Physical Therapy
For Facioscapulohumeral Muscular Dystrophy

FSH SOCIETY
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY
The purpose of this guide is to assist physical therapists and individuals with FSHD in developing a plan of care based on the best research evidence, clinician expertise, and patient preferences. To achieve this goal, individuals and their families need to understand the role of physical therapy and therapists in the management of individuals with FSHD, and for physical therapists to have the latest knowledge about FSHD and current management recommendations.
The goals of any physical therapy plan of care are to help individuals:

- maintain optimum health;
- prevent or delay secondary complications;
- maximize functional abilities; and
- improve or maintain quality of life.

Depending on the individual’s signs, symptoms, and functional abilities, the plan may include recommendations about appropriate activities, exercises, modalities for pain relief, guidance for management of fatigue, orthotics/braces, assistive devices, and environmental modifications.

We emphasize individualized plans of care rather than generalized recommendations because FSHD, unlike some of the other muscular dystrophies, is a highly variable condition. Patients can present with a specific combination of muscle involvement and progress at a rate that is unique to them. There is extreme variability relative to age of onset, degree of weakness, pattern of involvement, and rate of progression, even among individuals within the same family.

We hope that this guide will allow you to prepare for and make the most of your consultation with a physical therapist. Remember that FSHD is a rare condition, and unless the therapist is routinely involved in seeing patients with muscular dystrophies, the therapist may not be familiar with your condition and will appreciate any information you can share about FSHD. So please share this booklet with your therapist.

This guide is designed to supplement and complement information available from other lay* and professional** organizations. We look forward to your feedback to help us improve and update our recommendations.

* “About FSHD” brochure from the FSH Society.
** American Academy of Neurology Evidence-based Guideline summary for FSHD care for clinicians and summary for patients and families.
Who Are Physical Therapists?

Physical therapists are healthcare professionals who hold a graduate professional degree (MPT, DPT). They may also be certified specialists in an area such as pediatrics (PCS), geriatrics (GCS), neurology (NCS), orthopedics (OCS), etc. They are required to have a physical therapy (PT) license to practice in each state. They practice in a variety of settings including hospitals and nursing homes, outpatient clinics, home healthcare, and schools.¹

Many individuals with FSHD will probably first encounter a physical therapist in the multidisciplinary clinic where they receive care for their muscular dystrophy-related problems. In this setting, the physical therapist plays a consultative role providing evaluation, education, instructions, and recommendations based on individual patient needs. They may also act as a liaison and help coordinate care with school- or community-based therapists who may be providing direct care.

What can you expect when you see a physical therapist?

During your initial visit, the therapist will:

- obtain a history to understand the development and progression of the problems for which you are being seen;
- inquire about the tests and results that may have already been obtained;
- want to know about any other past or current medical conditions that may have an impact on your current problems;
- note any previous or current medical or surgical treatments you have had related to the muscular dystrophy or any other concomitant conditions;
- perform a detailed examination to document the muscle involvement, joint involvement, functional abilities and limitations, and any problems related to pain and/or fatigue.

After the history and physical examination, the therapist may have a further discussion with you regarding your current activity status based on your family role, occupational needs, or recreational interests. The therapist will inquire about your specific goals and will develop a plan to meet your exercise and activity needs that is compatible with your lifestyle and takes into account your daily routines.

The therapist will demonstrate and teach any specific exercises that may be appropriate and give you a prescription with specifics about frequency, repetition, and duration of exercise and activities. The therapist will also describe the symptoms that may require you to modify the program and/or seek re-examination, and discuss recommendations regarding progression of your individualized program. The number and frequency of visits necessary will be determined by the complexity of the program, your comfort with your own role and responsibilities related to the program, and the therapist’s need to monitor the program.

