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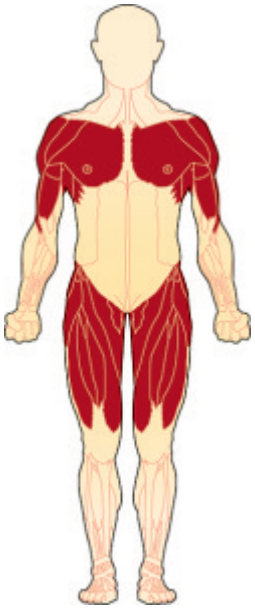
What is Limb Girdle Muscular Dystrophy (LGMD)?

- A term used for a GROUP of rare neuromuscular diseases which are inherited and known to cause muscle weakness & wasting.
- The muscles most affected are those closest to the body (proximal muscles), specifically the muscles of the shoulders, upper arms, pelvic area, and thighs.



- More than 30 forms of LGMD exist; with new forms (sub-types) being discovered each year.
- Some LGMD sub-types have a ‘dominant’ inheritance & some have a ‘recessive’ inheritance pattern.
- In some cases a family history exist & in others, no family history of the disease is known.
- Onset of symptoms can occur in childhood, adolescence, or even adulthood.
- LGMD occurs in all parts of the world & among all ethnic

groups.
- Males & females can inherit LGMD.
- No cure or treatment exists for these progressive diseases although promising research is being carried out which gives hope to many individuals.

Hugging a loved one • rolling over in bed • raising up from a seated position • reaching for an item in the cupboard • walking on a sandy or gravel surface • walking up stairs • wiping a tear from your eye • scratching an itch • bathing • getting dressed • lifting a glass for a sip of a beverage • writing a letter • walking on a windy day…..

For someone with LGMD, many or most of these daily activities can eventually become difficult or even impossible to perform independently, depending upon the stage of the progressive disease.

There are over 30 genetic sub-types (forms) of LGMD and all these rare muscular dystrophies cause weakness of the 'limb-girdle' muscles – the muscles surrounding the shoulders and hips. The severity, age of onset, and features of limb-girdle muscle dystrophy vary among the many sub-types of this condition and may be inconsistent even within the same family. Signs and symptoms may first appear at any age and generally worsen with time, although in some cases they remain mild.

In the early stages of limb-girdle muscular dystrophy, affected individuals may have an unusual walking gait, such as waddling or walking on the balls of their feet, and may also have difficulty running. They may need to use their arms to press themselves up from a squatting position because of their weak thigh muscles. As the condition progresses, people with limb-girdle muscular dystrophy may eventually require wheelchair assistance.

Muscle wasting may cause changes in posture or in the appearance of the shoulder, back, and arm. In particular, weak

shoulder muscles tend to make the shoulder blades (scapulae) “stick out” from the back, a sign known as scapular winging. Affected individuals may also have an abnormally curved lower back (lordosis) or a spine that curves to the side (scoliosis). Some develop joint stiffness (contractures) that can restrict movement in their hips, knees, ankles, or elbows. Overgrowth (hypertrophy) of the calf muscles occurs in some people with limb-girdle muscular dystrophy.

Weakening of the heart muscle (cardiomyopathy) occurs in some forms of limb-girdle muscular dystrophy. Some affected individuals experience mild to severe breathing problems related to the weakness of muscles needed for breathing.

Intelligence is generally unaffected in limb-girdle muscular dystrophy; however, developmental delay and intellectual disability have been reported in rare forms of the disorder.

There are many sub-types of LGMD, in fact more than 30 sub-types have been identified and more are being discovered each year.

- Sub-types with a “1” in the nomenclature (such as LGMD1A, LGMD1B) have an autosomal dominant inheritance pattern. In autosomal dominant inheritance, only one parent needs to transmit the mutated gene for the disorder to be inherited. Even though the matching gene from the other parent is unaffected, the faulty gene dominates. Autosomal dominant disorders typically appear in every generation without skips.
- Sub-types with a “2” in the nomenclature (such as LGMD2A, LGMD2B) have an autosomal recessive inheritance pattern. In autosomal recessive inheritance, both parents must be carriers of the faulty gene for their child to inherit the condition. A carrier harbors the faulty gene, but usually shows no symptoms. When a mutation is recessive, it can exist in a person without showing any symptoms so long as their other copy of the

same gene is healthy. Autosomal recessive disorders typically appear when there is no known family history of the disorder.

Diagnosis & Treatment:

The diagnosis of LGMD is often based on the clinical history, including a possible family history, physical examination and supporting laboratory studies. For some forms of LGMD, it is possible to make a definitive diagnosis by either a blood or saliva test to determine the specific genetic defect responsible for symptoms. A muscle biopsy is often helpful to determine if weakness is caused by muscular dystrophy, an inherited disorder, or by other acquired causes of muscle degeneration such as from inflammation or toxic exposure. It is very important for you to get a genetically confirmed LGMD diagnosis. (If your doctor tells you that you have an LGMD but doesn't genetically confirm it, your diagnosis is considered "incomplete".)

In addition to their primary care physician, individuals diagnosed with LGMD often are under the care of specialists who comprise a multidisciplinary team of doctors and other healthcare professionals. Although no cure or treatment currently exists for any form of LGMD, proper medical management is vital for maintaining function and quality of life.

The actual team of specialist may vary depending upon the form of LGMD (sub-type) and the stage of the illness. The multidisciplinary team of healthcare professionals that may help manage your care could include:

Specialty Doctors:

- **Neurologists** – doctors who specialize in the nervous system and the muscles
- **Cardiologists –** doctors who specializes in heart

function.

- **Pulmonologists** – doctors who specialize in disorders of the lungs and structures associated with the lungs, such as the muscles that control breathing.
- **Orthopedists –** doctors who specialize in the bones and joints and their associated structures, such as the muscles and tendons attached to the bones.
- **Physiatrists (rehabilitation doctors)** – doctors who specialize in what's known as "physical medicine and rehabilitation."
- **Psychiatrists** – doctors who specialize in mental health and emotional issues

Therapists:

- **Occupational therapists**
- **Physical therapists**
- **Respiratory therapists**

Other Healthcare Professionals

- **Dietitians and nutritionists**
- **Genetic counselors**
- **Psychologists**
- **Social workers**
- **Speech-language pathologists**

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