



Limb girdle muscular dystrophy 1C (LGMD1C)

Overview

LGMD1C (also known as caveolinopathy) is an autosomal dominant form of limb girdle muscular dystrophy (LGMD). The age of onset of muscle weakness is variable and ranges from childhood to adulthood.

LGMD1C is a rare disease and not many families affected by this muscular dystrophy have been reported. Because of this, the range of weakness and the progression of the condition of people with caveolin 3 deficiency are not well known.

Life expectancy is generally within a normal range because the heart and breathing muscles are usually not affected.

Symptoms

People with LGMD1C can have initial symptoms of weakness and wasting (loss of muscle bulk) in the hip, thigh and shoulder muscles.

Other people can show predominant distal muscle weakness (hand and forearm muscles in upper limbs and ankle and calf muscles in the lower limbs). This weakness is even on both sides of the body and usually is mild to moderate.

Upper leg weakness 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 in lifting objects.

When distal muscle involvement is present, people may have difficulties in walking because of foot weakness (foot drop) which causes them to stumble frequently. People who have hand weakness may have difficulties in performing simple tasks (for example opening bottles).

Rippling muscle disease is a condition in which people have visible ripples which move over the muscle. These can occur spontaneously or be induced by rapid tapping of the muscles. Many people affected by caveolinopathy, regardless of their symptoms, may show muscle rippling at the onset or later stages of the condition.

It is important to mention that the individual features of muscle weakness can be different from person to person, even within the same family.

Muscle hypertrophy (large muscles), especially calf hypertrophy, is often present in people with LGMD1C. Often people complain of muscle pain and cramps, especially in the legs and after exercise. Facial and neck muscles are not usually involved and therefore swallowing problems are unlikely.

Heart and breathing problems are usually not a feature in this condition. However, heart involvement with dilated cardiomyopathy has (rarely) been reported.

LGMD1C is a variable condition in terms of severity and the weakness, but usually the progression is slow to moderate and people remain able to walk (ambulant)

Causes

LGMD1C is caused by a mutation in the caveolin 3 gene, which gives instructions to produce a protein called caveolin 3 which is important to the muscle fibres.

People with mutations in the caveolin 3 gene can present with a broad spectrum of symptoms, which are classified as limb girdle muscular dystrophy (LGMD1C), distal myopathy, Rippling muscle disease myalgia, cardiomyopathy and hyperCKaemia.

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 is often mildly elevated, but in a few cases, CK elevation may be much more marked. Some people may not have any weakness and show only raised serum CK levels.

The diagnosis has to be confirmed by identifying a mutation in the caveolin 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 LGMD1C, however managing the symptoms of the condition improves 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 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.

Although joint contractures (tightening) are not a frequent feature of caveolinopathy, they can occur as a consequence of reduced mobility. Regular physiotherapy can be useful to maintain good

joint mobility. This can be carried out by a physiotherapist or people can be taught to do this by themselves in their own home.

Problems with breathing are usually not associated with LGMD1C, but many clinics obtain regular breathing assessments (forced vital capacity – FVC) as part of regular follow-up.

Regular cardiac assessment is usually not required because there is no strong evidence of heart muscle involvement in this condition. However, this can be discussed with your consultant on an individual basis.

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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.

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