



Limb girdle muscular dystrophy 2L (LGMD2L)

What is LGMD 2L (also known as anoctaminopathy)?

LGMD 2L is an autosomal recessive form of limb girdle muscular dystrophy (LGMD). The age of onset of muscle weakness is variable but most commonly in adulthood, between 20 and 50 years of age.

What causes LGMD 2L?

LGMD 2L is caused by mutations in the anoctamin 5 gene, which contains instructions to produce a protein important to the muscle fibres.

How is it diagnosed?

The diagnosis can be suspected when a doctor experienced in muscular dystrophy examines you. A serum creatine kinase (CK) blood test may also show raised levels (up to 30 to 50 times the normal range) which indicate a problem in the muscles. The diagnosis has to be confirmed by identifying the mutated gene (anoctamin 5 gene) which is done on a DNA sample from a blood test.

Is there a treatment or a cure?

To date there are no specific treatments for LGMD2L, 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 by 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 LGMD2L however they can occur as consequence of reduced mobility, and 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.

Foot drop can occur in LGMD2L. An orthopaedic opinion may be needed and orthoses (splints) are sometimes worn to help with this problem.

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

Version: 3, Published: November 2007, Updated: March 2012, Date of review: November 2013, Authors: The clinical neuromuscular team at the Institute of Genetic Medicine, Newcastle upon Tyne, incorporating the National Specialised Commissioning Team service for the limb girdle muscular dystrophies. Clinical neuromuscular team at Newcastle upon Tyne: Professor K.M.D. Bushby MD FRCP, Professor of Neuromuscular Genetics; Professor V. Straub MD, Professor of Neuromuscular Genetics; Professor H. Lochmuller MD, Professor of Experimental Myology; Dr M. Eagle, Consultant Physiotherapist; Dr M. Guglieri, Senior Research Associate, Honorary Consultant Geneticist; L. Hastings, Neuromuscular Nurse Specialist; A. Sarkozy, Specialty Doctor in Neuromuscular Genetics.





Breathing problems are uncommon in LGMD2L. However with progression of the muscle weakness, people with LGMD2L are at risk of developing mild breathing difficulties. Therefore regular monitoring of respiratory function (forced vital capacity – FVC) is recommended.

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 LGMD2L often have initial symptoms of weakness and wasting (loss of muscle bulk) in the thigh and calf muscles and it can be more severe on one side of the body. Some patients might show hypertrophy (increase of muscle bulk) of calf muscles. Hyperextension of the knees can also occur.

The weakness and wasting of the lower limbs can lead to frequent falls, difficulty in running, climbing stairs and rising from the floor. Some people predominantly show involvement of the distal muscles of the legs only (calf muscle) and they may initially complain of difficulties in standing on their tiptoes. They might also have difficulties in walking because of foot drop which causes them to stumble frequently. As the condition progresses, people can also develop some weakness in their arms, resulting in difficulties with lifting heavy objects.

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

Respiratory and heart problems appear to be rare in LGMD2L.

Females are often less severely affected than males, and might show symptoms only later in life.

The severity and progression of the disease is variable from person to person and even among members of the same family.

LGMD2L is a variable condition in terms of severity. The weakness is always progressive, however the rate of progression is usually slow and most people remain able to walk (ambulant) until late adulthood. Based on our current knowledge about this form of LGMD, life expectancy is generally within a normal range because the heart and breathing muscles are usually not affected.

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

Version: 3, Published: November 2007, Updated: March 2012, Date of review: November 2013, Authors: The clinical neuromuscular team at the Institute of Genetic Medicine, Newcastle upon Tyne, incorporating the National Specialised Commissioning Team service for the limb girdle muscular dystrophies. Clinical neuromuscular team at Newcastle upon Tyne: Professor K.M.D. Bushby MD FRCP, Professor of Neuromuscular Genetics; Professor V. Straub MD, Professor of Neuromuscular Genetics; Professor H. Lochmuller MD, Professor of Experimental Myology; Dr M. Eagle, Consultant Physiotherapist; Dr M. Guglieri, Senior Research Associate, Honorary Consultant Geneticist; L. Hastings, Neuromuscular Nurse Specialist; A. Sarkozy, Specialty Doctor in Neuromuscular Genetics.





Other relevant factsheets from Muscular Dystrophy UK

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



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

Disclaimer

While every reasonable effort is made to ensure that the information in this document is complete, correct and up-to-date, this cannot be guaranteed and Muscular Dystrophy UK shall not be liable whatsoever for any damages incurred as a result of its use. Muscular Dystrophy UK does not necessarily endorse the services provided by the organisations listed in our factsheets.

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

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

Version: 3, Published: November 2007, Updated: March 2012, Date of review: November 2013, Authors: The clinical neuromuscular team at the Institute of Genetic Medicine, Newcastle upon Tyne, incorporating the National Specialised Commissioning Team service for the limb girdle muscular dystrophies. Clinical neuromuscular team at Newcastle upon Tyne: Professor K.M.D. Bushby MD FRCP, Professor of Neuromuscular Genetics; Professor V. Straub MD, Professor of Neuromuscular Genetics; Professor H. Lochmuller MD, Professor of Experimental Myology; Dr M. Eagle, Consultant Physiotherapist; Dr M. Guglieri, Senior Research Associate, Honorary Consultant Geneticist; L. Hastings, Neuromuscular Nurse Specialist; A. Sarkozy, Specialty Doctor in Neuromuscular Genetics.

