CONDITION

Limb-girdle muscular dystrophies

Limb-girdle muscular dystrophies are inherited conditions that gradually weaken muscles around the hips and shoulders.

WHAT IT IS
Inherited muscle conditions causing hip and shoulder weakness

BODY SYSTEM
Muscles, sometimes heart and breathing

MAIN SYMPTOMS
Hip weakness, shoulder weakness, falls, difficulty climbing stairs

WHAT CAUSES IT
Inherited gene changes affecting muscle function

HOW IT IS DIAGNOSED
Genetic testing; muscle biopsy when results are unclear

MAIN TREATMENT
Rehabilitation, mobility support, and heart or breathing care

CAN IT BE CURED?
No - treatment supports function and manages complications

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble, choking, or fainting

LONG-TERM OUTLOOK
Usually progressive; pace and complications differ by genetic subtype

Overview

Limb-girdle muscular dystrophies, or LGMDs, are a large group of inherited conditions that cause muscle weakness. They mainly affect the muscles around the hips and shoulders, known as the limb girdles.

Weak hip muscles can make it difficult to climb stairs, rise from a chair, run, or keep your balance. Shoulder weakness may make lifting, reaching, and carrying harder. Some subtypes can also affect the heart, breathing muscles, or swallowing.

LGMD is not one single condition. Many genetic subtypes have been identified, and they can behave quite differently. Symptoms may begin in childhood, the teenage years, or adulthood. Some subtypes progress slowly, while others cause earlier or more extensive disability.

Modern subtype names may contain letters and numbers that describe how the condition is inherited and which gene is involved. Older names are still used in some records. A specialist can explain how an older name relates to the current classification.

Finding the specific subtype matters because it helps guide heart and breathing checks, family testing, and discussions about research or subtype-specific treatment.

Symptoms

Muscle weakness is usually similar on both sides of the body, although it may not be perfectly even. Early symptoms often come from weakness around the pelvis and hips.

You may notice:

Some people develop shortened, stiff joints called contractures. These commonly affect the ankles, knees, hips, or elbows. Curvature of the spine, known as scoliosis, can occur when trunk muscles are weak.

Certain subtypes cause enlarged-looking calf muscles. The calves may appear strong even though some muscle tissue has been replaced by fat or scar tissue.

Heart muscle weakness, called cardiomyopathy, and abnormal heart rhythms occur in some forms of LGMD. They may cause breathlessness, palpitations, ankle swelling, dizziness, or fainting. However, heart changes can sometimes be present without obvious symptoms.

Weak breathing muscles may lead to disturbed sleep, morning headaches, daytime sleepiness, a weak cough, or breathlessness when lying flat. Swallowing problems are less common but can occur in particular subtypes.

Sensation is usually normal because LGMD primarily affects muscle rather than sensory nerves. Learning and thinking are also usually unaffected, although there are exceptions among rare genetic subtypes.

- Difficulty running, climbing stairs, or standing from a low seat
- Frequent trips or falls
- A waddling walk or walking on the toes
- Trouble lifting your arms, reaching overhead, or carrying objects
- Shoulder blades that stick out, called scapular winging
- Muscle cramps, aching, or tiredness after activity

Causes and risk factors

LGMDs are caused by changes in genes needed for healthy muscle structure, repair, or function. A disease-causing gene change is also called a pathogenic variant.

Some forms are inherited in an autosomal recessive pattern. This means a person usually inherits one altered copy of the gene from each parent. The parents are often healthy carriers and may not know they carry the variant.

Other forms are autosomal dominant. In these conditions, one altered gene copy can be enough to cause symptoms. An affected parent may pass the variant to a child, although the age of onset and severity can differ within the same family.

A family history increases the chance of an inherited muscle condition, but its absence does not rule one out. Recessive conditions can appear unexpectedly, and a new genetic change can occasionally arise in one person.

LGMD is not caused by exercise, diet, infection, or something a parent did during pregnancy. These factors may affect day-to-day symptoms, but they do not create the underlying genetic condition.

Diagnosis

Diagnosis usually begins with a detailed history and examination. A clinician may assess how you walk, rise from the floor, lift your arms, and use muscles around your hips and shoulders. They will also ask about relatives with muscle weakness, heart conditions, or unexplained mobility problems.

Blood testing commonly includes creatine kinase, or CK. CK is an enzyme that leaks into the blood when muscle fibres are damaged. A high result supports a muscle problem but does not identify the LGMD subtype.

Genetic testing is central to diagnosis. A panel can examine many muscular dystrophy genes at once. Broader testing, such as exome or genome sequencing, may be considered when a panel does not find an answer. Results sometimes include a genetic change whose importance is uncertain, so interpretation by a specialist is important.

Other tests may include:

A muscle biopsy is not always needed when genetic testing gives a clear answer. When results are uncertain, biopsy findings may help distinguish LGMD from inflammatory, metabolic, or other muscle conditions.

Genetic counselling can help you understand the result, its meaning for relatives, and whether

family testing may be useful.

- Electromyography to measure electrical activity in muscles
- Muscle MRI to show patterns of affected and preserved muscle
- Muscle biopsy to examine tissue and muscle proteins
- An electrocardiogram and heart ultrasound
- Breathing tests to measure lung and respiratory muscle function

Treatment and management

There is currently no cure that corrects all forms of LGMD. Management aims to preserve movement and independence, reduce complications, and treat heart, breathing, swallowing, or bone problems when they occur.