Based on the examination and discussions, the therapist may also recommend orthotics/braces, assistive devices, and home and workplace modifications to allow you to function at your highest capabilities with the least expenditure of energy. The therapist may also recommend annual evaluations to monitor your condition and make appropriate recommendations.
Facioscapulohumeral Muscular Dystrophy (FSHD)

FSHD is one of the most common forms of inherited muscular dystrophy, affecting approximately one in 8,000 to one in 15,000 individuals. Children and adults of all ages and both genders can be affected. An estimated 10 to 30 percent of the cases arise from new mutations and, hence, there may not be a family history of the disorder. FSHD is almost always an autosomal dominant disorder, meaning that an affected individual who carries the genetic abnormality that causes FSHD has a 50-50 chance of passing the disorder on to each offspring. Over the past decade, major advances have occurred in our understanding of the genetics and molecular mechanisms underlying the condition. For the latest review of molecular mechanisms and genetics, please consult the “ABOUT FSHD” brochure on the FSH Society website at www.fshsociety.org.

For a copy of the American Academy of Neurology’s FSHD care guidelines summary for patients and clinicians, please contact the FSH Society or visit www.fshsociety.org.
CARE RECOMMENDATIONS FOR INDIVIDUALS WITH FSHD

An international group of clinicians has provided care recommendations based on clinical expertise and currently available research evidence. These recommendations include the following:

1. Individuals with childhood-onset FSHD require close monitoring in a multidisciplinary clinic.

2. All individuals with FSHD who have functional limitations should get an initial rehabilitation consultation that includes assessment of posture, gait, function, balance, and the need for orthoses.

3. The consultation should also address complaints related to pain and fatigue, and provide recommendations regarding an appropriate exercise regimen that includes stretching, strengthening, and aerobic training based on the current status of the individual and the current evidence related to exercise in FSHD.
Primary Impairments and Recommendations

Muscular impairments

FSHD is among the most common forms of muscular dystrophy, affecting children and adults of both sexes. The cardinal feature of FSHD is the progressive loss of muscle strength. The disease’s name comes from the typical pattern of weakness at onset: the face (facio), shoulder girdle (scapulo), and upper arms (humeral). However, the disease can differ in the typical initial pattern of weakness: Not every patient experiences facial muscle loss, and many develop muscle weakness in the legs and trunk.

Unlike other muscular dystrophies that present with symmetric involvement, asymmetric weakness is very common in FSHD. The reason for this pattern is not well understood. In the majority of individuals, the weakness progresses very slowly, and approximately 20 percent eventually end up needing a wheelchair for mobility.

The most common initial symptom is difficulty reaching above shoulder level. Foot drop is a less common symptom; such patients, however, almost always have asymptomatic scapular fixator and facial weakness on examination. Truncal weakness is an early and frequent manifestation that is easily overlooked during examination of patients. Weak abdominal muscles result in a protuberant abdomen and contribute to lumbar lordosis. Lower abdominal muscles are weaker than the upper abdominal muscles, causing a strikingly positive Beevor’s sign, a physical finding fairly specific for FSHD. (A Beevor’s sign is seen when a patient flexes the neck forward and the navel moves toward the head.)

In addition to muscles of the face, trunk, and extremities, muscles related to breathing can be involved in the later stages and, hence, regular monitoring of respiratory function is recommended in individuals with moderate to severe involvement. Unlike some of the other muscular dystrophies, cardiac muscle involvement is not present in FSHD, but up to 5 percent of individuals with FSHD may manifest atrial arrhythmias.
Non-muscular impairments

In addition to muscular involvement, retinal abnormalities and high-frequency bilateral sensorineural hearing loss are also associated with FSHD. Usually, these features are mild and do not cause symptoms. However, in the severe, early-onset FSHD, these complications, if not detected early, can lead to loss of vision in the case of retinal disease and problems with speech and cognitive development in the case of uncorrected hearing loss.

Specific Muscular Impairments and Recommendations

Facial muscle weakness

Inability to smile and difficulty with articulation because of facial muscle weakness remain major social concerns. In a small number of individuals, difficulty with swallowing has been reported; referral for swallowing evaluations is recommended in such cases.

