



Limb girdle muscular dystrophy 2A (LGMD2A)

What is LGMD2A (also known as Calpainopathy)?

LGMD2A is an autosomal recessive form of limb girdle muscular dystrophy (LGMD). It is one of the most common forms of LGMD. The age of onset of muscle weakness is extremely variable; the most common being between eight and 15 years, although it can range between two and 50 years.

What causes LGMD2A?

LGMD 2A is caused by mutations in the calpain 3 gene, which gives instructions to produce a protein important to the muscle fibres.

How is LGMD2A 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 which indicate a problem in the muscles. The diagnosis has to be confirmed by identifying a mutation in the calpain 3 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.

Is there a treatment or cure?

To date there are no specific treatments for LGMD2A, 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) can occur in LGMD2A and therefore regular physiotherapy is 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, knees and elbows. If ankle contractures impair mobility, referral for an orthopaedic opinion may be indicated. Orthoses (splints) are

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sometimes worn day or night to enhance good positioning of the ankle joints. In the case of severe contractures, minor surgical procedures may be necessary.

With progression of the muscle weakness, people with LGMD2A are at risk of developing 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 there is no involvement of the heart muscle in this condition.

What is the prognosis?

People with LGMD2A often have initial symptoms of weakness and wasting (loss of muscle bulk) in the hip, thigh and shoulder muscles. This weakness is usually even on both sides 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.

Shoulder and arm weakness can lead to difficulties in raising the arms above the head, and shoulder blade winging may be present (scapular winging). Some people complain of muscle pain, especially in the legs. Joint contractures (tightening) may be present and more frequently involve the ankles. Facial and neck muscles are not usually involved and therefore swallowing problems are unlikely.

Heart problems are not reported in this condition. People with LGMD2A are at risk of developing respiratory muscle weakness and experience breathing difficulties with the progression of the condition, but this is usually a very late complication.

LGMD2A 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 course of the condition can be mild and wheelchair use may be required many years after onset.

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.

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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 experiences
- ▶ 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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