CONDITION

LGMD

LGMD is a group of inherited conditions that mainly weaken the muscles around the hips and shoulders.

WHAT IT IS
Group of inherited muscle-weakening conditions

BODY SYSTEM
Muscles, sometimes heart and breathing muscles

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

WHAT CAUSES IT
Inherited gene changes affecting muscle function

HOW IT IS DIAGNOSED
Genetic testing, sometimes supported by muscle biopsy

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

CAN IT BE CURED?
No - current treatments manage symptoms

IS IT AN EMERGENCY?
Sometimes - severe breathlessness, fainting, or chest pain

LONG-TERM OUTLOOK
Often progressive; pace and complications differ by genetic type

Overview

LGMD stands for limb-girdle muscular dystrophy. It is the name for a group of inherited conditions that cause progressive muscle weakness. "Limb-girdle" refers to the muscles around the shoulders and hips, where weakness often begins.

LGMD is not one single condition. Many genetic types are known, and they can behave differently. Some begin in childhood and progress quickly. Others first cause symptoms in adulthood and change slowly over many years.

Most types mainly affect voluntary muscles, which are the muscles you use to move. Certain types can also weaken the heart or the muscles used for breathing. Knowing the genetic type can help your care team plan appropriate monitoring.

LGMD names have changed as researchers have learned more. Newer names may include "D" for dominant inheritance or "R" for recessive inheritance, followed by a number and the affected protein. Older records may use labels such as LGMD1 or LGMD2. These systems can refer to the same genetic condition.

Symptoms

Symptoms and age of onset differ considerably between genetic types. Weakness is usually similar on both sides of the body, although it may not be perfectly even.

Common early symptoms include:

Some people develop enlarged-looking calf muscles because muscle tissue has been replaced partly by fat and scar tissue. Others lose muscle bulk. Tightening around joints, known as contractures, can limit movement.

Weak abdominal and back muscles may affect posture. A curved spine, called scoliosis, can develop in some types.

Breathing problems can cause disturbed sleep, morning headaches, daytime sleepiness, a weak cough, or shortness of breath when lying flat. Heart involvement may cause palpitations, fainting, unusual breathlessness, or swelling around the ankles. These problems are more likely in certain genetic types and may occur before obvious symptoms.

Thinking and sensation are usually unaffected, although the exact features depend on the genetic diagnosis.

- Difficulty running, climbing stairs, or rising from a low chair
- Frequent trips or falls
- A waddling walk caused by weakness around the hips
- Difficulty lifting objects or raising the arms overhead
- Shoulder blades that stick out, called scapular winging

Causes and risk factors

LGMD is caused by changes in genes involved in the structure, repair, or normal function of muscle. When one of these genes does not work properly, muscle fibres become damaged over time.

The condition may follow one of two main inheritance patterns:

A family history can increase the chance of LGMD, but its absence does not rule it out. Recessive conditions can appear in a family with no known history. Symptoms can also be mild or unrecognised in other relatives.

LGMD is not caused by exercise, diet, infection, or anything a parent did during pregnancy.

- Autosomal recessive: A person has a changed copy of the gene from each parent. The parents are usually unaffected carriers.
- Autosomal dominant: One changed copy of the gene can cause the condition. It may be inherited from an affected parent or result from a new genetic change.

Diagnosis

Several muscle, nerve, metabolic, and inflammatory conditions can cause similar weakness. Diagnosis usually begins with a medical and family history and an examination of muscle strength, walking, joints, and reflexes.

A creatine kinase, or CK, blood test measures an enzyme released by damaged muscle. CK is often raised in LGMD, sometimes long before weakness is obvious, but it cannot identify the genetic type.

Other tests may include:

Genetic testing is the main way to confirm a specific type. Results are not always clear because testing may find a gene change whose importance is uncertain.

A muscle biopsy removes a small tissue sample for examination. It is now used less often, but it may help when genetic results do not provide an answer or when doctors need to examine particular muscle proteins.

Genetic counselling can help you understand the result, inheritance pattern, and possible meaning for relatives. Sometimes the diagnosis remains "unclassified LGMD" while testing and research continue.

- Genetic testing for known muscle-related gene changes
- Heart tests such as an electrocardiogram and echocardiogram
- Breathing tests that measure lung function
- Muscle MRI to show which muscles are affected
- Electromyography, which measures electrical activity in muscle

Treatment and management

There is currently no cure for inherited LGMD. Care focuses on preserving function, treating complications, and supporting independence. The plan should reflect the genetic type, current abilities, and whether the heart or breathing muscles are involved.

Physiotherapy can help maintain comfortable movement and limit joint tightening. Gentle or moderate aerobic activity may support general fitness when it is adapted to your abilities. Exhausting exercise or activity that causes prolonged muscle pain may be harmful. A physiotherapist familiar with muscular dystrophy can help set safe limits.

Occupational therapy can suggest practical changes for dressing, bathing, work, school, and household tasks. Braces, walking aids, powered wheelchairs, or home adaptations may reduce falls and fatigue. Using mobility equipment does not mean that you have stopped being active; it can conserve energy for activities that matter to you.