A physiotherapist can suggest stretches and suitable strengthening or aerobic activity. Moderate, low-impact movement may help maintain fitness without placing excessive strain on weak muscles. Exercise should be adjusted if it causes marked pain, extreme fatigue, or prolonged loss of function.

Occupational therapists can recommend ways to make daily tasks easier. Helpful equipment may include railings, raised seating, bathroom aids, braces, walking aids, powered wheelchairs, or devices that support reaching and arm movement. Using equipment can conserve energy and reduce falls; it does not mean that rehabilitation has failed.

Contractures may be managed with regular stretching, positioning, braces, or splints. Surgery is sometimes considered when joint tightening or scoliosis causes significant problems.

Heart care depends on the subtype and test results. It may include monitoring, medicines for weakened heart muscle or abnormal rhythms, or an implanted device such as a pacemaker. Because heart changes can be silent, scheduled checks may still be important when you feel well.

Breathing support can include assisted coughing, treatment of sleep-related breathing problems, or non-invasive ventilation delivered through a mask. Swallowing assessment and nutrition support may help if eating becomes tiring or food regularly goes down the wrong way.

Tell surgical and anaesthetic teams that you have, or may have, a muscular dystrophy. Certain anaesthetic plans require extra care in people with muscle conditions.

Research includes gene-based and other subtype-specific therapies. These treatments are not available for every form of LGMD, and participation in a clinical trial does not guarantee benefit.

Complications

Possible complications are influenced by the genetic subtype, severity, and length of time you have had symptoms. They can include:

Regular monitoring is intended to find treatable changes before they cause major symptoms.

- Falls, fractures, and reduced mobility
- Contractures or scoliosis
- Cardiomyopathy or abnormal heart rhythms
- Weak breathing and ineffective coughing
- Chest infections caused by difficulty clearing mucus
- Choking, weight loss, or food entering the airway
- Reduced bone strength, particularly when mobility is limited

Prevention

LGMD cannot currently be prevented in someone who has inherited a disease-causing genetic variant. Lifestyle choices do not remove that variant.

Genetic counselling can explain the chance of passing a specific subtype to a child. If the family variant is known, options may include testing during pregnancy, testing embryos during fertility treatment, using donated eggs or sperm, or choosing not to test. These are personal choices, and counselling should be non-directive.

Falls and some secondary complications may be reduced through suitable mobility aids, home adjustments, stretching, and recommended heart or breathing follow-up. These steps do not prevent LGMD itself.

Living with it and what to expect

Most LGMDs are long-term and progressive, meaning weakness tends to increase over time. The pace is not predictable from the diagnosis alone. The genetic subtype, age when symptoms began, and involvement of the heart or breathing muscles all affect the course.

Changes often develop over years rather than days. You may need to adapt how you manage stairs, work, school, transport, personal care, or exercise. Some people remain able to walk throughout life. Others eventually use a wheelchair for longer distances or all mobility. A

wheelchair can improve independence, access, and energy rather than simply representing loss.

Regular follow-up may include reviews of mobility, joint movement, heart health, breathing, swallowing, nutrition, and bone health. The timing of these checks should reflect the subtype and your previous results.

Fatigue, changing independence, and uncertainty can affect emotional health and relationships. Psychological support, practical advice, workplace or school adjustments, and contact with people who have similar conditions may help.

New symptoms should not automatically be blamed on LGMD. Common illnesses and treatable problems can still occur and deserve their own assessment.

When to get medical help

Arrange routine medical review if you develop new or gradually worsening weakness, repeated falls, difficulty climbing stairs, or trouble lifting your arms. These symptoms have many possible causes and need assessment rather than self-diagnosis.

Contact your usual clinical team promptly if you notice:

Seek emergency help for severe difficulty breathing, blue or grey lips, choking that prevents breathing, sudden collapse, or fainting with chest pain or a racing or irregular heartbeat.

- New palpitations, ankle swelling, or reduced exercise tolerance
- Morning headaches, daytime sleepiness, or breathlessness when lying flat
- A weaker cough or repeated chest infections
- Coughing during meals, food sticking, or unexplained weight loss
- A clear change in walking, falls, pain, or joint stiffness

Common questions

Is LGMD the same as Duchenne muscular dystrophy?

No. Both are inherited muscular dystrophies, but they are caused by different genetic changes and often have different patterns of inheritance and progression. Genetic testing helps distinguish them.

Can I exercise if I have LGMD?

Many people can benefit from carefully planned, moderate activity. Low-impact exercise and

stretching are often used to maintain fitness and joint movement. Exhaustive or high-resistance exercise may overwork vulnerable muscles. A physiotherapist familiar with neuromuscular conditions can help tailor activity safely.

Will I need a wheelchair?

Not everyone does. The likelihood and timing are strongly linked to the genetic subtype and severity. Some people use a wheelchair only for long distances, while others use one regularly to improve safety and conserve energy.

Can LGMD affect my heart or lungs?

Yes, some subtypes can weaken the heart or breathing muscles, while others rarely do. Problems may begin without clear symptoms. Your genetic result helps determine which monitoring is most relevant.

Can my children inherit LGMD?

They may, but the chance depends on whether your subtype is recessive or dominant and on your partner's genetic status. Genetic counselling can provide information based on your confirmed result and family circumstances.

Does LGMD affect life expectancy?

Many people live well into adulthood, but there is no single answer for all subtypes. Life expectancy is most affected when serious heart or breathing complications develop. Monitoring and treatment can help manage these risks.

Sources

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