Weakness of the orbicularis occuli (muscles around the eye) can cause incomplete closure of the eyelids during sleep, possibly leading to serious exposure keratitis and corneal scarring. Eye drops, ointments, taping, or patches have been recommended but have not always been successful. Surgical insertion of gold weight in upper eyelids or a supportive mesh in the lower eyelid have also been used with some success.
Scapular muscle weakness

Scapular muscle weakness and difficulty with overhead activities are a major problem. In addition, the weakness can lead to pain and fatigue. Severe weakness can cause persistent drooping of the shoulders. If severe enough, this can produce traction on the brachial plexus, a group of nerves in the shoulder/armpit region that supplies the arm. Sometimes this can stretch the nerves severely enough that they can be damaged, producing tingling/numbness, cramps, and further weakness.

The force necessary to keep the scapula stabilized has made the use of taping, slings, and spinal orthosis difficult. “Figure 8” braces for the upper back used for short periods of time have anecdotally provided pain relief and greater ease with specific activities. Surgical procedures may be considered, such as a well-known and tried technique called scapular fixation.

Abdominal muscle weakness

Weak abdominal muscles result in a protuberant abdomen and contribute to lumbar lordosis and complaints of backache and fatigue. Unfortunately, given the marked weakness, it is difficult to strengthen the abdominal muscles; thus, external supports such as abdominal binders and appropriate back supports are recommended.

Ankle muscle weakness

“Foot drop” (difficulty lifting the front part of the foot) caused by ankle dorsiflexor weakness is a common problem. Individuals may first notice problems when walking outdoors on uneven surfaces or when negotiating curbs and steps.

The benefits of specific types of ankle braces or ankle-foot orthoses (AFOs) and the correct timing for intervention have not been well studied. The recommendation regarding orthotic use remains very individualized, since it needs to take into consideration the pattern and severity of weakness; the type, intensity, and duration of activities in which the individual is involved; and any environmental factors related to home
and/or work. It is best to start the discussion regarding orthotic needs sooner rather than later and to see, experience, and discuss the pros and cons of the various options before making a decision.

**Pelvic girdle muscle weakness**

Involvement of the muscles around the hip can lead to difficulties with getting up from a seated position, negotiating stairs, and can make walking long distances difficult and time consuming. At this stage, an individual may want to consider using a scooter for outdoor mobility.

**Secondary Impairments and Recommendations**

**Pain**

Pain is a common complaint in individuals with FSHD. Depending on the survey, 70 to 90 percent of individuals report pain. The intensity and frequency vary and are often attributed to exertion or faulty posture secondary to weakness. As mentioned previously, the muscle involvement and weakness in FSHD are quite unique, leading to muscle imbalances and uneven tension across
joints. A good example is back pain secondary to the abdominal muscle weakness and lordosis of the spine.

Pain has a major impact on the quality of life of individuals with FSHD. The exact mechanisms underlying the reports of pain are not well understood, and neither is the natural history of pain. For example, it is not well understood whether pain worsens with age or is associated with severity of weakness, strenuousness of activity, type of activity, etc.

To effectively manage this symptom, it is essential to understand the location, nature, duration, and the activities and environmental factors that worsen or ease the pain. A wide variety of methods have been used to treat pain in individuals with FSHD. The use of non-steroidal anti-inflammatory medications, exercise (strengthening and range of motion), heat, and massage are the most common therapies used to manage pain.

**Fatigue**

As many as 60 to 90 percent of individuals with FSHD report problems with fatigue, often with negative impacts on function and quality of life. Like pain, the exact mechanisms underlying fatigue are not well understood, and the natural history is not well documented. Pain itself can cause fatigue.

In a recent report of a randomized controlled trial from the Netherlands, clinicians found that moderate aerobic exercise combined with cognitive behavioral therapy (CBT) reduces chronic fatigue. CBT is tailored to each individual to challenge negative patterns of thought in order to alter unwanted behavior patterns.

**Imbalance**

Due to muscle weakness, individuals with FSHD develop poor balance, which increases the risk of falling. Fear of falling may lead to restricted activity and decreased quality of life. PT can improve balance and make recommendations to reduce falls, and thereby enhance quality of life.
Exercise Recommendations

Individuals with FSHD often have questions about exercise. Exercises including flexibility/range of motion, strengthening/resistive, and cardiovascular/aerobic exercise are important for the management of the musculoskeletal manifestations of FSHD and for the overall health and fitness of the individual. More recently, the importance of neuromotor training, functional exercise, and balance training have also been recognized and recommended by the American College of Sports Medicine (ACSM).

