Speech and swallowing

- Speech and swallowing difficulties are common. Unmanaged difficulties can lead to inhalation of saliva, food, or drink into the lungs. In acute hospital setting, long-term swallowing management strategies should be implemented.
- Recurrent chest infections, weight loss, coughing, choking, and food sticking in the throat should be investigated. Refer to the speech and language therapy team when new symptoms start.
- Individuals may struggle with communication. Hospital teams should consult carers to understand how best to communicate with the individual. If individual needs to be involved in making decisions, refer to speech and language therapy team for assessment and advice.

Mobility and falls

- Falls are common due to muscle weakness and reduced balance. It is important to minimise fall risks in all environments and wear a seatbelt if using a wheelchair.
- Consider fractures if an individual has minor trauma, pain, tenderness, and limited or reduced mobility.
- Low-energy fractures can occur in people with poor mobility and contractures. If able to walk before fracture, internal fixation is preferable to casting as it helps to preserve muscle and speeds a return to walking. Contact the local team for orthotics and physiotherapy input early to maintain ambulation or supported standing capabilities. Local teams should liaise with specialist neuromuscular clinic teams for advice.

Anaesthetic precautions

- If using general anaesthetics or sedatives, a pre-assessment must be completed. Close liaison between surgical, anaesthetic, and respiratory teams is necessary. Longer monitoring post-operation is required to diagnose and treat any complications.
- Local anaesthetics and nitrous oxide are safe for minor dental procedures.

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MUSCULAR DYSTROPHY UK

Alert card

Congenital muscular dystrophies (CMD)

Name..... Date of birth.....

NHS/CHI/H&C number.....

If presenting at A&E, contact the specialist team at:

.....

as soon as possible on: .....

For information and support, contact us on our helpline 0800 652 6352 or email info@musculardystrophyuk.org.

Congenital muscular dystrophies (CMD)

CMD are a group of conditions affecting the muscles from birth or infancy. They may be diagnosed later in childhood. Symptoms and severity vary depending on subtype.

- Infants often have low muscle tone, hypotonia, and contractures in the ankles, hips, knees, and elbows.
- Initial problems may include difficulty holding the head up, delayed motor milestones, feeding or respiratory difficulties, and frequent chest infections.
- Certain subtypes can affect brain function and cause learning difficulties. In some rare forms, symptoms including seizures (LAMA2-related CMD) and involuntary movements and tremors (MICU1-related CMD) can develop later in life.

Respiratory

- Respiratory failure may occur without usual signs of respiratory distress and can be triggered by a chest infection. Signs include morning headaches, fatigue, and reduced appetite.
- If presenting with increased respiratory symptoms, carry out a blood gas test. Supplemental oxygen to achieve SpO2 over 94% may be needed. If CO_2 is raised or individual is deteriorating, early initiation of critical care support is advised. This may include non-invasive ventilation.
- Assess secretion management and consider cough augmentation techniques such as assisted coughing, breath stacking with a LVR bag, and/or cough assist device to clear lower airway secretions.
- General respiratory management includes regular screening and sleep studies to assess for nocturnal hypoventilation. Annual flu vaccination is recommended.

Cardiac

- Some subtypes may affect the heart. Regular Echo and ECG may be recommended to assess heart function. If individual presents with shortness of breath, palpitations, or loss of consciousness, it may indicate severe cardiac issues.
- LV-dysfunction may be mild or non-progressive. Asymptomatic cases should be treated empirically with standard treatments (e.g. ACE-inhibitors + beta blockers). Severe cases may need a pacemaker or defibrillator.
- Individuals who develop atrial fibrillation need treatment with blood thinning medication.
- LMNA-related CMD increases risk of cardiomyopathy. Common complications include supraventricular and ventricular arrhythmias, sudden death, complete heart block, and severe heart failure. Individuals with LMNA-related CMD will commonly need a defibrillator and need specialist cardiac care.

Nutrition and gastrointestinal

- Early identification of weight loss, poor appetite, chewing and swallowing difficulties, and prolonged mealtimes is essential. If not consuming an adequate diet, nutritional supplements, and vitamins must be considered. Refer to specialist or local dietetics service.
- A gastrostomy tube can be inserted to supplement oral food/fluid intake or meet nutritional needs. Medicines and water can be taken via tube. Before insertion, it is essential to have a respiratory, anaesthetic, and cardiac assessment, if needed.
- Carers and/or individuals must complete training on gastrostomy tube care. Individual must be referred to local dietetic and nutrition company nurse teams prior to discharge. Infections should be treated with appropriate antibiotics or topical medication.
- Constipation is common with age. Ensuring adequate fluid/fibre intake is important. Consider medication to manage constipation in the long term.