CONDITION GUIDE

Limb Girdle Muscular Dystrophy (LGMD)

Limb Girdle Muscular Dystrophy (LGMD) is the common name for a group of muscular dystrophies (a group of diseases that cause your muscles to become progressively weaker) that affect the pelvic (hip) and shoulder areas. The limb girdles are the groups of bones making up the shoulder and pelvic areas, and it is the weakness and atrophy (wasting) of the muscles connected to the limb girdles that has given LGMD its name.

Processing a new diagnosis

A diagnosis of LGMD can be a huge shock for the person with the condition, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your family's life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at https://youtu.be/A9XM71knDVw.

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on Psychology for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on Counselling for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on Social Work for more information.
- A genetic counsellor can help you understand how LGMD is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Visit our page on Genetic Counselling for more information.
- Your state or territory neuromuscular organisation can provide support, advice and information about living with LGMD. Visit our page on state and territory neuromuscular organisations for more information.

- Your GP can talk to you about a mental health plan and how you can use this to help you access the support and services you need. Visit our page on Wellbeing for more information.
- Other people and families living with LGMD have also experienced the emotional rollercoaster that comes with a diagnosis of LGMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with LGMD in The Loop Community on our Forum.

How to talk to your child and others about a new diagnosis

Having a conversation with a loved one about a diagnosis of LGMD, is not easy, in fact it will probably be one of the hardest talks you will have. But, like all difficult conversations it is important and necessary.

There are many reasons why people may not want to have this conversation, mostly centred on wanting to protect and not upset their loved ones and themselves.

But, avoiding the topic is not helpful. People may have noticed differences and changes that have occurred and/or heard the condition being discussed.

So it's crucial that you have a role in your loved one learning about their/your diagnosis. This will enable you to support them as they process the information, allow you to be on hand to answer any questions they may have and together you can learn about the journey ahead. Also, if you are the person with the condition it will provide you with support and someone to talk to.

It will also allow you to:

- Provide answers to questions in an age appropriate way
- Correct any misinformation they have heard or read
- Show them that you have faith in their ability to handle difficult conversations.

There is no right time to start this conversation. Research suggests the earlier you talk to your loved ones about your/their condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don'ts:

- Do be positive but also realistic.
- Do reassure them that no one has done anything wrong and this is not their fault.
- Do tell them they/you will do many wonderful things in their/your life – they/you may just do them differently.
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.

- Do use daily living examples that they can relate to.
- Don’t avoid answering questions and don’t shut the conversation down.

If you are having difficulty starting the conversation, just ask a couple of questions such as ‘How are you feeling today?’ or ‘Do you know why you/they are having difficulty with your/their muscles?’

Remember it’s okay to be upset during the conversation but try to avoid breaking down as this will only cause greater distress. And if speaking with a child, remember children are resilient and they generally handle information of this nature far better than adults.

Understanding LGMD and how it’s diagnosed

About LGMD

Limb girdle muscular dystrophy (LGMD) is the common name for a group of muscular dystrophies (a group of diseases that cause your muscles to become progressively weaker) that affect the pelvic (hip) and shoulder areas. The limb girdles are the groups of bones making up the shoulder and pelvic areas, and it is the weakness and atrophy (wasting) of the muscles connected to the limb girdles that has given LGMD its name.

What causes LGMD?

LGMD is a genetic condition caused by changes to certain genes (the parts of our cells that tell our cells to produce various proteins). A fault in a gene, such as missing or incorrect information, is also called a mutation. The genes associated with LGMD normally make proteins that play a vital role in how muscles function and repair. When one of these genes contains a mutation, the cells cannot produce the proteins needed for healthy muscles. Gradually the muscles become weaker and people will begin to experience the symptoms of LGMD.

Dozens of different mutated genes have been shown to cause the specific subtypes of LGMD1 and LGMD2. In about one in four cases of LGMD, the mutated gene is not yet known and therefore the subtype has not been identified.

How is LGMD diagnosed?

Diagnosis usually begins with a doctor (a neurologist) taking a full medical history, a family history and a clinical assessment. The doctor will ask questions about the age of onset of symptoms, the type of symptoms experienced, the persons progression through the developmental milestones, and if relevant, any issues a person is experiencing with their mobility such as if they are tripping/falling, are they have difficulty climbing stairs or how easy they can get off the floor etc.

