

Evidence-Based Guidelines and Beyond

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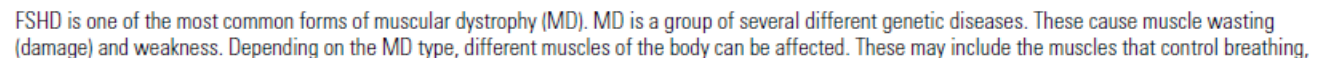
FSHD Connect Meeting

Las Vegas, June 9, 2018

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Best available evidence-based guideline for the management of FSHD



Evidence-based guideline summary:

☐ FSHD Genetic testing

- ☐ Is it accurate and reliable?

☐ Risk factors for disease severity

- ☐ Are there measurable factors that predict severity?

☐ Complications

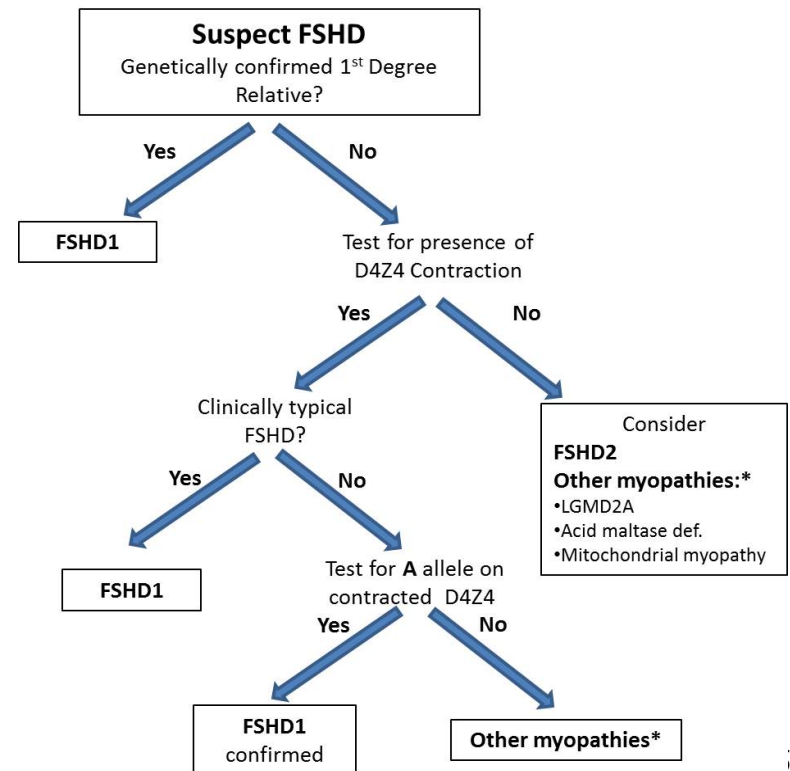
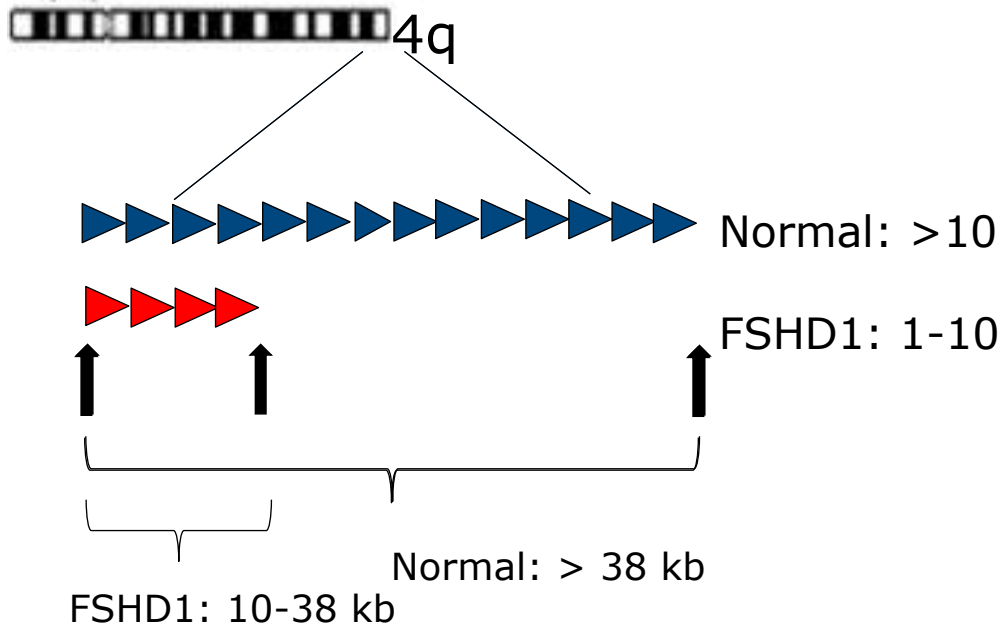
- ☐ What non-muscular complications occur in FSHD and who is most susceptible?

