Scottish Muscle Network

Facioscapulohumeral Muscular Dystrophy (FSHD) “at a glance” sheet

NOTE
This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined based on all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient’s case notes at the time the relevant decision is taken.

Diagnosis / Genetics

- an autosomal dominant condition
- molecular confirmation essential to diagnosis
- severity may vary within a family. Genetic counselling is advised

Clinical features and outlook

Weakness predominantly affects the face, shoulders upper arms and proximal leg muscles, sometimes with distal involvement too. Childhood onset may occur but is rare and usually more severe in phenotype. Some people may eventually require a wheelchair although many patients do maintain walking ability. With adult-onset disease the life span is expected to be normal.

Associated features and anaesthetic risks

Sensorineural hearing loss is associated with FSHD, especially in patients presenting in infancy. Retinal telangiectasia is a very rare feature in FSHD which may give rise to problems with vision. There is no specific anaesthetic risk - unless there is respiratory muscle weakness.

Bulbar problems

There can be difficulty with swallowing especially when there is severe facial and neck weakness. When the facial weakness is severe, it may interfere with speech and communication. Assessment by SALT may be helpful in some patients to assess swallow function and to ensure this remains safe and to assist with communication aids.

Pain

Is often reported and should be treated along conventional lines.

Cardiac manifestations

An ECG at diagnosis is recommended but clinically significant cardiac disease is not thought to be an association.

Respiratory manifestations

Respiratory muscle weakness may develop in those with more severe disease and is more likely in patients who develop camptocormia or lose independent ambulation. This can be treated by nocturnal non-invasive ventilation. Regular symptom review is advised with referral to appropriate respiratory physician as necessary.

Patients should carry an alert bracelet or similar.

Sources of additional information:

www.nn.nhs.scot/smn/
www.musculardystrophyuk.org
https://neuromuscular.wustl.edu/
www.fshd-registry.org.uk