The evidence available regarding the role of exercise in FSHD is limited. In extensive reviews on the topic published in 2010 and 2013, the authors examined the safety and efficacy of strength and aerobic training in neuromuscular diseases using randomized controlled trials (RCTs). They identified one study in which the benefits of six (6) months of exercise followed by an additional six (6) months of medication (albuterol) versus placebo were examined in 65 individuals with FSHD. Based on the findings of the study, the authors concluded that moderate-intensity strength training does no harm, but there is insufficient evidence that it offers benefits. In a 2007 review, evidence related to exercise in individuals with neuromuscular diseases was evaluated. Based on their analysis of the studies, the researchers concluded that the evidence suggests strengthening exercises in combination with aerobic exercises are “likely to be effective.”
Exercise: how much?

Given the evidence from the two (2) major reviews that exercise may be effective and that moderate-intensity exercise does not worsen disease progression, some general recommendations regarding exercise can be made to guide clinicians and individuals with FSHD using the current physical activity guidelines from the U.S. Department of Health and Human Services (HHS). (See www.health.gov/paguidelines.)

These suggest that for all individuals, some activity is better than none, and that the health benefits of physical activity far outweigh the risks. The guidelines recommend that children, adolescents, adults (ages 18-64), and older adults follow the appropriate guidelines to the best of their ability. Individuals with chronic conditions perform as much activity and/or exercise as their condition allows.

The HHS guidelines recommend about two (2) hours and 30 minutes each week of moderate-intensity aerobic exercise. The exercise can be spread out in 30-minute sessions five (5) times per week or even in episodes of at least 10 minutes three (3) times per day, preferably spread throughout the week. Muscle strengthening activities that involve all major muscle groups should be performed at least two to three (2-3) days a week.

A personalized program, baseline measures, and a daily log

Prior to beginning an exercise program, individuals should undergo baseline measures of strength and exercise capacity, provide a comprehensive description of their daily activities related to personal care, household duties, and work-related requirements. They should also share their preferred leisure activities and personal goals they would like to achieve. Based on all this information, a personalized program can be developed. The personal program should not only include instructions about the type of exercise, but also information about repetitions, intensity, duration, and guidance about signs and symptoms related to overwork, how much to cut back, and how to progress.
It is often helpful for individuals to maintain a daily log of activities of daily living, work-related activities, specific exercises performed, and symptoms experienced on the following day. By documenting physical tasks (both routine and exercise) along with physical well-being (including pain and fatigue), individuals with FSHD will be able to provide proper feedback to the therapist supervising the individualized program.

**Aerobic, flexibility, and strengthening exercises**

Moderate-intensity aerobic exercises are activities that can be performed while still continuing a conversation—without having to stop to catch your breath. Examples include walking briskly, biking on level ground or on a stationary bicycle, using hand cycles, ballroom and line dancing, lawn mowing, snow shoveling, and general gardening and household activities. Strengthening exercise can be performed in several ways—with resistance provided by gravity, water in a pool, or with equipment such as elastic bands, free weights, and machines. Yoga and Pilates types of exercises may also be recommended as part of a strengthening program, although there are no studies reported that have examined the effects of these specific interventions in individuals with FSHD.

Flexibility/range of motion (ROM) exercises are important in maintaining joint function and may play a role in reducing pain that is caused by muscular
imbalance or tightness. As muscles atrophy, resulting in weakness, gravitational pull may limit a person’s ability to move a body part through its entire range of motion. For example, as the dorsiflexors at the ankle become weaker while the plantarflexor remains stronger, an imbalance is created at the ankle, resulting in a tendency to develop tightness and loss of ankle range of motion. Another example: Due to the weakness of the shoulder stabilizers, overhead activities become difficult and individuals have difficulty raising their arms in a sitting or standing position but may have the ability to perform this movement when lying down in a supine position where gravity is eliminated.