- ☐ Approach to surveillance and prevention/mitigation

☐ Treatment

Genetic testing

Diagnosis		
Clinicians caring for people who may have FSHD type 1	Order genetic testing to confirm FSHD type 1 diagnosis in people with unusual patterns of signs and symptoms and no first-degree relatives with the disease	Moderate



Factors that Effect Disease Severity

Factors That Can Predict Disease Severity	
Clinicians caring for people with FSHD	Look for certain information from genetic test results (large D4Z4 deletion sizes) that may point to more significant disability at an earlier age

- ❑ 1-3 Repeats: 10-18 kb
 - ❑ Most severe, earlier onset, infantile onset
- ❑ 4-7 Repeats: 20-31 kb
 - ❑ Most common, very variable in overall severity
- ❑ 8-10 Repeats: 33-38 kb
 - ❑ Later onset and more likely to have no symptoms

Non-muscular Complications

Monitoring for Complications: Breathing (Pulmonary)

Clinicians caring for people with FSHD	Order baseline pulmonary (breathing) function testing for all patients with FSHD. Monitor regularly if patient has abnormal test results or any combination of severe weakness near the lungs or chest, kyphoscoliosis, wheelchair dependence, or related conditions that affect breathing
Clinicians caring for people with FSHD and 1) poor results from pulmonary function tests or 2) symptoms of extreme daytime tiredness or poor sleep	Refer patients for pulmonary or sleep medicine consultation for possible nighttime sleep monitoring or nighttime breathing machine in order to improve quality of life
Clinicians caring for patients with FSHD who do not get regular pulmonary function testing	Order testing before any surgery requiring general anesthesia (such testing may uncover hidden breathing problems)

Monitoring for Complications: Cardiac (Heart) Abnormalities

Clinicians caring for patients with FSHD	Refer for cardiac (heart) evaluation if the patient has clear signs or symptoms of heart disease (shortness of breath, chest pain, palpitations). Routine screening is not needed in patients with no clear signs or symptoms
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Non-muscular Complications

Monitoring for Complications: Retinal Vascular Disease (Eye Disease)	
Clinicians caring for patients with FSHD and certain genetic signs (large deletions)	Refer the patient to an experienced ophthalmologist (eye doctor) for dilated indirect ophthalmoscopy (an eye test to examine the retina). Eye doctor should use presence of disease on first screening in order to determine how often to follow up with the patient
Monitoring for Complications: Hearing Loss	
Clinicians caring for young children with FSHD	Screen at diagnosis and yearly afterward until the children start school (hearing loss not always present at diagnosis and can be progressive)
Monitoring for Complications: Pain	
Clinicians caring for patients with FSHD	Ask regularly about pain. May refer for physical therapy as a helpful first step. If pain continues, may prescribe nonsteroidal anti-inflammatory drugs for acute (short-term) pain and antidepressants and epilepsy drugs for chronic (long-term) pain

Treatment

Treatment: Pharmaceutical Drugs	
Clinicians caring for patients with FSHD	Should not prescribe albuterol, corticosteroid, or diltiazem for improving strength
Treatment: Scapular Surgical Fixation (Surgery to Lock Shoulder Blade)	
Clinicians caring for patients with FSHD	Might offer surgical scapular fixation (surgery to lock the shoulder blade) to certain patients after careful consideration of arm muscle problems, potential gain in range of motion, rate of disease progression, and potential for poor outcomes from surgery and long periods of bracing
Treatment: Aerobic Exercise	
Clinicians caring for patients with FSHD	Might encourage patients to do low-intensity aerobic exercise program (program might be developed with help of physical therapist). Might use guidelines for physical activities for people with disabilities from US Department of Health and Human Services ¹
Clinicians caring for patients with FSHD who are interested in strength training	May refer patients to physical therapists to develop safe exercise program using appropriate weights and resistance and is tailored to the patient's abilities

