

MANAGING FSHD: ONE PATIENT'S PERSPECTIVE

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1. Introduction. I am 56 years old and was diagnosed with FSHD in 1975. I began to use a power wheelchair at age 32, while I was still able to walk a bit indoors under ideal conditions, and lost the ability to walk at 38 when I broke my femur in a fall at home. This paper draws on my experiences, those of many other people I've known with FSHD, and advice from many sources over many years. I'm not a doctor, physical therapist or other medical or scientific professional. **This paper is not medical advice, nor is it science. It is *imperative* that you consult your own medical professionals before beginning or modifying any of the therapies described in this paper or following the suggestions herein.**

This paper is being distributed at the FSH Society's 2014 FSHD Connect Meeting. **However, it contains *solely my own views* and not necessarily those of the FSH Society or any of its officers, directors, employees or members.**

This paper includes advice on dealing with medical professionals and discusses certain therapies. By using the word "therapy," I do not mean to suggest that FSHD can be treated (yet!), but rather that its physical, mental and emotional effects can be managed, coped with and mitigated.

It is critical always to remember that each of us is unique and FSHD is highly variable, even among family affected members. What works for one person may not necessarily work for others. My goal in this paper is to provide a wide range of suggestions in the hope that each reader will find something useful for his or her unique situation.

Some suggestions may be obvious, but we often take things for granted so much that we forget to step back and take another look. Also, it's sometimes easy to forget that managing your FSHD requires, first and foremost, managing your overall health. Healthy habits and practices will help you manage your FSHD, while unhealthy ones such as smoking, poor diet and excessive stress will make it harder to cope with FSHD, even if they may not directly weaken your muscles.

2. FSHD Patients are Like Professional Athletes. Paradoxically and ironically, FSHD patients have much in common with professional athletes. Both groups function at the limits of what their bodies can do, both are constantly reminded of the limits, both unavoidably risk injury and overuse, and both have a heightened awareness of their bodies. An FSHD patient must see himself or herself as an athlete in the game of life, paying close attention to his or her body and caring for it diligently, the way an athlete does.

3. The Patient has Ultimate Responsibility. For me, a good thing about having FSHD has been the privilege of being cared for by resourceful, compassionate, dedicated and brilliant doctors, physical therapists, occupational therapists, respiratory therapists and other medical professionals. Not all FSHD patients have been so fortunate. But regardless of how good, bad or indifferent the medical care each of us receives, the tenet that each patient has the right and ultimate responsibility to make decisions about his or her healthcare applies to managing FSHD, and perhaps especially so.

The specifics and extent of each person's progression are different, lifestyle and other environmental factors and choices vary greatly, very little certainty exists about potential therapies, expert opinions frequently differ, and though knowledge is expanding, much remains unknown. Tradeoffs are often involved - therapies take time, effort, money, energy and resourcefulness, and what one person may consider worthwhile, another may consider too difficult, risky and costly.

Medical professionals vary in their knowledge, experience, empathy, bias, resources, access to expertise, and communication skills. It's impossible for any one professional - no matter how experienced, empathetic and brilliant - to know about or have experience with every potentially useful therapy. Just because a professional may not suggest something doesn't mean it shouldn't be explored. On the other hand, it's important to be extremely wary of claims that an effective therapy or practice exists that medical professionals, researchers, patient organizations and drug companies are completely ignorant about, or that, for selfish reasons, they are trying to hide from patients. Patients must be proactive - but prudent - in learning about, seeking and trying therapies and strategies.

It is critical to communicate effectively, frankly and completely with your medical caregivers. Tell them not only about your FSHD symptoms, but also about: job and lifestyle factors; other medical conditions; sources of stress in your life; your home environment; your activities of daily living; medications you are taking; and all therapies you have tried, are doing and are considering. Remember that your doctor and therapist have many patients, and what's foremost in your mind may not be prominent in theirs. Something you told them last year may not be recorded in the chart notes and they may not remember it.

4. A Team Approach is Crucial. A wise neurologist, primary care physician or physiatrist (a medical doctor specialized in physical medicine and rehabilitation) knows that his or her knowledge and experience are limited, especially as medical knowledge increases over time, and will actively embrace and bring in experts from other disciplines. Dealing effectively with FSHD requires a team approach, and each member of the team (doctor, PT, OT, respiratory therapist, medical equipment specialist, etc.) should have experience with FSHD patients, or at least with patients who have other muscular dystrophies. If expertise is not available where you live, I urge you to consider traveling for a consultation with experts.

