Limb-girdle muscular dystrophies (LGMDs) are a group of rare progressive genetic disorders that are characterized by wasting (atrophy) and weakness of the voluntary muscles of the hip and shoulder areas. The various forms of LGMD are caused by mutations in many different genes. These genes provide instructions for making proteins that are involved in muscle maintenance and repair.

It is difficult to determine the prevalence of limb-girdle muscular dystrophy because its features vary and overlap with those of other muscle disorders. Prevalence estimates range from 1 in 14,500 to 1 in 123,000 individuals.

Muscle weakness and atrophy in LGMD are progressive and may spread to affect other muscles of the body.

LGMD subtypes are classified based on inheritance pattern and genetic cause.

Limb-girdle muscular dystrophy type 2 (LGMD2) includes forms of the disorder that have an inheritance pattern called autosomal recessive, in which two flawed copies of the causative gene — one from each parent — are required for the disease to occur.

The age of onset, severity, and progression of symptoms of LGMD subtypes may vary greatly from case to case, even among individuals in the same family. Some individuals may have a mild, slowly progressive form of the disorder; others may have a rapidly progressive form of the disorder that causes severe disability.

There are no cures for any of the LGMDs, but medications and therapy can help manage some symptoms and potentially slow the course of the disease.
The heart can be affected in LGMD, but this doesn't occur as often as it does in some other forms of muscular dystrophy. Heart problems can take two forms — weakness of the heart muscle (cardiomyopathy) and abnormal transmission of signals that regulate the heartbeat (conduction abnormalities or arrhythmias). The heart should be monitored for these complications.

LGMD, like other muscular dystrophies, is primarily a disorder of voluntary muscles. These are the muscles you use to move the limbs, neck, trunk, and other parts of the body that are under voluntary control.

Pain isn't a major symptom of LGMD, although limited mobility sometimes leads to muscle soreness and aching joints.

Respiratory (breathing) function can decline over time, and this, too, should be monitored regularly.

The brain, the intellect, and the senses are unaffected in most subtypes of LGMD.

What should I know about LGMD?

1. Disease progression in each type of LGMD cannot be predicted with certainty, although knowing the underlying genetic mutation can be helpful. Some forms of the disorder progress to loss of walking ability within a few years and cause serious disability, while others progress very slowly over many years and cause minimal disability.
2. LGMD can begin in childhood, adolescence, young adulthood, or even later. Both genders are affected equally.
3. Individuals may first notice a problem when they begin to walk with a “waddling” gait because of weakness of the hip and leg muscles. They may have trouble getting out of chairs, rising from a toilet seat or climbing stairs.
4. Weakness in the shoulder area may make reaching over the head, holding the arms outstretched, or carrying heavy objects difficult. It may become increasingly hard to keep the arms above the head for such activities as combing one’s hair or arranging things on a high shelf. Some people find it harder to type on a computer or other keyboard and may even have trouble feeding themselves.
5. The heart can be affected in LGMD, but this doesn’t occur as often as it does in some other forms of muscular dystrophy. Heart problems can take two forms — weakness of the heart muscle (cardiomyopathy) and abnormal transmission of signals that regulate the heartbeat (conduction abnormalities or arrhythmias). The heart should be monitored for these complications.
6. LGMD, like other muscular dystrophies, is primarily a disorder of voluntary muscles. These are the muscles you use to move the limbs, neck, trunk, and other parts of the body that are under voluntary control.
7. The involuntary muscles, except for the heart (which is a special type of involuntary muscle), aren’t affected in LGMD. Digestion, bowel and bladder functions, and sexual function, which are carried out by involuntary muscles, remain normal.
8. Pain isn’t a major symptom of LGMD, although limited mobility sometimes leads to muscle soreness and aching joints.
9. The brain, the intellect, and the senses are unaffected in most subtypes of LGMD.

How is LGMD treated?

Treatment is aimed at the specific symptoms present in each individual.

Physical therapy may help strengthen large muscle groups, allow greater motion in the joints and maintain mobility.

A pacemaker can be used to stimulate a normal heartbeat in individuals who experience arrhythmia.

Supportive aids such as railings, braces, walkers, or wheelchairs may help prevent falls, alleviate fatigue, and preserve mobility.

Speech therapy can help individuals who experience slurred speech.

Please talk to your medical provider to obtain more information on these treatments.

Occupational therapy can help individuals focus on specific activities and functions, particularly the use of the hands, specific to a person’s job, recreation, or daily living.

Assisted ventilation can help those with breathing problems.
**Cardiac arrhythmia**
Abnormal heartbeat

**Cardiomyopathy**
A condition in which the heart muscle is weakened, making it harder for the heart to pump blood to the body

**Conduction defect**
Irregular electrical control of the heartbeat

**Contracture**
A shortening of muscles or tendons around joints that can limit mobility

**Dysphagia**
Difficulty swallowing

**Dysarthria**
Slurred speech

**Hypertrophy**
Overgrowth of muscle

**Limb girdle**
The shoulder girdle is the bony structure that surrounds the shoulder area, and the pelvic girdle is the bony structure surrounding the hips; collectively these are called the limb girdles

**Muscular dystrophy**
A term that refers to a number of diseases that cause progressive loss of muscle mass, resulting in weakness and, sometimes, loss of mobility

**Mutation**
A flaw in the DNA code

**Myalgia**
Muscle pain

To learn more about LGMD, visit mda.org or contact the MDA National Resource Center at 833-ASK-MDA1 (275-6321).