Subtypes of LGMD1

- **LGMD1B** is caused by mutations in the lamin A/C gene. The phenotype can vary and can selectively involve only the muscle or skin or be multi-systemic. LGMD1B is characterised by predominant proximal weakness in the lower limbs, contractures and cardiac arrhythmias and dilated cardiomyopathy with risk of sudden death. Respiratory insufficiency occurs as muscle weakness progresses; NIV may be required in more severely affected patients. CK is moderately elevated.

- **LGMD1C** is caused by mutations in the caveolin 3 gene and is characterised by an onset usually in the first decade, a mild-to-moderate proximal muscle weakness; however distal weakness can also occur. Muscle hypertrophy, especially enlarged calves, is a common feature as are cramps and rippling muscle disease. Cardiac and respiratory function are generally not affected; however, dilated cardiomyopathy has been rarely reported.

Medication and anaesthetic precautions

- It is essential that the anaesthetist is aware of the diagnosis of LGMD1 to allow appropriate pre-operative assessment and post-operative monitoring.
- LGMD1 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 LGMD1 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.
- It is advised to check vitamin D levels and bone mineral density on a regular basis, especially following a fall or fracture.

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Cardiac
- Cardiomyopathy and/or dysrhythmias are very common in some subtypes of LGMD1, whereas some other forms don't have cardiac complications.

Respiratory
- Symptoms of nocturnal hypoventilation may signal the development of significant respiratory muscle weakness and 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.

Recommendations and precautions
- Swallowing difficulties are rarely reported in LGMD1 patients, however if present, these should be assessed by a SALT.
- Bowel function is generally normal in LGMD1 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 LGMD1 can have central nervous system involvement with intellectual disability and/or epilepsy and rarely movement disorders.