Speech and Language Therapy/Swallowing

- Swallowing difficulties can be common in CMD.
- Symptoms such as recurrent chest infections, unintentional weight loss, the sensation of food and drink sticking, or feeling the need to clear the throat when eating or drinking, should be investigated more thoroughly.
- Refer to a specialist Speech and Language Therapist for an up-to-date swallowing assessment and/or the nutrition team for consideration of alternative means of hydration/nutrition, such as gastrostomy.

Orthopaedics

Low-energy fractures can occur in children with poor mobility and joint contractures.

- In the limbs, these can appear as 'greenstick' or impacted fractures and can be difficult to see on X-ray.
- A high level of suspicion is required if a child has minor trauma, pain, tenderness and limited, reduced mobility.
- Refer to specialist paediatric orthopaedic services for fracture management.

GI Nutritional issues

- Gastrostomy tube leakage can occur and may need replacement.
- Infection should be treated with appropriate antibiotics or topical preparations.
- If the site is very swollen, the tube may have to be removed to relieve pain.
- If tube is removed/or falls out it is important to keep entry site open using XX and to contact the gastrointestinal specialist as soon as possible.

Muscular Dystrophy UK
0800 652 6352 / info@musculardystrophyuk.org
www.musculardystrophyuk.org

Muscular Dystrophy UK
Fighting muscle-wasting conditions

Alert card

Congenital muscular dystrophy

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**

Health & care information you can trust
The Information Standard

Congenital muscular dystrophy (CMD)

CMD is a neuromuscular condition caused by genetic mutations that lead to a lack of various proteins vital for healthy muscle structure or function.

- Symptoms in small babies include hypotonia (floppiness) and low muscle tone. Contractures (tightness) in the hip, ankle, knee and elbow joints are common.
- In children that do not have contractures, initial problems may be difficulties holding the head, and delays in sitting and walking.
- Some forms of CMD can have associated brain changes on a magnetic resonance imaging (MRI) scan.
- Some children with CMD, and brain changes visible on an MRI scan, may have learning difficulties with or without epileptic seizures.

Respiratory

- Respiratory failure in CMD may present without the usual signs of respiratory distress. Always consider underlying respiratory failure.
- If presenting with respiratory symptoms or oxygen need, measure SpO2 in air and CO2 (transcutaneous, end-tidal or blood gas).
- Titrate oxygen therapy to achieve SpO2 94-98% and monitor CO2.
- If CO2 is raised, consider early initiation of mask ventilation.
- Manage respiratory infections with chest physiotherapy, and consider use of nebulised saline and in-exsufflator (cough assist device).
- For hypersecretion, consider use of glycopyrrolate 40-100 micrograms/kg oral max 2mg six-hourly (use IV solution 200 mcg/ml) or oral atropine drops.
- Collect cough swab or sputum for culture and use broad-spectrum antibiotics.
- Consult early with senior to discuss need for ITU care and escalation of respiratory support.

Cardiac

- The likelihood of heart involvement depends on the underlying CMD mutation and this guides the intensity of cardiac surveillance (i.e. Echo and ECG).
- Cardiomyopathy occurs commonly in MDC1C (fukutin mutation) around age 10 years, progressing to heart failure. Periodic cardiac imaging is recommended from diagnosis.
- LV-dysfunction may be mild or non-progressive (MDC1A; FKRP).
- Even asymptomatic LV-dysfunction should be treated empirically with conventional regimes (e.g. ACE-inhibitors or angiotensin-receptor blockers; +/- beta-blockers; etc).
- The possibility of severe LV-dysfunction should be considered when CMD patients present acutely or for other aspects of their condition.