Muscular Dystrophy New Zealand

Limb Girdle Muscular Dystrophy

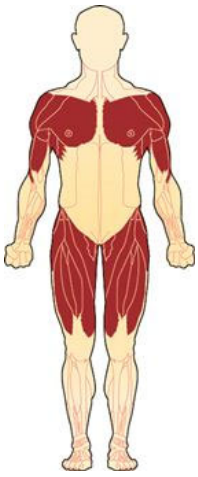
Limb Girdle Muscular Dystrophy (LGMD affects the proximal muscles (those closest to the centre of the body) of the hip and shoulder areas – the pelvic and shoulder girdles, also known as the limb girdles. These muscles weaken and waste away (atrophy), and as a progressive disorder, it may involve other muscles over an individual's lifetime.

Symptoms include difficulties running, walking with a 'waddling' gait, having trouble getting out of chairs, rising from a toilet seat, or climbing stairs. As this weakness progresses, the person may require the use of assistive mobility devices.
Weakness usually occurs first in the legs but then progresses to the arms and shoulder area which can make reaching over the head, holding the arms outstretched, or carrying heavy objects difficult.

Diagnosis has improved markedly in recent years and is usually completed through DNA analysis and a muscle biopsy if needed.
It is recommended that genetic counselling is arranged so that inheritance issues and any risks to other family members can be discussed
https://www.geneticheathservice.org.nz/

The onset of one of the LGMD conditions can occur in childhood, or symptoms may not be apparent until adolescence or adulthood. Males and females are equally affected, however it is difficult to give generalised information on how the condition will progress because different LGMDs progress at different rates, even within the same family. In addition, each different type has some specific features and characteristics, such as age of onset of symptoms and particular muscles involved.

The brain, intellect and senses are unaffected.



Causes of Limb Girdle Muscular Dystrophy

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. When these proteins are not produced properly due to a faulty gene or genes, the cells in the muscles fail to function as they should.

Types of Limb Girdle Muscular Dystrophy

Most types of LGMD are inherited in an autosomal recessive manner and some in an autosomal dominant manner. The old classification of these conditions was 1 for a dominant inheritance and 2 for a recessive inheritance followed by a letter showing order of discovery. However, with more genes being discovered and the alphabet all used up there needed to be a change in classification - which occurred in 2017. Consequently the term LGMD is now followed by the letter D when the disease is inherited in an autosomal dominant manner and by an R if it is inherited in a recessive manner. Subsequently, the disease is given a number in the order in which the disease is discovered over time and finally, the name of the protein that is not (or incorrectly) produced. For example LGMD 2A is now known as LGMD R1 Calpain 3 related.
https://ern-euro-nmd.eu/wp-content/uploads/2019/10/New_names_for_limb_girdle_muscular_dystrofies_Oct2019.pdf

Recessive and Dominant Inheritance

Autosomal Recessive Inheritance
An autosome is any of the chromosomes that is not a sex (X or Y) chromosome. When a disease or genetic trait is recessively inherited two copies of the gene are required (i.e one copy from each parent) for the disease or genetic trait to be expressed.

Autosomal Dominant Inheritance
An autosome is any of the chromosomes that is not a sex (X or Y) chromosome. When a disease or genetic trait is dominantly inherited only one copy of the gene (i.e one copy from one parent) is required for that disease or genetic trait to be expressed.

Progression

Progressively, muscles of the face and distal muscles, such as the lower legs, feet, forearms and hands, may become affected and lead to considerable weakness. Calf muscles may appear unusually large (pseudo hypertrophy) as fatty deposits accumulate and replace lost muscle tissue.

Mobility may become increasingly restricted and 20-30 years from onset, individuals with some types of LGMD may need to use a wheelchair. Wheelchair options can be discussed with an occupational and/or seating therapist.

Fatigue can be common across all the LGMD's and for some of the sub-types muscle and joint pain can also be a feature.

Late symptoms can also include contractures as scar tissue replaces normal elastic tissue. Contractures result in prevention of normal movement in the joint and makes the tissue resistant to stretching. These most commonly occur in the ankles and surgery may be an option to release them. For some individuals, contractures may be an early sign.

Scoliosis, an abnormal curvature of the spine, can also become an issue. Spinal bracing may be required, and in more severe cases spinal fusion surgery. An orthopaedic specialist is beneficial in monitoring the scoliosis.

Weakness of the breathing and heart muscles can affect people with some types of LGMD so regular monitoring by a respiratory physician and/or cardiologist is recommended. Early detection and prompt treatment can be lifesaving. People without a genetic diagnosis should also be carefully monitored as their risk of heart and breathing problems is unknown.

Many researchers have noted that progression of LGMD is often faster and more severe when the onset is earlier, in comparison to individuals who develop LGMD later in adolescence or adulthood.

Additional Management of Limb Girdle Muscular Dystrophy

From an early stage, it is important to undergo regular exercise and stretching programmes, with the help of a physiotherapist, to maintain muscle strength, range of movement and flexibility. Swimming is an excellent option to exercise and mobilize all muscles and joints.

Treatment of Limb Girdle Muscular Dystrophy

Although there are no current treatments yet for these conditions research is continuing and there are now a number of pre-clinical and clinical trials underway.

See more at: https://clinicaltrials.gov/

In New Zealand patients can join Pūnaha Io Neuro-Genetic Registry & BioBank for access into trials and other research opportunities: https://www.mda.org.nz/Our-Research/Registry--Biobank

Support

There are a number of different Facebook groups (some are closed groups) for these conditions, either generic or sub type specific, and they are a good forum to learn from others' experiences or discuss issues.

- NZ Support Group
- Limb Girdle Muscular Dystrophy Beyond Labels and Limitations.
- C3 Community for R1/2A
- National Limb Girdle Muscular Dystrophy
- Women Living with MD

Resources and References:

- MedlinePlus - Health information: https://medlineplus.gov/ency/article/000711.htm
- LGMD Awareness: https://www.lgmd-info.org/about-us/
- LGMD Type 2: https://rarediseases.info.nih.gov/diseases/438/limb-girdle-muscular-dystrophy-type-2d
- Muscular Dystrophy Association: https://www.mda.org/disease/limb-girdle-muscular-dystrophy