Facio-Scapulo-Humeral (FSH) Muscular Dystrophy

Standards of care and management in children and young people

Introduction

FSH Muscular Dystrophy is an autosomal dominant disorder with a prevalence of up to 1:20,000. It may present at any time in life and can affect individuals differently. Within the paediatric population onset of symptoms is more usually in teenagers, but can present in infancy and early childhood in more severe forms. In contrast some gene carriers may remain asymptomatic into old age. In affected individuals muscle weakness slowly evolves through life and the management of this and associated features require a co-ordinated approach to on-going care. These guidelines and statements are based on a recently published European Neuromuscular Centre (ENMC) evidence based standard and literature review¹ and a consensus within the Scottish Muscle Network. They are applicable to clinically affected infants and children up to age 16 years of age.

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 ¹.
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. All affected children and their parents should be offered referral for genetic counselling. 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.

Physiotherapy

FSH is characterised by proximal weakness but axial weakness can be also a major feature. Pain can be a problem, more commonly in affected adults. With progressive muscle weakness, specific advice on postural management, fatigue management and pain management by a physiotherapist familiar with neuromuscular disorders is appropriate. Advice on physical activities is also an important aspect of the management of the condition.

All children and young people with significant functional impairment should be referred for physiotherapy assessment and advice as appropriate.

Medication

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

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Cardiac care

There are isolated reports of both atrial tachy-arrhythmias and of right bundle branch block but this has not yet been reproduced in long term or wider studies.

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

Respiratory care

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 ii. Pragmatic guidance here is based on ref 1.

Respiratory review with ongoing follow up including FVC should be offered to all non-ambulant children and young people, those with scoliosis or with other known respiratory problems. In other cases enquiry should be made at medical review regarding symptoms of respiratory insufficiency, and referred to respiratory services when clinically indicated.

All non-ambulant children and young people with significant functional impairment should have a formal respiratory paediatric review prior to elective general anaesthesia.

Swallowing

Recent papers have described dysphagia in severe FSH dystrophy iii iv. However there is no evidence this is a major issue prior to loss of ambulation and no specific recommendations are made. Enquiry about difficulties with swallow should be made at routine medical reviews. Very rarely lack of facial expression and feeding difficulties
may be a presenting feature in the severe infantile form of FSH MD and this should be
considered in a differential diagnosis of facial diplegia.

**Deafness**
Sensorineural hearing loss is recognized feature of FSH and may be a major manifestation
in paediatric onset cases." vi. Infants and young children will not complain of hearing loss,
and this may impact on their general development and learning.

Infants and pre-school children with FSH should be referred for audiology screening
and older children and young people should be referred if concerns about hearing
are raised. Symptoms suggestive of evolving hearing difficulties should be sought
at review appointments.

**Visual symptoms**
Retinal vasculopathy is common in FSH and in the paediatric group and can affect visual
function vii. Progression in adult life is rare.

All symptomatic children and young people with FSH should have a formal
ophthalmological assessment looking for retinal vascular disease. Young children
should be seen for review on an annual basis, as they are unable to recognise
visual deterioration. Once a child is school age no further follow up is required
unless visual symptoms develop. Families should be given information about
visual symptoms, with advice to contact GP for ophthalmology referral if
appropriate.

**Patient information**
Many of the recommendations above carry the implication of actions to follow certain
events. We would therefore recommend that
All children and young people with FSH are offered written FSH specific information, including the use of General Anaesthesia, and appropriate support organisations.

References

iv Facioscapulohumeral muscular dystrophy; a radiological and manometric study of the pharynx and esophagus, Stubgen J, Dysphagia, 2008, 23, 341 - 347
vi Facioscapulohumeral muscular dystrophy; hearing loss and other atypical features of patients with large 4q35 deletions, Trevisan C P et al, European Journal of Neurology, 2008, 15, 1353 - 1358