Evidence-Based Guidelines and Beyond

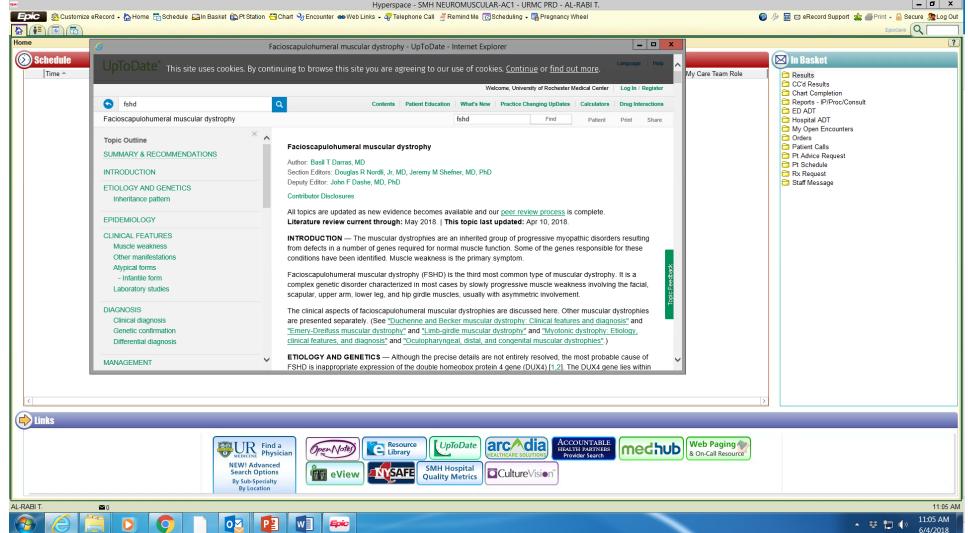
Rabi Tawil, MD

FSHD Connect Meeting

Las Vegas, June 9, 2018







FSHD Care Guidelines 2015

Best available evidence-based guideline for the management of FSHD



(AANEM



EVALUATION, DIAGNOSIS, AND MANAGEMENT OF FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

This is a summary of the Americ dystrophy (FSHD).

Please refer to the full guidel classifications of evidence a







Diagnosis of FSHD

Clinical Context

When clinical presentation of FS straightforward. If, in such circuindividual. However, atypical precounseling, especially with the r

In the most common FSHD type, from >10 repeats to 1–10 repeat to the repeats (PRIN). Available and specific (EVID). In studies the does not appear to improve diag chances of a false-positive result presentation is atypical for FSHI.

Level B

Clinicians genetic c This fact sheet is provided to help you understand the current evidence for diagnosing and managing facioscapulohumeral muscular dystrophy (FSHD).

FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

Summary of Evidence-based Guideline for PATIENTS and their FAMILIES

The American Academy of Neurology (AAN) is the world's largest association of neurologists and neuroscience professionals. Neurologists are doctors who identify and treat diseases of the brain and nervous system, including neuromuscular disorders. The American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) is an association of neurologists, doctors of physical medicine and rehabilitation (PMR), and other health care professionals. PMR doctors specialize in rehabilitation. The AAN is dedicated to promoting the highest quality patient-centered neurologic care. The AANEM is dedicated to advancing the care of patients with muscle and nerve disorders.

Experts from the AAN and AANEM carefully reviewed the available scientific studies on diagnosing and managing FSHD. The following information is based on evidence from those studies and other key information. The information summarizes the main findings of the 2015 AAN and AANEM guideline on FSHD.

To read the full 2015 guideline, visit AAN.com/guidelines.

FSHD is a muscle disease with two genetic causes. People with either genetic cause show similar signs and symptoms. In the majority of cases, genetic testing can confirm an FSHD diagnosis. Doctors should know about and help manage related health problems.

What is FSHD?

FSHD is one of the most common forms of muscular dystrophy (MD). MD is a group of several different genetic diseases. These cause muscle wasting (damage) and weakness. Depending on the MD type, different muscles of the body can be affected. These may include the muscles that control breathing,

Predictors of Severit Clinical Context

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

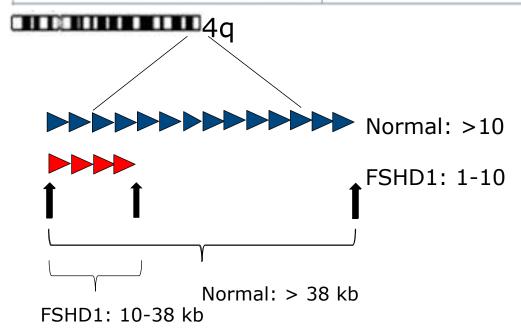
Genetic testing

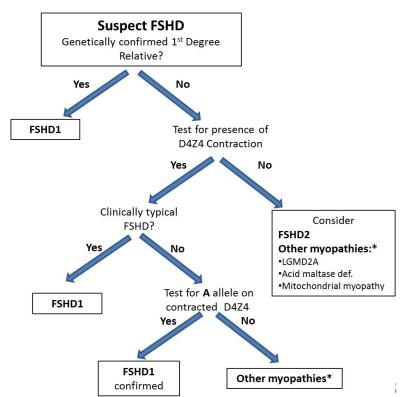
Diagnosis

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 symtpoms

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	

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 (Su	rgery 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

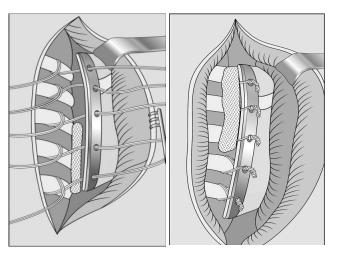


Scapular Fixation

- □Choosing the right surgeon
- □Choosing the right procedure



□Scapulothoracic fusion





Courtesy of John Kissel

Treatment

Treatment: Pharmaceutical Drugs		
Clinicians caring for patients with FSHD	Should not prescribe albuterol, corticosteroid, or diltiazem for improving strength	
Treatment: Scapular Surgical Fixation (Su	rgery 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	

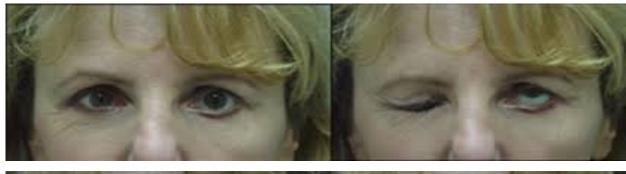
Limitations of Evidence-based Guidelines in FSHD

- □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 nonrepresentative 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

Home Overview Our Team Clinical Services Education & Training Research Grand Rounds News Neurology History Contact Us

Registry Home

<u>UR Medicine</u> / <u>Neurology</u> / National Registry for Myotonic Dystrophy & Facioscapulohumeral Dystrophy

Make a Gift

About the Registry

Join the Registry

Research Studies

Information for Researchers

Information for Patients

Publications

Newsletters

Links

National Registry for Myotonic Dystrophy (DM) & Facioscapulohumeral Dystrophy (FSHD)



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



Join The Registry

Contact Us

Registry Coordinators: Jim Hilbert and Liz Luebbe

Toll free: (888) 925-4302

<u> Email us</u>

Help us make a difference!



News

2017 Newsletter 🖪

New Award Will Advance Muscular Dystrophy Research

Study Identifies Patients'
Priorities in Treating
Rare Muscular
Dystrophy

New Guidelines for Care in FSHD

Moxley and Thornton Honored for Myotonic



MEDICINE of THE HIGHEST ORDER