small sample of muscle for laboratory examination.

Electromyography may sometimes be used to study electrical activity in muscle. It can help distinguish a muscle condition from a problem involving nerves, but it does not usually confirm the genetic subtype by itself.

Once muscular dystrophy is suspected or confirmed, other tests may check which organs are affected. These can include:

Genetic counselling can help you understand the test result, how the condition may have been inherited, and what it could mean for relatives.

- An electrocardiogram and heart imaging
- Breathing tests, including measurements while sitting or lying down
- Sleep or overnight oxygen and carbon dioxide monitoring
- Swallowing, nutrition, hearing, or eye assessments
- Blood tests for hormone or metabolic problems in relevant subtypes

Treatment and management

There is currently no cure that reverses all forms of muscular dystrophy. Treatment aims to preserve movement and independence, support breathing and heart function, manage symptoms, and reduce complications.

Care often involves specialists in neurology, rehabilitation, heart care, respiratory care, physiotherapy, occupational therapy, and genetics. The exact team depends on the subtype and your needs.

Physiotherapy may use gentle movement, positioning, stretching, and individually planned activity to maintain comfort and joint movement. Occupational therapy can suggest ways to make school, work, self-care, and household tasks easier.

Braces, walking aids, wheelchairs, standing equipment, or home adaptations may improve safety and independence. Using mobility equipment does not mean that other movement must stop. It can reduce fatigue and help you participate in daily life.

Some medicines can slow muscle decline in particular forms. Corticosteroids like prednisone or hydrocortisone are commonly used in Duchenne muscular dystrophy, while some gene- or mutation-specific treatments are available only for people with certain genetic results. Benefits, monitoring needs, and possible side effects differ, so these treatments require specialist assessment.

Heart problems may be treated before or after symptoms appear. Treatment can include medicines for weakened heart muscle or abnormal rhythms. Regular heart monitoring matters because changes may be present without obvious warning signs.

Breathing support can include assisted coughing, treatment of sleep-related breathing problems, and non-invasive ventilation delivered through a mask. Swallowing and nutrition support may include changes in food texture, feeding strategies, or a feeding tube when eating is no longer safe or sufficient.

Surgery may sometimes be considered for severe contractures, spinal curvature, or other specific problems. Emotional support, education planning, workplace adjustments, and help

with practical needs are also part of care.

Complications

Weak muscles can lead to contractures, reduced mobility, falls, spinal curvature, and difficulty with everyday tasks. Long periods of limited movement may also weaken bones.

If breathing muscles become weak, coughing may not clear mucus effectively. This can increase the effect of chest infections or cause breathing to become too shallow, especially during sleep.

Some muscular dystrophies can cause cardiomyopathy, which is weakness of the heart muscle, or abnormal heart rhythms. These problems may need monitoring even when you feel well.

Swallowing weakness can cause weight loss, dehydration, choking, or food entering the lungs. Myotonic dystrophy can also be associated with cataracts, daytime sleepiness, hormone problems, and heart rhythm changes.

Certain anaesthetic medicines and medical procedures may carry extra risks in some subtypes. Tell surgical and anaesthetic teams about the muscular dystrophy, even before a minor procedure.

Prevention

Inherited muscular dystrophies generally cannot be prevented through lifestyle changes. Exercise, diet, supplements, or avoiding particular activities cannot remove the gene change that causes them.

When a specific genetic change is known, genetic counselling can explain reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilisation. Whether to use these options is a personal decision.

Early diagnosis does not prevent the condition, but it can allow heart, breathing, mobility, and swallowing problems to be monitored and treated sooner.

Living with it and what to expect

The course differs greatly between muscular dystrophy subtypes and between people with the same subtype. Some forms progress slowly over decades. Others cause significant movement,

breathing, or heart problems earlier in life.

Muscle weakness that has already developed usually does not fully recover. However, treatment, equipment, and environmental changes may help you continue activities and conserve energy. Needs may change over time, so equipment and therapy plans often require review.

Regular follow-up may include checks of muscle function, joints, breathing, swallowing, nutrition, bone health, and the heart. The schedule is based on the specific subtype, age, symptoms, and previous results.

Many people attend school, work, form relationships, raise families, and take part in their communities. What this looks like may change as physical needs change. Planning ahead for transport, housing, personal assistance, education, and employment can make transitions easier.

Life expectancy is close to typical for some forms but shortened in others, especially when breathing or heart complications are severe. Modern supportive care has improved outcomes for several subtypes, but no single forecast applies to everyone.

When to get medical help

Call emergency services for:

Seek urgent medical advice for new or quickly worsening breathlessness, a weak cough during a chest infection, repeated choking, difficulty swallowing liquids, fainting, or a new fast or irregular heartbeat. Morning headaches, unusual daytime sleepiness, or disturbed sleep can be signs of shallow breathing overnight and also need prompt assessment.

Arrange routine follow-up if weakness, falls, pain, stiffness, or fatigue are gradually increasing. Also mention skin problems from braces, unwanted treatment effects, weight change, or equipment that no longer meets your needs.

Children with delayed movement skills, frequent falls, or loss of previously gained abilities should be assessed. These signs can have many causes and do not by themselves confirm muscular dystrophy.

- Sudden or severe difficulty breathing
- Blue, grey, or very pale lips or skin
- Choking with an inability to breathe, speak, or cough
- Severe chest pain, collapse, or loss of consciousness

- Sudden marked weakness with difficulty staying awake

Common questions

Is muscular dystrophy the same as multiple sclerosis?

No. Muscular dystrophies are inherited conditions that primarily damage muscles. Multiple sclerosis involves the immune system attacking the protective covering of nerves in the brain and spinal cord.

Can muscular dystrophy start in adults?

Yes. The genetic change is present from birth, but symptoms from some types may not appear until the teenage years or adulthood. Myotonic, facioscapulohumeral, limb-girdle, and oculopharyngeal muscular dystrophies can have adult onset.

Does everyone with muscular dystrophy eventually use a wheelchair?

No. This is strongly influenced by the subtype, severity, age at onset, and available treatment. Some people never need a wheelchair. Others use one for longer distances before they need it for most mobility.

Can girls and women have muscular dystrophy?

Yes. Many types affect all sexes. Duchenne and Becker muscular dystrophy mainly affect boys, but girls and women with a related gene change can sometimes have muscle weakness or heart involvement.

Is muscular dystrophy painful?

Weakness itself may not be painful, but muscle cramps, joint stiffness, contractures, posture changes, and overuse can cause discomfort. New, severe, or persistent pain deserves assessment rather than being assumed to be part of the condition.

Is exercise safe with muscular dystrophy?

Appropriate activity can support fitness, movement, and well-being, but exhausting or high-impact exercise may strain vulnerable muscles. The safest type and intensity depend on the subtype, heart and breathing health, and current strength. A physiotherapist familiar with neuromuscular conditions can help plan activity.

Sources

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