Tests that your doctor may use to assist with the diagnosis of LGMD include:

- Muscle enzyme blood test – testing the creatine kinase (CK) level. This test can also be useful for indicating the subtype of LGMD as the degree of CK elevation can help differentiate between different subtypes.
- Electromyogram (EMG) – this test involves inserting small needles into the muscles to measure their electrical activity. An EMG will detect the presence of myotonia in a high portion of people with DM.
- Magnetic Resonance Imaging (MRI) scan - used to identify muscles for biopsy and show doctors the pattern of muscle involvement
- Genetic testing - this type of blood test looks for the specific genetic changes that cause LGMD. Further information about genetic testing can be found on the **Healthdirect website**.
- Muscle biopsy – sometimes doctors will remove a tiny piece of muscle for examination under a microscope to see if there are abnormalities of the muscle fibres.

The first three tests listed above (CK test, EMG and MRI) help narrow down which genes and proteins should be examined in the biopsy and genetic test.

Once a diagnosis is made, it can be possible to also identify the subtype of LGMD through clinical assessment. Different subtypes can be diagnosed by features such as involvement of the cardiac (heart) and respiratory (breathing) systems, presence of muscle hypertrophy (muscle enlargement), contractures (muscle shortening) and scapular winging (shoulder blades sticking out due to shoulder muscle weakness).

LGMD subtypes

There are 2 main types of LGMD, Type 1 and Type 2, however there are currently there are more than 20 different known subtypes. The subtypes of LGMD each differ in their age of onset, their severity, the progression of the condition, the symptoms experienced and how they are inherited.

Once a diagnosis or LGMD is made, further testing will help identify the subtype. The types of LGMD are generally classified by letters and numbers that indicate which faulty gene is known or suspected to be involved.

Ten Most Common LGMD and Their Features

LGMD Type: 1A

Gene affected: myotilinAge of onset: AdultBreathing usually affected: NoHeart usually affected: YesComments:

- Vary Rare
- Mutations in this gene also cause myofibrillar myopathy
- Speech and swallowing difficulties common

LGMD Type: 1B

Gene affected: lamin A/CAge of onset: 5 to 20 yearsBreathing usually affected: YesHeart usually affected: YesComments:

- Mutations in this gene also cause Emery-Dreifuss muscular dystrophy and congenital muscular dystrophy
- Usually slow progression
- Contractures common

LGMD Type: 1C

Gene affected: caveolin 3Age of onset: Any ageBreathing usually affected: NoHeart usually affected: NoComments:

- May have weakness in distal muscles (feet, ankles, calves, hands and wrists) and 'rippling muscle disease'
- Cramps and muscle pain after exercise are common
- Usually slow progression

LGMD Type: 2A

Gene affected: calpain 3Age of onset: Early teens usually, can range from 2 to 50 years of ageBreathing usually affected: NoHeart usually affected: NoComments:

- A common form of LGMD worldwide
- Not usually very rapidly progressive
- Joint contractures may be present

LGMD Type: 2B

Gene affected: dysferlinAge of onset: 15 - 25 usually (variable)Breathing usually affected: NoHeart usually affected: NoComments:

- Usually slow progression
- Muscle pain and swelling in calves can be present

LGMD Type: 2C, 2D, 2E, 2F (sarcoglycanopathies)

Gene affected: gamma, alpha, beta or delta sarcoglycanAge of onset: Usually in childhoodBreathing usually affected: YesHeart usually affected: YesComments:

- Rate of progression of the condition is extremely variable
- Joint contractures and scoliosis may be present

LGMD Type: 2I

Gene affected: FKRPAge of onset: 10 to 20 years (may be earlier or later, with a range from two to 40 years).Breathing usually affected: YesHeart usually affected: YesComments:

- Common in the UK and Northern Europe
- Rate of progression of the condition is extremely variable
- Joint contractures may be present

Source: Muscular Dystrophy Australia – Limb Girdle factsheet.

The genetics of LGMD

LGMD's are classified as Type 1 or Type 2. This classification is based on how they are inherited.

Type 1 or LGMD1

- Less common than Type 2
- Inherited in an autosomal dominant pattern, with a few rare exceptions, meaning it only takes one mutated gene from a parent to pass on the condition
- The single faulty gene is sufficient to over-ride the normal functioning copy of the gene inherited from the other parent
- The chance of passing on an autosomal dominant condition to an offspring is 50% or 1 in 2.

