

FACT SHEET

LIMB GIRDLE MUSCULAR DYSTROPHIES

Limb Girdle Muscular Dystrophies (LGMD) are a group of muscle conditions that cause weakness and wasting of the proximal muscles in the arms and legs.

INCIDENCE: 1 : 14,500 to 1 : 123,000 live births depending on type

AGE OF DIAGNOSIS: infancy to adulthood as onset of symptoms varies depending on type

OCCURRENCE: effects both males and females equally

LIFE EXPECTANCY: infancy to adult. Some people have a normal life expectancy depending on type and age of onset. With early onset the course is generally more rapid

Limb Girdle Muscular Dystrophies (LGMD) have three common features across types; they are genetic; cause progressive proximal muscle weakness; and have a similar pattern of muscles affected. LGMDs typically affect the upper arms, hips, thighs, pelvis, and shoulder area. There are more than 20 different types depending on the gene responsible. There is currently no cure for LGMDs. Research is leading to a greater understanding about LGMDs and this will lead to improved diagnosis and treatments.

Treatments focus on keeping the child/adult healthy and active for as long as possible. Regular physiotherapy aims to prolong function and maintain the muscles used for mobility and breathing. People with LGMD may stop walking in their teens and require a wheelchair for mobility. Others continue walking and have normal strength into late adulthood. Occupational therapy improves the skills of everyday living and focuses on equipment to enhance mobility. The person with LGMD will need an experienced physiotherapist and occupational therapist to ensure their needs are being met. As LGMD progresses, a person's needs will change, and the amount of support and care needs will increase. It is important that they are assessed at regular intervals to plan their future medical, physical, social and psychological supports.

As children become adolescents and adults they will have goals and desires similar to their peers. This may include relationships, work options, study or planning for holidays and so on. Access and inclusion are paramount to ensure independence for education and community involvement. Issues related to puberty, relationships and sexuality may require professional intervention.

The rate of progression varies, however generally the earlier the onset the more severe the symptoms. Children diagnosed with LGMD will often lose the ability to walk in their teenage years. In adulthood, the weakness associated can progress significantly and may affect the person's abilities to function within the home, at school and or at work. People with LGMD experience increased physical difficulties and require support to help with issues such as physical access, transport, social exclusion, financial constraints, health risks, low self-esteem and daily support needs.

Planning ahead for services & equipment is essential, particularly given the complex & varied nature of LGMDs

Given their changing needs, people with LGMD are at risk of becoming depressed and socially isolated, so it is important that a variety of opportunities are encouraged, for example accessible work options, sport and recreation. A person with LGMD may rely on others for a significant level of care support as their condition progresses, and this may affect family dynamics. Enough support needs to be provided to the person with LGMD so that their carer can have a break

As LGMDs progress, medical complications may arise, therefore the following areas should be monitored: tightening of the joints; respiratory care including prevention and treatment of chest infections; sleep studies; non-invasive mechanical respiratory support; heart concerns and annual reviews; speech, swallowing and eating; continence issues; curvature of the spine

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and possible surgery; and psychological issues as they grapple with the loss of independence and increased reliance on others such as family, friends or paid carers.

It is important that service provision is centered around quality of life outcomes & meets the needs of the individual

Genetic testing and counselling should be offered to people with LGMD, parents and family members for future pregnancy planning. Families and individuals affected by chronic conditions benefit most when empowered to have control over their life choices. Through seeking information about what is available, people will be empowered to make the right decisions to achieve their goals. Despite the challenges a person with LGMD faces, there are many opportunities available to support people to live fulfilled and productive lives. Goal setting and positive attitudes contribute to achieving success in many facets of life.

CONSIDERATIONS FOR PLANNING SHOULD INCLUDE BUT ARE NOT LIMITED TO THE FOLLOWING:

- A consistent General Practitioner (GP)
- Specialists for monitoring and medical care planning eg. neurologist, respiratory, heart physicians
- Support coordination due to varied nature of LGMDs and complex care needs
- Physiotherapy eg. hydrotherapy, stretching
- Occupational therapy eg. to increase independence, equipment assessment
- Assistive technology eg. scooter, manual wheelchair, power wheelchair, shower chair, hoist, slings
- Orthotics eg. leg splints and ankle-foot orthoses (AFO's)
- Comfort/sleeping eg. air pressure relieving mattress, electronic bed with high/low options
- Continence management eg. urinary bottles, pans
- Nutrition support eg. dietician, supplements, straws, feeding equipment
- Opportunities to socialise and be away from home eg. MDNSW Camps, accessible travel
- Assistive ventilation devices eg. CPAP or BiPAP machines, cough assist machines
- Home modifications for access to all areas eg. ramps, lifts, bathrooms, bedrooms, kitchen, outdoor areas, pool
- Vehicle modifications/transport eg. steering wheel conversion, taxis to and from school or work and the community
- Adapted sporting and recreation equipment eg. boccia equipment, sports wheelchair
- Support workers eg. showering, toileting, meal preparation, community access
- Professional psychological support eg. counsellor, clinical psychologist
- Future goal planning eg. independent living, study, work choices
- Financial entitlements eg. Centrelink, Medicare, Companion Card

USEFUL RESOURCES

MDNSW www.mdnsw.org.au

MDNSW NDIS Toolkit www.mdnsw.org.au/our-services/support-coordination-your-choice/ndis-faqs

The Loop - Your Neuromuscular Resource Hub www.theloopcommunity.org