If FSHD affects one person in 14,000 (this is a conservative estimate; actual incidence may be higher), and if your internist has 1,500 patients at any time, then you are probably his or her only current patient with FSHD and maybe the only one in an entire career. My primary care doctor, an internist, is superb, but he isn't an expert on muscular dystrophy, so I also see my neurologist annually, who is an expert. Even though there is no cure or treatment for FSHD, my neurologist and his team of PT, OT, equipment expert, respiratory therapist and social worker monitor my progression, see how I'm doing, and suggest changes and strategies. My primary care doctor and neurologist know each other and communicate regularly.

5. FSHD Complicates Other Health Issues and Can Have Important Indirect Effects. It's important to be aware of how FSHD can affect things that are not direct or obvious. For example, you may begin to feel pain in your knee and be unaware of the source, knowing that you haven't recently had a fall or injury. The x-rays are normal, and an orthopedist who lacks experience in muscular dystrophy may not be able to find the cause of the pain. But it may be that your leg has gotten weaker by just enough that the amount of standing, walking and climbing stairs you normally do has begun to strain your knee. However, you may not yet actually feel or notice that your leg has gotten weaker. A neurologist or orthopedist with experience in muscular dystrophy may have an educated hunch about the cause, and if he is perceptive and knows you well, may even observe a small change in your gait. He might recommend a soft knee support and more frequent rests when standing or walking, which might solve the problem.

This example illustrates the importance of understanding the less obvious effects of FSHD. It's also important, however, not to attribute to FSHD a symptom that the doctor and patient simply can't explain. If the symptom is not either a common direct symptom of FSHD or a logical indirect effect, the doctor and the patient must continue trying to find the cause and not just assume it's due to FSHD.

The side effects of some medications and other medical treatments may be more problematic for FSHD patients than for other people. For example, a medication for urinary problems may cause mild dizziness that able-bodied people can manage safely, but greatly increases the risk of falls and injury for an FSHD patient who walks with difficulty or who uses a wheelchair and needs stability and concentration to transfer in and out of the wheelchair. An urologist may focus only on how successful the drug usually is in treating this type of urinary problem, urging the patient to take the drug and tolerate the dizziness. The FSHD patient needs to explain to the urologist why the side effects of this medication are far more problematic for him than for most patients, and to press the doctor to consider an alternative treatment.

Some x-rays, other diagnostic tests and medical treatments may be more difficult for FSHD patients with significant mobility limitations (as for others with similar limitations) than for able-bodied people. Make sure your doctor is aware of your mobility limitations so he or she can work with you to provide alternatives. For example, my dental hygienist cleans and x-rays my teeth, and my dentist examines them, while I remain in my Permobil wheelchair with the back reclined. (Procedures such as filling a

cavity do require transferring to the dental chair.) My ophthalmologist checks my eye pressure with a portable device and performs other eye tests while I remain in my wheelchair (although a complete eye exam requires transferring to the doctor's chair).

There are often alternatives to the most common or advanced diagnostic or treatment methods, but the doctor may be out of the habit of ordering them. Sometimes a low-tech method works just fine and is easier for wheelchair users.

Recuperation from illness or injury is slower and more difficult for many people with FSHD. It's important to be aware of this, accept it and explain it to your doctor. Most doctors are very willing to advocate for their patients, but you need to give yours detailed facts to help him advocate for you. A minor surgery that requires one night's stay in a hospital for most people might require two or three nights for an FSHD patient, whose body may not have that extra bit of "oomph" to bounce back as fast from the trauma of the surgery and the post-operative inactivity. The FSHD patient may also benefit from having the 24x7 help that's available in a hospital. Having help may reduce the risk of falling at a time when the patient is weak and in pain. Paradoxically, it also may permit the patient to begin exercising more quickly than if he were at home without help.

Inactivity is more problematic for FSHD patients because of the muscle breakdown of FSHD. To reduce the risk of flu and the resulting inactivity, I've had flu shots for as long as I can remember, even at a time when young and middle-aged adults typically weren't encouraged to get flu shots. Some surgeries can be done using either a general or a local anesthetic; FSHD patients who can tolerate the pain may consider using a local to shorten the period of inactivity.

6. Respiratory Impairment, Ventilatory Support and Stacked Breathing Exercises. Although it had generally been thought that FSHD doesn't involve respiratory impairment, in recent years some doctors have found that respiratory impairment, including sleep apnea and respiratory insufficiency, is present in some FSHD patients. Whether the cause is due to FSHD, other factors or a combination has not been conclusively proven. It's logical to expect that weak chest and diaphragm muscles have a negative affect on breathing, so as these muscles weaken in FSHD patients, breathing may be affected. And abdominal weakness common to FSHD patients may exacerbate this. Fatigue is often part of FSHD because our muscles have to work harder, but there also may be additional causes of fatigue such as interrupted sleep due to respiratory problems. If you feel generally fatigued, or you begin snoring loudly or in a different pattern than usual, or your spouse or partner notices that your breathing while in bed is labored and interrupted, consider discussing breathing tests with your doctor, including forced vital capacity and nocturnal oxymetry tests. These are easy, non-invasive tests that don't require a hospital stay.