Current recommendations regarding flexibility/ROM exercises are that they be done daily or at least three (3) times a week. If tightness has already developed, a gentle stretch should be maintained for 10 to 30 seconds and the exercise repeated two to four (2 to 4) times. Stretching should be performed when the muscles are warmed up, either after cardiorespiratory exercise or after a bath or use of a hot pack. Individuals may also participate in flexibility exercises that are more dynamic in nature. These include yoga- and Pilates-based activity that can either be done individually or in a class setting.

Weakness occurs as part of the disease process; however, weakness may also develop as a result of disuse. Exercises may help minimize the disuse weakness, but there is also a concern that too much exercise or inappropriate exercise may hasten or worsen weakness and, therefore, finding the right balance of exercises for each individual is important.

**Hydrotherapy (aquatic/water therapy)**

There have been no controlled trials studying the effects of hydrotherapy in FSHD. Physical therapists encourage hydrotherapy in all types of muscular dystrophy because water can be used to assist as well as resist movements, is a fun medium for younger individuals to exercise in, and the warmth and buoyancy can be relaxing. Also, hydrotherapy can be an efficient means of exercise, as
many muscles can be exercised at the same time. On a practical basis, however, adults may avoid hydrotherapy because of accessibility, safety issues, or social concerns.

**Neuromuscular electrical stimulation**

The use of neuromuscular electrical stimulation (NMES) has been studied for decades in individuals with muscular dystrophies and other chronic conditions causing atrophy and weakness. The goal has been to prevent disuse atrophy in those unable to exercise actively.

An open, pilot study of NMES in nine (9) individuals with FSHD who underwent stimulation of the deltoid, trapezius, and quadriceps muscles five (5) days per week for five (5) months reported increases in strength and in the 6-minute walk test. The authors concluded that the use of NMES appears to be safe, feasible, and well tolerated, but requires further study with multicenter randomized controlled trials.
Providing physical therapy for the patient with FSHD introduces special challenges for the therapist. More than other muscular dystrophies, FSHD can be quite asymmetrical. For example, it is not unusual to grade a muscle on one side as a 4 or above by MMT and the corresponding muscle on the other side as a 1 or 2 grade. There often appears to be no rhyme or reason to the specific muscle group strengths. However, there are usually some predictive patterns:

- The upper and lower abdominals are among the first muscle groups to show clinical weakness and are rarely graded above a 3.
- The middle and lower trapezius and serratus anterior muscles also weaken early and are rarely above a 3 grade. The middle deltoid, however, usually is only mildly weakened.
- Shoulder external rotators are weaker than shoulder internal rotators.
- Biceps brachii is usually preferentially involved.
- Knee flexors are weaker than knee extensors.
- Ankle dorsiflexors are weaker than ankle plantar flexors.
- Despite, on occasion, radical strength imbalances between a muscle antagonist and agonist, significant joint contractures are rare.

Because this autosomal dominant disease is slowly progressive, and perhaps because other members of the family may also have “toughed it out” with symptoms for many years, patients sometimes are not referred to a physical therapist until they have endured decades of chronic pain, weakness, and disability.
The therapist seeing a patient with FSHD for the first time must begin by gathering information regarding the person’s occupation, leisure activities, and role as a parent or caregiver to another person. This should involve detailed questioning of the patient’s job description, hours at work, responsibilities of keeping a house or apartment, childcare, pet care, and any other regular physical tasks. It is necessary for the therapist to have a clear sense of the percent of time each day the person with FSHD spends sitting at a computer, standing, walking, climbing stairs, getting into and out of a vehicle, doing laundry, preparing meals, walking a dog, carrying a toddler, etc.

A detailed strength evaluation should then follow. Typically, as this is completed, the therapist has become aware of significant muscle imbalances. Very weak muscles are often overstretched. Strong muscles may be tight, but again, are probably not producing significant contractures.

The person with FSHD almost inevitably is suffering from chronic pain. The areas most often affected are the cervical area, the upper back, the lower back, and the posterior knee. The areas of pain will correspond with the weakest (and thus overstretched) muscles.

