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.

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.

Other relevant factsheets from the Muscular Dystrophy Campaign

The limb girdle muscular dystrophies (LGMD)

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