

Anaesthetics/sedation

- ▶ Some patients can experience an increased sensitivity to sedatives, inhaled anaesthetics and neuromuscular blockade. It is essential that the anaesthetist is aware of the diagnosis of FSHD so that appropriate plans can be made for post-operative monitoring.
- ▶ Patients with compromised respiratory function have a higher anaesthetic risk.
- ▶ Local anaesthetics and nitrous oxide are safe, e.g. for minor dental procedures.

Liver/GI

Liver

- ▶ Serum creatine kinase (CK) and 'liver' enzymes (AST/ALT, but not gamma GT) may be mildly raised owing to the muscle involvement. The clinical setting dictates whether further investigation of the apparent liver dysfunction is indicated.

Gastrointestinal (GI)

- ▶ Constipation is common in FSHD patients with substantially reduced mobility, but may need assessment to exclude other causes.
- ▶ When dysphagia occurs in FSHD, patients are at risk of aspiration pneumonia.

Other possible manifestations

- ▶ Conjunctivitis and ulceration of the cornea can occur owing to limited blinking and inability to properly close the eyes, also when sleeping. The patients should consider using artificial tears and protect their eyes during sleep.
- ▶ Retinal vasculopathy, usually asymptomatic, may affect FSHD patients.
- ▶ High-frequency sensorineural hearing loss is common, not usually symptomatic.
- ▶ Substantial facial muscle weakness may lead to misinterpretation of emotional expression, particularly in those with severe, childhood-onset FSHD.

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**Muscular
Dystrophy UK**
Fighting muscle-wasting conditions



Alert card

Facioscapulohumeral muscular dystrophy (FSHD)

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@muscular dystrophyuk.org or
call our **Freephone helpline 0800 652 6352**

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Facioscapulothumeral muscular dystrophy (FSHD)

FSHD is a muscular dystrophy characterised by progressive muscle weakness affecting the facial, scapular, axial, upper arm and lower leg muscles. Wrist and hand muscles as well as those in the hips may be affected but usually later in the condition. Bulbar muscles can be affected in the more severe cases and at late stages of the condition. Extraocular and respiratory muscles tend to be spared. The severity of the condition varies from patient to patient even in the same family and is partly dependent on the severity of the mutation.

Respiratory

- ▶ Respiratory function is usually normal. In a minority of cases, however, type 2 respiratory failure may occur owing to weak breathing muscles, causing shortness of breath and nocturnal hypoventilation.
- ▶ Patients can be prone to chest infections owing to respiratory failure. Patients reporting dysphagia might also be at risk of aspiration pneumonia.

Respiratory (continued)

- ▶ Immunisations should be kept up-to-date, including the flu and pneumococcal vaccines.
- ▶ If breathing function is impaired and if supplemental oxygen is required during a respiratory crisis it must be carefully controlled (aim for SpO₂ target range of 88-92%) and carbon dioxide levels monitored. Non-invasive ventilation (NIV) may be required.
- ▶ Assisted coughing with chest physiotherapy and breath-stacking techniques with an AMBU bag help to clear lower airways secretions during acute chest infections, or prophylactically when respiratory function is compromised. This can also be facilitated by a cough assist device.

Cardiac

- ▶ Heart function is usually not affected. Potential cardiac symptoms (palpitation, fainting, dizziness and shortness of breath) require appropriate investigations. Coincidental cardiac problems, unrelated to FSHD, are more likely than causally related problems.

Pain

- ▶ Chronic pain, probably secondary mechanical in origin rather than being directly related to the myopathy, is very frequent in FSHD patients. Mechanical cervical and lower back pain is common especially in patients who are developing an axial myopathy lumbar lordosis (often evident as a protuberant abdomen).

Fractures and falls

- ▶ Owing to weakness and poor balance, patients with FSHD are at high risk of frequent falls.
- ▶ Consider checking vitamin D levels and bone mineral density, especially following a fall or fracture.
- ▶ If ambulant, internal fixation is preferable to casting as it helps to preserve muscle by allowing earlier mobilisation.
- ▶ Orthotics input is important, especially for ankle weakness. Orthotics can also be used to support the axial myopathy and periscapular weakness.