**Treatment of pain**

Ask the patient to keep an activity log for at least several weeks. This can be as detailed as the patient needs it.
to be, but at minimum, it should have columns for the type of activity, the time of day (15 minutes to blocks of several hours), and symptoms noticed. If the person is accurate and thorough, such a log may well provide very useful information concerning the correspondence of some specific activities and symptoms of fatigue or pain.

Treatment of the pain should include instruction for any appropriate stretching or strengthening exercises, evaluation for possible bracing (especially to assist in ankle dorsiflexion, scapular retraction, or to provide abdominal support), and modalities such as moist heat, cold, TENS, massage, acupuncture, etc. Medication may also be required.

The use of lightweight, off-the-shelf, or custom-made ankle-foot orthoses (AFOs) works well to correct foot drop. The new stance-control knee-ankle-foot orthoses (KAFOs) may benefit a minority of FSHD adults who have significant quadriceps weakness (especially unilaterally) or those who present with knee pain due to chronic genu recurvatum caused by an imbalance between knee flexors that are weaker than extensors. The type of stance-control orthosis that has double uprights at the ankle usually benefits an individual with FSHD in terms of increased stability and decreased fatigue.

Most importantly, the therapist must teach the individual with FSHD strategies to avoid the cycle of chronic musculoskeletal pain. This includes education of any postural deficiencies and ways to minimize them, an honest appraisal of the individual’s lifestyle—both leisure and work—and suggestions for alterations that might result in less emphasis on the weakest muscles. Suggestions could be as simple as trying certain adaptive equipment for bathing, dressing, or housekeeping at home, or a more supportive or height-appropriate desk chair, or ergonomically correct keyboard and mouse for a laptop or computer station at work.

Even if individuals choose not to use a recommended orthotic device or adaptive equipment, they need to understand the principle behind the recommendation.
References


ABOUT THE AUTHORS

Shree Pandya, PT, DPT, MS, is associate professor of neurology at the University of Rochester School of Medicine and Dentistry. Since the 1980s, Dr. Pandya has participated in clinical care, research, education, and advocacy related to muscular dystrophy. She has helped develop evidence- and consensus-based recommendations for the management of the most common muscular dystrophies.

Kate Eichinger, PhD, PT, DPT, NCS, is a physical therapist in the Neuromuscular Disease Center at the University of Rochester. She is certified by the American Board of Physical Therapy Specialties in Neurologic Physical Therapy. In addition to the clinical care of individuals with adult and pediatric neuromuscular conditions, she is involved in natural history studies and clinical trials of patients with FSHD and other neuromuscular disorders.
Become part of the solution!

Too often, we hear people say they’ll volunteer when there’s a treatment. But that day will never arrive unless patients participate in research now. Equally important are family members, both affected and unaffected. Comparing a parent or sibling who has very mild symptoms with a person who has more severe symptoms could provide insight for future treatments. Ironically, the mildly affected and unaffected are least likely to volunteer for research, yet they may hold the key to a treatment.

By volunteering for research, you will help move us a step closer to a breakthrough.

For more information or to support the work of the FSH Society, visit www.fshsociety.org.

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☐ Please have someone from the FSH Society call me.

Tell us about you:

☐ I have FSHD
☐ I have family members with FSHD
☐ I treat patients with FSHD
☐ I am a researcher interested in FSHD
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Medication alert: If narcotics are necessary for pain control, respiratory function must be closely monitored.
☐ Patient uses BiPAP ventilation support. Monitor for CO2 retention.
☐ Administer oxygen only with BiPAP ventilation.

EMERGENCY CONTACT NAME & PHONE: _______________________________________

PHYSICIAN NAME & PHONE: ________________________________________________

If patient is incapacitated and non-responsive, all medical information and healthcare decisions should be disclosed, discussed, and decided with:

____________________________________

SIGNED: ________________________________________________________________

MEDICAL ALERT: Facioscapulohumeral muscular dystrophy (FSHD) patient

NAME: ____________________________ BLOOD TYPE: ____________________________

Medication alert: If narcotics are necessary for pain control, respiratory function must be closely monitored.
☐ Patient uses BiPAP ventilation support. Monitor for CO2 retention.
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