



# Limb girdle muscular dystrophy 1B (LGMD1B)

## Overview

**LGMD1B is an autosomal dominant form of limb girdle muscular dystrophy (LGMD). The age of onset of muscle weakness is variable; the most common presentation is before 20 years, however some people may present with symptoms when they are older.**

Life expectancy depends upon the identification and treatment of the associated involvement of the heart and the breathing muscles.

## Symptoms

**People with LGMD1B 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 the shoulders 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).

As the condition progresses, the distal muscles (hand and forearm muscles in upper limbs and ankle and calf muscles in the lower limbs) can also be involved. People may experience difficulty in doing simple tasks owing to hand weakness (for example opening bottles) and in walking because of foot weakness (foot drop) which causes them to stumble frequently.

Some people complain of muscle pain, especially in the legs. Less often, calf hypertrophy (large calves) may be present. As the condition progresses, people may develop joint contractures (tightening) in the arms (elbows) and legs (ankles). Facial and neck muscles are not usually involved. However, some people may show mild facial weakness, with difficulties in inflating their cheeks, whistling and may experience fatigue in chewing.

People with LGMD1B are at risk of heart problems. These heart problems can be mild to severe even when weakness does not have an impact on a person's daily activities. Problems with the heart can begin at the onset of weakness and would tend to increase with time.

The heart involvement in LGMD1B usually consists of rhythm and conduction disturbances and less frequently dilated cardiomyopathy. People with heart problems can experience symptoms of breathlessness, tiredness or palpitation (funny beats). However, some people can have heart problems even when they do not show symptoms.

Less frequently, people with LGMD1B may develop respiratory muscle weakness and experience breathing difficulties with the progression of the condition. Breathing symptoms can include; poor sleep, nightmares, tiredness or headaches after waking up in the morning, lack of appetite and falling asleep during the day.

LGMD1B is quite a variable condition in terms of severity and the weakness, but usually the progression is slow to moderate and people remain on their feet and able to walk (ambulant).

## Causes

**LGMD1B is caused by a mutation in the Lamin A/C gene, which gives instructions to produce a protein important to the muscle fibres.**

Mutations in the Lamin A/C gene also cause autosomal dominant and recessive Emery-Dreifuss muscular dystrophy, a congenital muscular dystrophy, an isolated cardiomyopathy, an autosomal recessive peripheral neuropathy (Charcot-Marie-Tooth disorder type 2B1) and other rare unusual conditions with no major muscle involvement (lipodystrophy, Hutchinson-Gilford Progeria and mandibuloacral dysplasia).

## 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 within the normal range or mildly elevated. Unusually, in a few cases, CK elevation may be much more marked.

The diagnosis has to be confirmed by identifying a mutation in the Lamin A/C 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 LGMD1B, however managing the symptoms of the condition improves a person's quality of life.**

Keeping mobile is important for all people affected with muscular dystrophy. There are no guidelines about the type or intensity of activities, however it is recommended that advice on exercise should be discussed with your consultant because of the associated heart problems.

Joint contractures (tightening) or foot drop can occur in LGMD1B 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. An orthopaedic opinion may be indicated and orthoses (splints) are sometimes worn to enhance good positioning of the ankle joints or to help with foot drop if there is weakness in the feet.

Because of the risk of problems with the heart in LGMD1B, regular heart checks are required and these should include ECG (electrocardiogram) and echocardiogram. Many treatments are available and may include the insertion of a device (defibrillator) which controls the heart rate. This will be discussed with you by a cardiologist.

With progression of the muscle weakness, people with LGMD1B may develop breathing difficulties. Therefore, regular monitoring of respiratory function (forced vital capacity – FVC) is recommended. Sometimes overnight studies are indicated (pulse oximetry).

## **Disclaimer**

While every reasonable effort is made to ensure that the information in this document is complete, correct and up-to-date, this cannot be guaranteed and Muscular Dystrophy UK shall not be liable whatsoever for any damages incurred as a result of its use. MuscularDystrophy UK does not necessarily endorse the services provided by the organisations listed in our factsheets.

If you have feedback about this factsheet or want to request references, please email [info@muscular dystrophyuk.org](mailto:info@muscular dystrophyuk.org).

## **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](mailto:info@muscular dystrophyuk.org).

Version: 03 / Date published: 1 November 2007 / Original author: The clinical neuromuscular team at the Institute of Genetic Medicine, Newcastle upon Tyne, incorporating the National Specialised Commissioning Team service for the limb girdle muscular dystrophies. Clinical neuromuscular team at Newcastle upon Tyne: Professor K.M.D. Bushby MD FRCP, Professor of Neuromuscular Genetics; Professor V. Straub MD, Professor of Neuromuscular Genetics; Professor H. Lochmuller MD, Professor of Experimental Myology; Dr M. Eagle, Consultant Physiotherapist; Dr M. Guglieri, Senior Research Associate, Honorary Consultant Geneticist; L. Hastings, Neuromuscular Nurse Specialist; A. Sarkozy, Specialty Doctor in Neuromuscular Genetics / Updated: 1 March 2012 / Updated by: / Date of review: 1 January 1970