Type 2 or LGMD2

- Makes up about 90% of the LGMDs
- Is inherited in an autosomal recessive pattern, meaning a person with LGMD2 has inherited 2 faulty genes, one from their mother and one from their father
- The parents are known as carriers as they each carry one copy of the faulty gene but typically do not show signs and symptoms of the condition as the other 'normal' copy of the gene is enough to prevent the condition developing
- For carrier parents to have a child with LGMD Type 2, both parents must pass the altered gene on to their child
- If both parents are carriers the likelihood of a child inheriting the condition is 25% or 1 in 4.

Long-term outlook

At this time, progression in each type of LGMD cannot be predicted with certainty. Some forms of LGMD 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.

The progression of the condition and its impact differs between subtypes so knowing the underlying genetic mutation can be helpful in predicting the course of the condition.

Generally, it is thought that the earlier LGMD appears the faster the progression and the greater disability a person will experience. When the condition begins in adolescence or adulthood, it does not tend to be as severe, progression is slower and people are more likely to have a normal life expectancy.

As progression of the condition is unpredictable, early identification and intervention can greatly assist in the management and progression of LGMD.

For more information about living with LGMD, overcoming some of the day-to-day challenges and where to get the right support, visit:

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- Social Work
- Psychology

Understanding and planning for changes

What are the main symptoms of LGMD?

There over 20 different subtypes of LGMD, each of which can cause slightly different symptoms. The key symptom all types of LGMD have in common is initial weakness in the muscles of the shoulder and pelvic (hip) areas. Legs are commonly affected before arms so some of the first signs of LGMD include frequent falls and difficulty climbing steps, running and rising from the floor. When the condition progresses to the arms, people will have difficulty raising their arms above their head to do such things as brushing or washing hair, holding their arms outstretched or carrying heavy objects.

Usually the muscles of the face, the brain, learning and memory, and senses are not impacted.

Other symptoms are dependent on the subtype:

Symptom: Muscle hypertrophy (muscle enlargement) Subtypes:

- LGMD1C
- LGMD2C-F

- LGMD2I

Symptom: Contractures (tightening or shortening of the muscle leading to joint stiffness) Subtypes:

- LGMD1B
- LGMD2A – but milder

Symptom: Spinal stiffness or rigidity Subtypes:

- LGMD1B
- Occasionally in LGMD2A

Symptom: Scoliosis (curvature of the spine) Subtypes:

- LGMD2C-F

Symptom: Cardiac (heart) involvement Subtypes:

- LGMD1B
- 2C-F
- 2I
- Infrequent in LGMD1C, 2A and 2B.

Symptom: Respiratory muscle weakness Subtypes:

- LGMD2C-F
- 2I
- Tends to be insignificant in 2A and 2B.

Source: EFNS Guideline on diagnosis and management of limb girdle muscular dystrophies

Progression and monitoring

Progression in each type of LGMD can’t be predicted with certainty but generally muscles become weaker over time. Some forms of LGMD can progress quickly, with people losing their ability to walk within a few years, while other subtypes progress very slowly over many years and cause minimal disability.

Generally, the earlier the onset of the condition, the more severe the disability. For example, a child diagnosed with LGMD will often lose their ability to walk in their teenage years.

As LGMD progresses, medical complications can arise. Depending on the LGMD subtype, your medical team might monitor the following:

- Joint mobility, for tightness of the joints

- Respiratory function, including prevention and treatment of chest infections, sleep studies, and the need for non-invasive mechanical respiratory support
- Heart concerns
- Speech, swallowing and eating
- Incontinence issues
- Curvature of the spine and the need for possible surgery.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at https://youtu.be/_6oJ9Mc0UO8.

Life stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our Living Life section.

Where to find more information about living with LGMD

Limb Girdle Muscular Dystrophy fact sheet From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

Muscular Dystrophy Australia Limb Girdle Muscular Dystrophy fact sheet From Muscular Dystrophy Australia, June 2018.

International organisations

Muscular Dystrophy Association (US) This website has excellent information about LGMD and neuromuscular conditions. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.

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Support for LGMD

Treatment and management

Currently there is no specific treatment for LGMD however early diagnosis and intervention may assist in managing the condition and slowing its progression. People living with LGMD without a known subtype should be closely monitored as their risk of heart and breathing problems is unknown.

Medical management includes:

Respiratory system

Weakness in the respiratory (breathing) muscles can lead to respiratory complications in some people with LGMD. Early signs of breathing difficulties include frequent chest infections, daytime sleepiness and morning headaches. People experiencing these symptoms may benefit from a non-invasive ventilator device (CPAP) or a cough assist machine (link to equipment). This is particularly important for people with subtypes LGMD2C-F and 2I.