Scapular Fixation: Choosing the Right Patient



Scapular Fixation

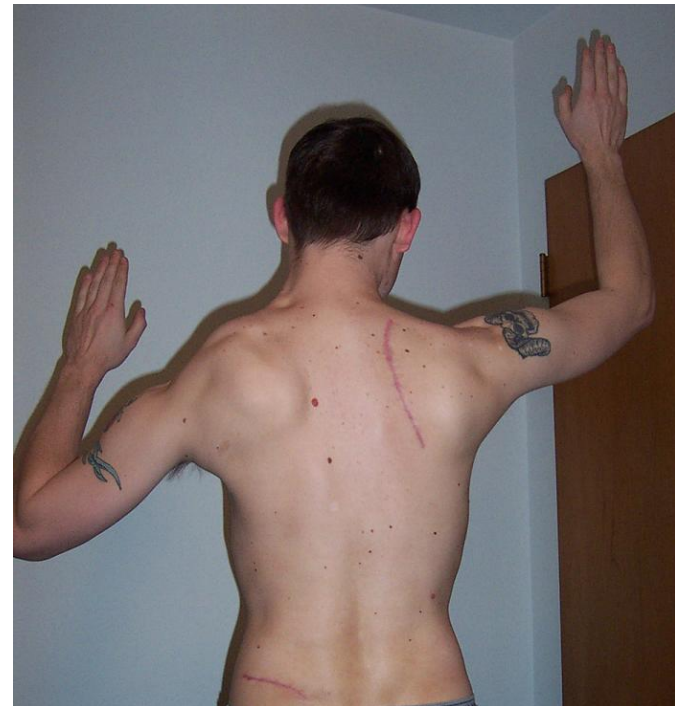
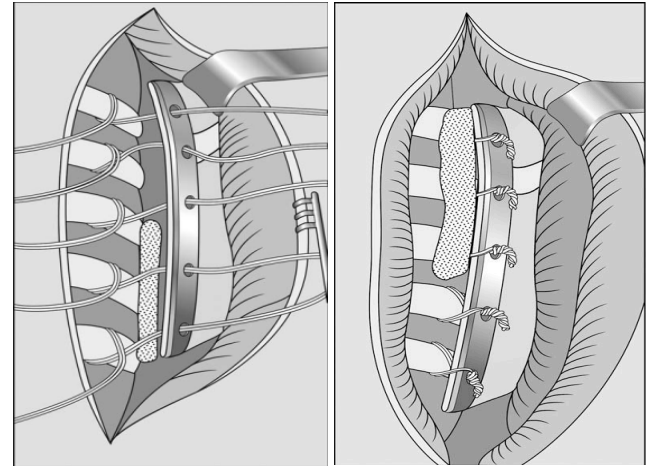
☐ Choosing the right surgeon ✓

☐ Choosing the right procedure

☒ String

☒ Wiring

☐ Scapulothoracic fusion ✓



Courtesy of John Kissel

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Limitations of Evidence-based Guidelines in FSHD

- ❑ FSHD has a wide spectrum of disease severity and rate of progression
 - ❑ Cannot have a one-size fits all guidelines
- ❑ Most studies are retrospective, do not involve large numbers of patients and often involve a non-representative group of patients

BEYOND EVIDENCE-BASED GUIDELINES

Weakness of Eyelid Muscles

- ❑ Weakness can lead to conjunctival dryness, irritation and exposure keratitis.
- ❑ Consider implantation of gold weight in the upper eyelid. Routinely used for facial palsy.



Weakness of the Lower Facial Muscles

- ❑ Weakness with eversion of the lower lip results in poor pronunciation, drooling and spillage of food when eating.
- ❑ Various minimally-invasive surgical techniques have been utilized in individual patients leading to cosmetic as well as functional improvement.

How do we Improve Care Guidelines

- ❑ Need for more longitudinal information in larger numbers of individuals with FSHD
- ❑ This can only be practically done in a registry
- ❑ The National FSHD Registry has about 1000 patients
- ❑ Given the estimated prevalence of FSHD, there is potentially 20,000 individuals with FSHD in the US

Neurology: National Registry for DM and FSHD

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Help Us Advance Research

The National Registry advances research in myotonic dystrophy (DM) and FSHD by helping patients to participate in clinical studies. These studies help doctors, investigators, and care providers better understand the biology, progression, and other important issues in DM and FSHD.

Help Us Learn What's Important to You and Your Family

Registry members complete questionnaires every year about their symptoms. We collect and track your clinical information which can lead to a better understanding of DM and FSHD, how symptoms change over time, and how to improve care for patients.



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