Facio-Scapulo-Humeral (FSH) Muscular Dystrophy

Standards of care and management in symptomatic adults

Introduction

FSH Muscular Dystrophy is an autosomal dominant disorder with a prevalence of up to 1:20,000. Onset may be at any time in life and some gene carriers may remain asymptomatic into old age. Muscle weakness slowly evolves through life and the management of this and associated features require a co-ordinated approach to long term care. These guidelines and statements are based on a recently published European Neuromuscular Centre (ENMC) evidence based standard and literature review and a consensus within the Scottish Muscle Network. They are applicable to clinically affected adult populations only (over 16 years of age). Some patients may be recognized as gene carriers but show no clinical manifestations (usually identified in the course of pedigree genetic advice). In these patients the nature of any ongoing follow up and management is left to the discretion of the Clinical Geneticist involved to decide on the basis of patient age, wishes and co-morbidity.

Diagnosis

The diagnosis of FSH is established by the identification of a partially deleted D4Z4 repeat array on one chromosome 4. Patients with FSH have 1 - 10 D4Z4 units. The test for this mutation is readily available but interpretation of these reports is complex and requires further investigation in some 5% of cases. It is therefore recommended that
The diagnosis of FSH can be based on the detection of a short fragment at 4q35 on Southern Blot AND a typical clinical phenotype. In atypical cases the clinical features and molecular result need to be discussed with a clinical geneticist because of the possibility of both false positive and false negative results from the standard Southern Blot test. Advice should always be sought for further genetic input addressing the pedigree. The diagnosis of FSH in patients with intermediate alleles on Southern Blotting requires careful clinical and genetic correlation and should be discussed with a clinical geneticist.

Physical therapy

Axial weakness is a major feature of FSH and pain, especially in the low back a major problem affecting up to 77% of patients. There are no specific therapies for this but education regarding physical fitness and a full physical therapy evaluation is recommended for these patients.

All patients with functional limitations should be considered for a rehabilitation medicine referral.

There is support from one study for the role of aerobic training in FSH and this should be considered in patients able to participate.

Aerobic exercise therapy should be considered as part of a neuro-rehabilitation strategy.

There is no evidence to support the role of drug treatment and no specific regimes or drugs are recommended.
No specific drug treatment is recommended.

**Cardiac disease**

There are isolated reports of both atrial tachy-arrythmias and of right bundle branch block\(^\text{vii}\) but this has not yet been reproduced in long term or wider studies.

No routine cardiac surveillance in recommended. Any complaints suggestive of a rapid heart beat should be investigated fully for a alternative mechanism. An ECG at diagnosis is recommended.

**Respiratory disease**

Although it has been stated that respiratory insufficiency requiring overnight ventilatory assistance develops in less than 1%, up to 30% of symptomatic patients have weakness of respiratory muscles \(^\text{viii}\). Pragmatic guidance here is based on ref 1.

Respiratory review with ongoing follow up including FVC should be offered to all non-ambulant patients, those with scoliosis or with additional lung disease. In other patients enquiry should be made at review regarding symptoms of respiratory insufficiency.

All non-ambulant patients should have a formal respiratory physician review prior to elective general anaesthesia.
Swallowing

Recent papers have described dysphagia in severe FSH dystrophy. However, there is no evidence this is a major issue prior to advanced disease and no specific recommendations are made.

Deafness

Sensorineural hearing loss is recognized feature of FSH and may be a major manifestation in paediatric onset cases. In adults however there is debate over frequency (perhaps up to 75%) and severity of deafness. We would agree with the ENMC recommendation that adults diagnosed with FSH muscular dystrophy do not require audiological assessment unless they become symptomatic. Symptoms suggestive of evolving deafness should be sought at review appointments.

Visual symptoms

Retinal vasculopathy is common in FSH and in the paediatric group can affect visual function. Progression in adult life is rare but

All adults symptomatic with FSH should have a formal ophthalmological assessment looking for retinal vascular disease. No further follow up is required unless patients develop visual symptoms.
Scapular fixation

A Cochrane review concluded that scapular fixation improved shoulder function in muscular dystrophy \(^{xvi}\). However the selection criteria and best technique remain to be defined. Some patients may experience a small reduction in FVC following surgery.

All adults should be referred for consideration of the procedure if upper limb function is impaired by failure of scapular fixation. Referrers should identify a surgeon with a declared interest in the procedure.

Patient information

Many of the recommendations above carry the implication of actions to follow certain events. We would therefore recommend that

All adults with FSH are offered written FSH specific information regarding General Anaesthesia, visual and aural symptomatology with a contact number for further information.
References

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