It is recommended that the flu shot be given to people with respiratory muscle weakness to reduce the risk of illness. See the Commonwealth Department of Health website for further information.

Colds, coughs or other respiratory viruses need to be treated promptly by your doctor or medical team. Visit our Respiratory Care page for more information.

Cardiac system

Heart complications, such as weakness of the heart muscle, are generally rare but are often experienced by people with LGMD types 1B, 2C-2F and 2I. People with these types of LGMD should see a cardiologist to monitor heart function. If a problem is detected medication may be used or a pacemaker or defibrillator implanted. Visit our Cardiology page for more information.

Spine

Curvature of the spine, also known as scoliosis, can sometimes occur, particularly for people with subtypes LGMD2C-F or wheelchair users. Correct fitting/seating by an occupational therapist is important.

Management of spinal stiffness or rigidity is particularly important for people with subtypes LGMD1B and sometimes 2A.

Exercise and physical activity

Currently there are no guidelines relating to exercise and the role of a physiotherapist in managing LGMD. General principles suggest that gentle exercise is important for all LGMD subtypes as it helps maintain muscle strength but is also important for general health and wellbeing.

Stretches developed by a physiotherapist can assist with contracture management, as can the use of orthoses (splints). This is particularly important with people with subtypes LGMD 1B and 2A, who may require surgery to release contractures.

As exercise recommendations for LGMD are based on general principles, it is important to seek advice from a doctor of physiotherapist before starting. People living with LGDM should be aware that extreme tiredness, muscle pain and cramps during or after exercise can mean that you have pushed your body too far.

Source: EFNS Guideline on diagnosis and management of limb girdle muscular dystrophies

LGMD Registries

There are a number of international patient registries for people with specific LGMD subtypes to join. See Limb Girdle Patient Registries.

Other helpful support services

The video is available at https://youtu.be/BDaCNsd9R_s.

- Your state or territory neuromuscular organisation can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to equipment, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn

more about what services are available and how to access them here. Visit our pages on Psychology, Counselling, Social Work and Wellbeing for more information.

- Living with LGMD may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on Disability Support Workers for more information.
- Other people and families living with LGMD have also experienced the emotional rollercoaster that comes with a diagnosis of LGMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with LGMD in The Loop Community on our Forum.

How to have better conversations when communicating your needs

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Employers

Educators

Living Life: Education

Support for LGMD

Treatment and management

Currently there is no specific treatment for LGMD however early diagnosis and intervention may assist in managing the condition and slowing its progression. People living with LGMD without a known subtype should be closely monitored as their risk of heart and breathing problems is unknown.

Medical management includes:

Respiratory system

Weakness in the respiratory (breathing) muscles can lead to respiratory complications in some people with LGMD. Early signs of breathing difficulties include frequent chest infections, daytime sleepiness and morning headaches. People experiencing these symptoms may benefit from a non-invasive ventilator device (CPAP) or a cough assist machine (link to equipment). This is particularly important for people with subtypes LGMD2C-F and 2I.

It is recommended that the flu shot be given to people with respiratory muscle weakness to reduce the risk of illness. See the Commonwealth Department of Health website for further information.

Colds, coughs or other respiratory viruses need to be treated promptly by your doctor or medical team. Visit our Respiratory Care page for more information.

Cardiac system

Heart complications, such as weakness of the heart muscle, are generally rare but are often experienced by people with LGMD types 1B, 2C-2F and 2I. People with these types of LGMD should see a cardiologist to monitor heart function. If a problem is detected medication may be used or a pacemaker or defibrillator implanted. Visit our Cardiology page for more information.

Spine

Curvature of the spine, also known as scoliosis, can sometimes occur, particularly for people with subtypes LGMD2C-F or wheelchair users. Correct fitting/seating by an occupational therapist is important.

Management of spinal stiffness or rigidity is particularly important for people with subtypes LGMD1B and sometimes 2A.

Exercise and physical activity

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About LGMD

Limb Girdle Muscular Dystrophy fact sheet From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.**Muscular Dystrophy Australia Limb Girdle Muscular Dystrophy fact sheet**From Muscular Dystrophy Australia, June 2018.

International Organisations

Muscular Dystrophy Association (US) This website has excellent information about LGMD and neuromuscular conditions. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.

TREAT-NMD Information about Limb Girdle Muscular Dystrophy This international network has information about LGMD and research registries.

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