RESOURCES FOR INDIVIDUALS LIVING WITH FSHD

The FSH Society (http://www.fshsociety.org)

The FSH Society is the world’s largest grassroots network serving facioscapulohumeral muscular dystrophy (FSHD) patients, their families, and research advocates. It offers online and in-person forums to exchange FSHD information around the world, sharing expert guidance, patient experiences, and recommendations that can help other patients and families. It also reports on scientific and medical advances in the field, including clinical trials. All information on the site is reviewed by established experts. The FSH Society’s website offers downloadable resources, including the following:

- FSH Watch newsletters
- About FSHD patient brochure
- Physical Therapy brochure
- Living with FSHD Series, FSHD: A Guide for Schools
- A Guide for Family and Friends
- Health Tips for FSHD patients

FSH Society Yahoo Group Forum (https://groups.yahoo.com/neo/groups/fshsociety)

This Yahoo Group Message board enables patients to raise questions, provide one another with guidance, form friendships, and discuss options for managing health issues and living with FSHD. It is an active message board that delivers useful information. The message board has search capabilities for easy access to past posts.

The FSH Society (https://www.facebook.com/FSHSociety)

This FSH Society Facebook page posts news and events, and enables message exchange on a wide range of FSHD topics.

Private Facebook groups

There are a number of private, moderated Facebook groups for individuals affected by FSHD (FSH Friends), young people (Teens/Young Adults with FSHD), women (Women with FSHD), and parents of children affected by FSHD. Messages posted in these private groups cannot be seen by non-members. Membership is by invitation, which can be easily arranged by emailing info@fshsociety.org.

These resources are extremely valuable and can help individuals deal with different aspects of FSHD.

In addition to the Internet resources, the FSH Society has a service to connect individuals with a Peer-to-Peer Team member. This service spans the U.S. and many other parts of the world. The Society can match patients to peers by various criteria, including age, gender, shared interests, occupation, and geographic area.

While this devastating disease has a real impact on patients and families, it can be immensely comforting for them to know that they are not alone. These resources give individuals opportunities to have personal conversations with others and receive encouragement and support to help guide their future.

The FSH Society thanks Bill Maclean for helping to create this resource page. This Clinician Guideline is distributed by the FSH Society with permission of the American Academy of Neurology.

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This is a summary of the American Academy of Neurology (AAN) guideline on the evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy (FSHD).

Please refer to the full guideline at AAN.com/guidelines for more information, including the complete clinical context and definitions of the classifications of evidence and recommendations.

Diagnosis of FSHD

Clinical Context

When clinical presentation of FSHD is typical and the inheritance pattern is consistent with autosomal dominant inheritance, clinical diagnosis is usually straightforward. If, in such circumstances, the diagnosis is genetically confirmed in a first-degree relative, genetic testing is not necessary for each affected individual. However, atypical presentations are not uncommon. In the setting of atypical or sporadic cases, genetic confirmation is important for genetic counseling, especially with the recent discovery of two genetically distinct forms of FSHD (PRIN).

In the most common FSHD type, FSHD type 1 (FSHD1), disease results from contraction of a DNA repeat sequence, termed D4Z4 repeat, on one copy of 4q35 from >10 repeats to 1–10 repeats. In addition, the contraction must occur in the presence of one particular (A variant) of two (A/B) sequence variants distal to the repeats (PRIN). Available molecular testing for FSHD1, which measures only the presence of a repeat contraction on initial testing, is highly sensitive and specific (EVID). In studies that utilized strict diagnostic criteria for FSHD1, determining whether a contraction occurs on an A variant genetic background does not appear to improve diagnostic specificity (EVID). However, in clinical practice, strict clinical diagnostic criteria might not be adhered to, increasing the chances of a false-positive result (INFER). In consequence, determining that a D4Z4 contraction is occurring on an A variant is warranted when the clinical presentation is atypical for FSHD. At present, commercial genetic testing in FSHD is limited to FSHD1 testing.

Level B

Clinicians should obtain genetic confirmation of FSHD1 in patients with atypical presentations and no first-degree relatives with genetic confirmation of the disease.

Predictors of Severity in FSHD

Clinical Context

Factors that predict disease severity in FSHD are important for counseling patients and for screening for and managing potential complications (PRIN). The D4Z4 deletion size appears to be somewhat predictive of the overall rate of disease progression (EVID). D4Z4 deletion size should be used cautiously for predicting disease progression rate in any particular individual due to other sources of variation affecting disease severity, including intramuscular factors (INFER). Clinical experience suggests that patients with severe childhood-onset disease almost invariably have very large deletions (i.e., contracted D4Z4 allele of 10–20 kb or 1–4 repeats), suggesting a much more robust correlation between disease severity and large deletions (EVID).

Level B

Large D4Z4 deletion sizes (contracted D4Z4 allele of 10–20 kb or 1–4 repeats) should alert the clinician that the patient is more likely to develop more significant disability and at an earlier age. Patients with large deletions are also more likely to develop symptomatic extramuscular manifestations.

