



Limb girdle muscular dystrophy 2B (LGMD2B)

What is LGMD2B (also known as Dysferlinopathy)?

LGMD2B is an autosomal recessive form of limb girdle muscular dystrophy (LGMD). The age of onset of muscle weakness is extremely variable, the most common age being between 20 and 30 years.

What are the causes LGMD2B?

LGMD2B is caused by mutations in the dysferlin gene, which contains instructions to produce a protein important to the muscle fibres. Mutations in the dysferlin gene can also cause another condition, called Miyoshi myopathy which predominantly shows symptoms of distal weakness, especially involving the calf muscles in the lower limbs.

How is LGMD2B diagnosed?

The diagnosis can be suspected by findings on a muscle biopsy or when a doctor experienced in muscular dystrophy examines you. A serum creatine kinase (CK) blood test may also show raised levels (up to 100 times the normal range) which indicate a problem in the muscles. The diagnosis has to be confirmed by identifying a mutation in the dysferlin gene which is done on a DNA sample from a blood test. This is often done following a clue from the muscle biopsy or examination. Because this is a very large gene, testing is very lengthy and results may take several months to become available.

Is there a treatment or a cure?

To date there are no specific treatments for LGMD2B, however careful management of the symptoms of the condition can improve a person's quality of life.

Keeping mobile is important for all people affected with muscular dystrophy. There are no guidelines about the type or intensity of activities however it is recommended that any exercise undertaken is done within your limitations and ensuring you remain comfortable. Extreme tiredness, muscle pain and cramps during or after activities can mean that you have pushed yourself too hard and therefore those activities should be avoided. Swimming is a good activity because it promotes movement of all muscles without increased strain.

Joint contractures (tightening) are not a frequent feature in LGMD2B however they can occur as a consequence of reduced mobility. Regular physiotherapy is therefore recommended. This can be carried out by a physiotherapist or people can be taught to do this by themselves in their own home. These types of exercises can include the stretching of all joints, in particular the ankles.

This factsheet is under review, due for updating later in 2017. If you have any queries, please contact us.

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Foot drop can occur in Miyoshi myopathy and less frequently in LGMD2B. An orthopaedic opinion may be indicated and orthoses (splints) are sometimes worn to help with this problem.

Breathing problems are uncommon in LGMD2B. However with progression of the muscle weakness, people with LGMD2B are at risk of developing mild breathing difficulties. Therefore regular monitoring of respiratory function (forced vital capacity – FVC) is recommended. Sometimes overnight studies are indicated (pulse oximetry).

Regular cardiac assessment is usually not required because to date there is no clear evidence of heart muscle involvement in this condition.

What is the prognosis?

People with LGMD2B often have initial symptoms of weakness and wasting (loss of muscle bulk) in the hip, thigh and shoulder muscles. Wasting of calf muscle is also a predominant feature in the majority of people, and they may complain of difficulties in standing on their tiptoes.

The weakness can be more severe on one side of the body and leg involvement is present before shoulder and arms. This can result in frequent falls, difficulty in running, climbing stairs and rising from the floor. As the condition progresses, people can have problems with walking. The distal muscles (hand and forearm muscles in upper limbs and ankle and calf muscles in the lower limbs) can also be involved and occasionally people affected by LGMD2B show both thigh and foot weakness. It is worth mentioning, however, that these symptoms can also be present in some people at the onset of the condition. When this distal muscle involvement is present, people may have difficulties in walking because of foot drop which causes them to stumble frequently.

Shoulder and arm weakness can lead to difficulties in raising the arms above the head, and in lifting objects. Some people complain of muscle pain and swelling in the legs, especially in the calves, but this is usually transient.

Facial and neck muscles are not usually involved and therefore swallowing problems are unlikely.

Whether heart problems are present in LGMD2B is under debate at the moment. Patients with LGMD2B appear rarely to develop clinically significant heart problems.

People with LGMD2B are at risk of developing respiratory muscle weakness and experience breathing difficulties with the progression of the condition, but this is usually a late complication.

LGMD2B is a variable condition in terms of severity and the weakness is always progressive with time although the rate of progression varies from person to person. The rate of progression is usually slow and most people remain able to walk (ambulant).

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Life expectancy is generally within a normal range because the heart and breathing muscles are usually not affected. In later stages of the condition, breathing difficulties can occur but are usually less severe than in other muscular dystrophies. These symptoms can include poor sleep, nightmares, tiredness or headaches after waking up in the morning, lack of appetite and falling asleep during the day.

Other relevant factsheets from Muscular Dystrophy UK

- ▶ The limb girdle muscular dystrophies (LGMD)
- ▶ Inheritance

We're here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child's muscle-wasting condition, and let you know of progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experience's
- ▶ tell you about – and help you get – the services, equipment and support you're entitled to.

If you would like your GP or other health professional to have more information about the Limb-girdle muscular dystrophies, we have some relevant materials. We've developed an online training module for GPs, as well as one for physiotherapists working with adults with muscle-wasting conditions. Contact our helpline or email us to find out more.

If you have feedback about this factsheet please email info@muscular dystrophyuk.org.



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Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on **0800 652 6352** or **info@muscular dystrophyuk.org** from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.muscular dystrophyuk.org

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