Regular heart and breathing checks are important for types known to affect these systems. Treatment may include medicines for heart problems, help with clearing mucus, or non-invasive ventilation. Non-invasive ventilation supports breathing through a mask, often during sleep.

Speech and swallowing specialists can help if chewing, swallowing, or coughing becomes difficult. Dietitians may advise on maintaining nutrition without recommending unnecessary supplements.

Pain, cramps, fatigue, anxiety, and low mood also deserve attention. Treatment may include pacing, physical approaches, psychological support, and appropriate symptom-relieving medicines.

Gene-based and other targeted treatments are being studied for several forms of LGMD. A clinical trial may offer access to an experimental treatment, but benefit is not guaranteed and eligibility is specific to each study.

Complications

Possible complications depend strongly on the genetic type and severity. They can include:

Not everyone develops these problems. Scheduled monitoring can find some complications before they cause noticeable symptoms.

- Loss of walking ability or increasing need for physical assistance
- Falls, injuries, contractures, or scoliosis

- Weak cough and repeated chest infections
- Reduced breathing during sleep
- Cardiomyopathy, meaning weakness of the heart muscle
- Arrhythmias, which are abnormal heart rhythms
- Difficulty swallowing or maintaining adequate nutrition

Prevention

LGMD cannot currently be prevented in someone who has inherited the gene changes that cause it. Exercise, diet, and supplements do not prevent the underlying condition.

Genetic counselling can explain the chance of passing a specific type to children. If the genetic change is known, family-planning options may include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices rather than required steps.

Preventive care after diagnosis focuses on reducing avoidable complications. This may include vaccinations, fall prevention, joint care, and planned heart and breathing monitoring based on the genetic type.

Living with it and what to expect

LGMD usually changes over months or years rather than from day to day. The rate is different across genetic types and between people with the same type. Periods of stable function can occur, but lost muscle strength is not usually recovered.

Some people continue walking throughout life. Others gradually need braces, a walking aid, or a wheelchair. Heart and breathing problems do not always match the amount of limb weakness, so monitoring remains important even when mobility seems stable.

Planning before tasks become difficult can protect independence. This might include workplace or school adjustments, accessible transport, energy conservation, and equipment for bathing or transfers.

Regular follow-up may involve a neuromuscular clinic, physiotherapy, heart checks, and breathing tests. The schedule is usually based on your genetic type and test results. Bring up new falls, reduced stamina, sleep problems, swallowing changes, or increasing difficulty with everyday tasks at routine appointments.

Many people with LGMD study, work, have relationships, travel, and raise families. Support needs may increase over time, but the timing and extent cannot be predicted from the LGMD label alone.

When to get medical help

Arrange a routine appointment if you notice gradual difficulty climbing stairs, rising from chairs, lifting your arms, or keeping up with usual activities. These symptoms have many possible causes and do not necessarily mean LGMD.

If you already have LGMD, contact your care team promptly about increasing falls, new swallowing difficulty, a weaker cough, morning headaches, daytime sleepiness, or shortness of breath when lying down. These may need assessment but are not always emergencies.

Seek urgent medical help for dark urine with severe muscle pain or weakness, especially after strenuous activity or illness. This can be a sign of substantial muscle breakdown.

Call emergency services for:

These symptoms can have causes other than LGMD, but they need immediate assessment.

- Severe or rapidly worsening difficulty breathing
- Blue or grey lips, confusion, or inability to speak because of breathlessness
- Choking with inability to breathe
- Chest pain, fainting, or a sustained rapid or irregular heartbeat

Common questions

Is LGMD the same as muscular dystrophy?

LGMD is one group within the muscular dystrophies. The term muscular dystrophy covers many inherited conditions that damage muscle, including Duchenne, Becker, facioscapulohumeral, myotonic, and limb-girdle types.

Will I need a wheelchair?

Not everyone with LGMD needs one. The likelihood and timing are influenced by the genetic type, age when weakness began, and rate of progression. Some people use a wheelchair only for long distances to reduce fatigue and falls.

Does LGMD affect life expectancy?

Many people have a near-normal lifespan, particularly when heart and breathing muscles are

not significantly affected. Some types can shorten life because of serious heart or respiratory complications. Genetic diagnosis and regular monitoring provide more useful information than the general LGMD label.

Can exercise make LGMD worse?

Appropriate activity is generally encouraged, but very intense or exhausting exercise may damage vulnerable muscles. A suitable plan often includes low-impact aerobic movement, stretching, and carefully selected strengthening. Stop and seek advice if activity causes severe pain, dark urine, or weakness lasting longer than expected.

Can LGMD be passed to my children?

It can be, but the chance depends on whether your type is dominant or recessive and whether your partner carries a change in the same gene. Genetic testing and counselling can clarify the chance for your family.

Why has the name of my LGMD changed?

Researchers have replaced older number-based labels with names that show the inheritance pattern and affected gene or protein. Your underlying condition has not changed. The newer name is intended to make the diagnosis more precise.

Sources

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