Monitoring for Complications of FSHD

Pulmonary Complications

Clinical Context

Our systematic review revealed that some patients with FSHD develop respiratory muscle weakness that can result in respiratory failure and need for mechanical ventilator assistance (i.e., nocturnal bilevel positive airway pressure), although this complication is uncommon (EVID). Patients with chronic respiratory failure from neuromuscular-related weakness often do not have classic symptoms of ventilatory failure (i.e., overt dyspnea). Impending respiratory failure, therefore, may begin with respiratory insufficiency mainly during sleep, resulting in excessive daytime somnolence or nonrestorative sleep. Respiratory insufficiency in patients with FSHD, therefore, may be evident only through pulmonary function testing (PRIN). Respiratory failure constitutes a major source of morbidity in patients with most muscular dystrophy types and can severely disrupt sleeping, daily activities, and quality of life (QOL) (PRIN).

Early intervention with noninvasive mechanical ventilation leads to improved survival and QOL (RELA).

Level B

Clinicians should refer patients with clinical findings of impending respiratory insufficiency to pulmonology services for evaluation and potential intervention with nocturnal bilevel positive airway pressure and other means of respiratory support.
Treatment of FSHD

Pharmacologic Interventions

Clinical Context

As of this writing, no evidence exists for any effective pharmacologic interventions that improve strength or slow disease progression in FSHD. Randomized, controlled trials of albuterol were negative (EVID). Uncontrolled, open-label trials of corticosteroid and diltiazem showed no benefit. A controlled early phase II study of MYO-D02, a myostatin inhibitor, also failed to show benefit.

Level B

In patients with FSHD, clinicians should not prescribe albuterol, corticosteroid, or diltiazem for improving strength.

Surgical Scapular Fixation

Clinical Context

In patients with FSHD, limited shoulder range of motion due to periscapular muscle weakness is a major source of functional limitation (PRIN). Moreover, in many patients, bedside manual scapular fixation can result in significant improvement in shoulder range of motion (PRIN). Postoperative complications are infrequent but include hemato- or pneumothorax, pain, infection, non-union, and reduced lung capacity. Scapular fixation appears to be generally safe and may be effective for improving shoulder range of motion (EVID).

Level C

Surgical scapular fixation might be offered cautiously to selected patients after careful consideration of the overall muscle impairment in the involved arm, assessment of potential gain in range of motion by manual fixation of the scapula, the patient’s rate of disease progression, and the potential adverse consequences of surgery and prolonged posturgical bracing.

Aerobic Exercise

Clinical Context

Aerobic exercise in FSHD appears to be safe and potentially beneficial (EVID), as has been shown in many other muscle diseases (REL). Aerobic fitness is important for overall health (PRIN). To minimize injury from falls or overuse, the type of aerobic exercise should be tailored to the patient’s initial distribution of weakness. For example, a stationary bicycle rather than a treadmill should be recommended for patients with leg weakness (PRIN). Although clear data on aerobic exercise and motor impairment are lacking, the evidence shows that regular aerobic exercise may be safe and perhaps beneficial for improving aerobic fitness and reducing the risk of muscle fatigue (EVID).

Level C

Clinicians should screen all young children with FSHD at diagnosis and yearly thereafter until these children start school, as hearing loss may be present at diagnosis and can be progressive.

Cardiac Abnormalities

Clinical Context

Our systematic review revealed very little evidence for structural cardiac abnormalities in FSHD. Also, data are insufficient to suggest that patients with FSHD are susceptible to cardiac arrhythmias (EVID). Routine electrocardiographic/echocardiographic testing is therefore unnecessary in patients with FSHD who are asymptomatic (INFER).

Level C

Patients with FSHD should be referred for cardiac evaluation if they develop overt signs or symptoms of cardiac disease (e.g., shortness of breath, chest pain, palpitations). However, routine cardiac screening is not essential in the absence of cardiac signs or symptoms.

Retinal Vascular Disease

Clinical Context

Our systematic review suggests that symptomatic retinal vascular disease in the form of an exudative retinopathy (Coats disease) is very rare in FSHD but tends to affect patients with large deletions almost exclusively (EVID). Untreated exudative retinopathy can lead to significant visual loss, which may be prevented by early intervention (INFER).

Level B

Clinicians should refer patients with FSHD and large deletions (contracted D4Z4 allele of 10–20 kb) to an experienced ophthalmologist (e.g., retina specialist) for dilated indirect ophthalmoscopy (Level B). The presence and severity of retinal vascular disease at initial screening should be used to determine the frequency of subsequent monitoring.

Hearing Loss

Clinical Context

Our systematic review shows that the available studies fail to capture the prevalence and clinical relevance of hearing loss in FSHD (EVID). In clinical practice, most patients with FSHD and hearing loss requiring the use of a hearing aid have childhood-onset FSHD with large D4Z4 deletions. Two recent studies support this clinical impression (EVID). Moreover, one of the studies suggests that hearing loss is progressive in some patients. Adults and older children are cognizant of the hearing loss onset, and therefore intervention can occur early when required. However, failure to detect hearing loss in infants and younger children may significantly delay or impair language development (PRIN).

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Pain

Clinical Context

Pain is a common complaint in FSHD and appears to be mostly musculoskeletal in origin (EVID). Pain compounding muscle weakness can have a significant impact on QOL (INFER). Physical therapists often can provide insight into the mechanism of pain in patients with weakness (PRIN). Nonsteroidal anti-inflammatory medications are useful for acute pain, and antidepressants or antiepileptics for chronic musculoskeletal pain (PRIN).

Level B

Pain patients with FSHD who do not get regular pulmonary function testing should be tested prior to surgical procedures requiring general anesthesia, as such testing may uncover asymptomatic respiratory compromise.

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