If indicated by the tests, BI-PAP or other mechanical ventilation at night can increase oxygen and greatly improve sleep. The tests should be repeated periodically to ensure that the settings on the machine are appropriate. Your doctor should consult a

respiratory therapist as early as possible and the RT should remain involved in monitoring your progress. Because many doctors, even experienced neurologists, don't associate FSHD with respiratory problems, your doctor may be reluctant to order respiratory tests; be persistent if you feel fatigue or the other symptoms described above.

Stacked breathing exercises can be helpful. They involve a facemask attached to a football-shaped plastic bladder. A helper holds the facemask around the mouth of the person doing the exercise. The person inhales rapidly several times in succession without exhaling, while the helper squeezes the bladder to push air into the person's lungs. This exercises the chest muscles and increases oxygen intake. These exercises don't require much time and a family member can help you do them.

7. Studies Have Limits. Because of the extreme variability among FSHD patients in which muscles are affected and the extent to which they are affected; environmental factors such as diet, exercise, stress, climate, job; and a myriad of other factors, it is extremely difficult not only to generalize but also to quantify and prove whether or not many of the therapies are effective. Even for an individual patient, the progression isn't linear, so it's impossible to prove what would have happened had a particular therapy been utilized or not. (Even if the FSHD progression is demonstrably slower after a course of therapy, how can it be proven that the slowdown was due to the therapy and wouldn't have happened anyway?)

Physical therapy doesn't seem amenable to double blind studies of the type that are done in drug trials. Also, measurement tools don't necessarily capture the whole story - a patient may truly feel and function better even though improvement can't be objectively demonstrated. Or the amount demonstrated may be small even though the patient feels and actually functions significantly better. The 1 to 5 scale, with pluses and minuses, typically used to measure muscle strength is a fairly gross measurement and may not be capable of capturing increments that may be small on a macro level but are significant for a patient's ability to function. Therefore, in determining whether or not a therapy has been effective, great weight should be given to the patient's opinion, subjective though it may be, and to functional improvement in activities of daily living.

8. The Benefits of Exercise. Stretching, floor exercises such as modified stomach crunches and pelvic tilts, and light weights (weight machines, pulleys, light dumbbells) and other resistance exercises (such as theraband) have the following benefits: maintaining range of motion and preventing stiffness and contractures; improving circulation; and minimizing or mitigating potential weight gain by burning calories. But the risk of overuse is ever-present, so it's essential to monitor closely how you feel. When in doubt about an exercise, don't do it. When in pain, stop.

A very complex, difficult and controversial issue is whether weights and other resistance exercises have the potential to strengthen affected muscles or, at least, slow the FSHD progression. It may not really be possible to provide a definitive answer, but because of the benefits described above, these should be attempted (slowly and cautiously) so long as the affected muscles aren't being overtaxed or inflamed.

Also, some physical therapists and physiatrists believe that resistance exercises may possibly increase or maintain the strength of muscles that are unaffected or only minimally affected by FSHD. Because the unaffected/minimally affected muscles vary among patients, and because it is difficult to isolate these muscles from the affected ones and exercise them sufficiently without overtaxing and inflaming the affected ones, what is appropriate differs for each patient and can be determined only by experience and in consultation with a physical therapist.

Above all, don't undertake an exercise program without consulting a physical therapist or occupational therapist who knows about muscular dystrophy.

9. The Physical Therapist's and Patient's Roles. Initially, it is crucial to have a PT who is experienced with FSHD patients directly administer certain of the exercises (e.g. stretching) and, for other exercises, teach you and observe you to ensure you are doing them correctly and are not overtaxing the muscles. However, insurance companies won't pay for physical therapy indefinitely, PTs' time is in great demand, and physical therapy is time-consuming for the patient. So the goal should be to learn the exercises and, after a course of physical therapy sessions, be able to do them on your own or with the help of family members or friends. But it's important to ask your doctor to authorize physical therapy when you feel your condition is changing significantly, periodically to monitor how you perform the exercises, and to treat specific conditions or events such as strained or pulled muscles or injuries from falls. It's useful to go to a PT for a "tune up" every so often.

10. Aerobics. Exercise bicycles (both lower body and upper body) and other forms of aerobic exercise are very beneficial for heart health, circulation, stress relief, good sleep, maintaining range of motion, preventing stiffness and contractures, and burning calories.

