

Subtypes of LGMD2

- ▶ **LGMD2A** is caused by mutations in the calpain 3 gene and is characterised by variable condition onset, progressive proximal weakness and muscle atrophy. Cardiomyopathy has been reported rarely. Respiratory function may decline over time but there is rarely severe respiratory impairment.
- ▶ **LGMD2B** is caused by mutations in the dysferlin gene and is characterised by heterogeneous phenotypes ranging from mild late-onset to severe forms. Cardiac dysfunction has been reported rarely. Respiratory function may decline over time but there is rarely severe respiratory impairment.
- ▶ **LGMD2C/D/E/F** are caused by mutations in the sarcoglycan genes (γ , α , β , δ respectively). They are characterised by onset in childhood with severe disability over time and cardiac and respiratory involvement can often be severe.

Subtypes of LGMD2

- ▶ **LGMD2I** is caused by mutations in the FKRP gene. (Mutations in the same gene can also cause a form of congenital muscular dystrophy.) Onset is usually in late childhood and the phenotype can be mild to severe. Patients can have calf hypertrophy. Dilated cardiomyopathy and respiratory impairment are common. Mild intellectual disability has also been reported in some patients.
- ▶ **LGMD2L** is caused by mutations in the anoctamin 5 gene and is characterised by adult onset with slow progression over years. Cardiac and respiratory function are usually normal; however dilated cardiomyopathy has been reported in some patients.

For consensus care guidelines agreed by doctors and patient groups across the world, visit:
www.treat-nmd.eu/care/sma/care-standards



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Muscular Dystrophy UK
Fighting muscle-wasting conditions

Alert card

▶ Limb girdle muscular dystrophy Types 2

Name _____

Date of birth _____ NHS number _____

If presenting at an emergency department, contact the neurology/neuromuscular team and respiratory team at:

as soon as possible on:

Activate your alert card today to receive your vital care plan:

Email info@musculardystrophyuk.org or call our **Freephone helpline 0800 652 6352**



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Limb girdle muscular dystrophy Types 2

Limb girdle muscular dystrophy Types 2 (LGMD2) are a group of muscular dystrophies that predominantly cause weakness in the shoulder and pelvic girdle and are inherited in an autosomal recessive pattern. The group is further divided into subtypes based on the underlying genetic cause, with a progressive alphabetical letter indicating the chronological order of gene identification.

LGMD2 patients may need to use walking aids, can have difficulties climbing stairs and lose the ability to walk. Creatine kinase (CK) levels can be normal to moderately elevated.

The various subtypes of LGMD2 can differ in terms of condition onset, progression, severity and involvement of other systems. Prognosis and management, therefore, are not uniform across the subtypes of LGMD2. Nonetheless, early identification of complications and risk factors is crucial.

Cardiac

- ▶ Cardiomyopathy and/or dysrhythmias are very common in some subtypes of LGMD2, whereas some don't have cardiac complications.

Respiratory

- ▶ Symptoms of nocturnal hypoventilation may signal the development of significant respiratory muscle weakness and the need for intervention. Non-invasive ventilation (NIV) may be required. If supplemental oxygen is required during a respiratory crisis, this must be carefully controlled and carbon dioxide levels monitored, especially in the context of chronic respiratory failure.
- ▶ Assisted coughing with chest physiotherapy and breath-stacking techniques with an AMBU bag help to clear lower airways secretions. This can also be facilitated by a cough assist device.
- ▶ Immunisations should be kept up-to-date, including the flu and pneumococcal vaccines.

Medication and anaesthetic precautions

- ▶ It is essential that the anaesthetist is aware of the diagnosis of LGMD2 to allow appropriate pre-operative assessment and post-operative monitoring.
- ▶ LGMD2 patients may experience increased sensitivity to sedatives, inhaled anaesthetics and neuromuscular blockade.
- ▶ Local anaesthetics and nitrous oxide are safe (e.g. for minor dental procedures).

Fractures and falls

- ▶ Owing to weakness, contractures and poor balance, patients with LGMD2 are at high risk of frequent falls.
- ▶ If the patient is ambulant before fracture, internal fixation is preferable to casting as it helps to preserve muscle and speeds a return to walking.
- ▶ Orthotics input is often important, especially for ankle weakness.

Fractures and falls (continued)

- ▶ It is advised to check vitamin D levels and bone mineral density on a regular basis, especially following a fall or fracture.

Recommendations and precautions

- ▶ **Swallowing difficulties** are rarely reported in LGMD2 patients, however if present, they should be assessed by a SALT.
- ▶ **Bowel function** is generally normal in LGMD2 patients, however some patients can experience constipation. If this is severe, it may require specialist input to exclude other causes.
- ▶ **Liver** enzymes (AST/ALT/alkaline phosphatase) may be mildly raised on blood tests in up to 50 percent of patients. The clinical setting dictates whether further investigation is indicated.
- ▶ Some subtypes of LGMD2 can have **central nervous system** involvement with intellectual disability and/or epilepsy and, rarely, movement disorders.