Muscular Dystrophy UK

Fighting muscle-wasting conditions

Limb girdle muscular dystrophy 2I (LGMD2I)

Overview

LGMD2I is an autosomal recessive form of limb girdle muscular dystrophy (LGMD). It is one of the most common forms of LGMD, especially in Northern Europe.

The age of onset of muscle weakness is extremely variable, the most common being between 10 and 20 years of age. It can also range between two and 40 years.

Life expectancy and quality of life depend upon the identification and treatment of the associated complications such as heart and breathing problems.

Symptoms

People with LGMD2I 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 weakness can result in frequent falls, toe-walking or in a particular 'waddling gait' (swaying from side to side). This can also cause people to have hyperlordosis (arched back). People can have difficulty in running, climbing stairs and rising from the floor. As the condition progresses, mobility becomes increasingly more difficult.

Shoulder and arm weakness can lead to difficulties in raising the arms above the head, and in lifting objects. Some people may complain of muscle pain and cramps, especially in the legs, even before the onset of muscle weakness.

Calf hypertrophy (large calves) and macroglossia (large tongue) can be present.

People with LGMD2I can develop joint contractures (tightening) and more frequently they involve the ankles. Facial and neck muscles are not usually involved and therefore swallowing problems are unlikely. Unlike congenital muscular dystrophy type 1C, Learning difficulties and eye problems are not features of LGMD2I.

People with LGMD2I are at risk of heart and breathing problems. These problems can occur even when weakness is mild. However, as the condition progresses, heart and breathing involvement tend to increase.

People with heart problems can experience symptoms of breathlessness and tiredness. However, some people can have heart problems even when they do not show symptoms.

Breathing problems are common in LGMD2I and these may occur before losing the ability to walk (loss of ambulation). The first symptoms of breathing involvement can include poor sleep, nightmares, tiredness or headaches after waking up in the morning, lack of appetite and falling asleep during the day. As LGMD2I can involve the diaphragm, the first symptoms may be difficulty in breathing when lying flat.

LGMD2I is a variable condition in terms of severity. The weakness is always progressive with time although the rate of progression varies from person to person. Same people may be only mildly affected, whereas others may show a relatively rapid deterioration of weakness, resulting in loss of independent ambulation in early adulthood.

Causes

LGMD2I is caused by mutations in the Fukutin-related protein gene (FKRP), which gives instructions to produce a protein important to the muscle fibres.

Mutations in the FKRP also cause a form of severe congenital muscular dystrophy (MDC1C).

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 (which measures muscle damage) may also show raised levels which indicate a problem in the muscles.

The diagnosis has to be confirmed by identifying a mutation in the FKRP 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 LGMD2I, 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 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 LGMD2I 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

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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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