Muscular Dystrophy UK
Fighting muscle-wasting conditions

Limb girdle muscular dystrophy 2A (LGMD2A)

Overview

Limb girdle muscular dystrophy 2A (LGMD2A) also known as Calpainopathy 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.

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.

Symptoms

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.

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Causes

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

Diagnosis

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.

Treatment

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 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.

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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@musculardystrophyuk.org.

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