Limb-girdle muscular dystrophy

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Limb-girdle muscular dystrophy (LGMD) is a highly variable group of inherited disorders that cause weakness and wasting of the proximal skeletal muscles. These are the muscles closest to the body such as the hip and shoulder areas. These conditions are progressive and worsen over time, often leading to loss of mobility.

The onset of LGMD can occur in childhood or symptoms may not be apparent until adolescence or adulthood. LGMD is often faster and more severe when the onset is earlier, in comparison to individuals who develop LGMD later in adolescence or adulthood.

The main features of LGMD are:

- Weakness in hip and thigh muscles result in an unusual walking gait, and may lead to difficulty climbing stairs and getting up from a seated position.
- Muscle weakness in the shoulder area can make reaching over the head or carrying heavy objects difficult. Some individuals find it increasingly hard to type and may have trouble feeding themselves.
- Postural changes – weak shoulder muscles tend to make the shoulder blades protrude (scapular winging) and some individuals may have spinal curvature.
- 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.
- Mobility may become increasingly restricted and 20-30 years from onset, individuals may require a wheelchair.
- Cardiac problems can arise such as weakness of the heart muscle (cardiomyopathy) or abnormal heartbeat (conduction abnormalities or arrhythmias). The heart must be monitored regularly.
- Respiratory muscles may also be affected resulting in breathing difficulties and should be monitored closely.

Classification of LGMD

The classification of LGMD has changed in the last two years. Previously LGMD1 represented the condition when the mode of inheritance was autosomal dominant (one of the parents has the gene defect and children have a 50 percent chance of inheriting the disease) and LGMD2 represented the condition which was inherited in an autosomal recessive manner (both father and mother need to have the gene defect to pass it onto their children, children have a 25 percent chance of inheriting the disease).

Specific gene defects were assigned a letter of the alphabet in the order that they were discovered. For example, LGMD2A was caused by a defect in the Calpain 3 gene, and was the recessive gene (hence the 2) defect to be discovered first (hence the A).

The new classification has the letter "D" when the disease is inherited in an autosomal dominant manner and 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 the name of the protein that is not or incorrectly produced. For example, LGMD2A is now known as LGMDR1 Calpain 3-related.

The list of classifications is available on MDANZ website.

LGDM community

There are several different Facebook groups for these conditions, either generic or sub type specific, and they are a good forum to learn from others' experiences or discuss issues. These are listed on MDANZ website. R