11. Swimming and Water Exercise. Swimming is very beneficial for many of the same reasons as exercise bicycles. Swimming has a low impact on the bones and muscles and a low risk of injury. The buoyancy of the water provides resistance exercise and also enables people to do things they might not be able to do on land. For example, I was able to walk a bit in the water even after I lost the ability to walk on land. Stretching exercises in the water are also helpful. For myself and many people I know with greatly reduced mobility, being in the water provides a freedom of movement we can't find anywhere else and a welcome feeling of exhilaration, energy and relief from stress.

12. Massage and Watsu. Massage is excellent for preventing contractures, maintaining circulation and reducing stress. Watsu, a form of massage administered in the water, combines the benefits of ordinary massage and being in the water. I really enjoy being in the water, and I feel great after a Watsu treatment. Both massage and Watsu should be done only by certified professionals. www.Watsu.com.

13. Standing Frame. For people who can no longer walk, standing in a standing frame, daily if possible, can be very helpful. It provides weight bearing to strengthen bones, helps maintain range of motion and prevent contractures, relieves pressure on the posterior, increases circulation, reduces stress, and realigns/improves functioning of the internal organs. Standing and walking in a swimming pool (with ankle weights if necessary) provides similar benefits. A PT should check to ensure you are doing these therapies correctly and that the standing frame settings are correct, your posture is good and you don't overdo it.

14. Occupational Therapy at Home and at Work. People whose FSHD has progressed beyond mild will benefit by regular consultation with an occupational therapist for: improving daily living activities to increase function and reduce stress, fatigue and risk of injury; positioning and seat cushions (if the patient is in a wheelchair); and functioning at work (ideally, with an OT who is a workplace ergonomics expert). A good OT will be expert in assistive technology. Many employers are willing to pay for an ergonomics consultation at the workplace; in most cases where the employee has a "disability" per the Americans with Disabilities Act, the employer is legally required to do so. Home visits are also essential: there is no substitute for having an OT observe the actual conditions of daily living and recommend ways to improve safety, efficiency and comfort. Home visits also benefit caregivers by teaching them how to reduce their stress, burden and risk. Ideally, an OT and a PT should do the home visit together.

15. Durable Medical Equipment. A good durable medical equipment (DME) dealer is very valuable, and it's important to find a dealer you respect. However, do not rely only on a DME dealer for recommendations. Consult an independent occupational therapist about equipment such as wheelchairs, shower/commode chairs, canes, walkers, lifters, ramps, electric beds, etc. Ideally, the DME dealer will be an OT or will have one on staff. The dealer also should be willing to work with your OT. Attending "abilities expos" and similar trade shows is an excellent way to see and try equipment. Emotionally difficult as it may be, it's a good idea to check out equipment well before you really need it. Above all, be skeptical about advice from dealers selected by your medical insurance company or about direct advice from the insurance company. Medical insurance companies (even so-called nonprofit ones) are concerned about their bottom line, not your health.

16. Driving Evaluation; Adaptive Driving. If you feel uncomfortable or unsafe driving, get evaluated by an adaptive driving expert. Some hospitals have adaptive driving programs; these are usually administered by an OT who is also a certified driving instructor. Evaluations are expensive and, unfortunately, insurance usually doesn't cover them. But if driving is necessary to maintain your employment, the rehabilitation agencies of some states may pay part or all the cost of the evaluation and adaptive driving equipment. An increasing variety of adaptive driving technology is available.

17. Get Plenty of Sleep. Study after study has confirmed that getting enough sleep is vital for everyone. Even an hour per night less than optimum sleep time probably

has significant negative long-term effects on able-bodied people. Because FSHD patients get fatigued more easily than able-bodied people and our muscles are under constant additional stress, it's essential to get plenty of sleep, and to get it consistently - don't shortchange yourself and then try to catch up.

18. Stress Reduction. Stress harms the body in many ways. Although there may be no scientific proof that stress specifically worsens FSHD progression, it's reasonable to believe that it does. Stress reduction techniques such as meditation, yoga, and breathing exercises can be very beneficial.

19. Alternative Bodywork. Gentle, non-stressful, low impact bodywork techniques such as the Alexander Technique and Feldenkrais Method can be helpful. These techniques aim to make us aware of how we actually use our bodies and how we can improve our habits. Though these techniques won't strengthen muscles, they can improve posture and breathing, help maintain balance and range of motion, reduce stiffness and pain, reduce fatigue and improve overall health. I have studied the Alexander Technique and found it to be very beneficial. www.AlexanderTechnique.com. www.Feldenkrais.com.

20. Anti-clotting Medications when Flying. FSHD patients who can't walk are at risk of developing blood clots on long plane flights. Per my neurologist's suggestion, I take a prescription medication, Lovenox, before flights to reduce the risk of clotting.

21. Scapular Fixation. This term refers to a variety of surgical procedures that stabilize the scapula by attaching it to the back of the rib cage to prevent it from "winging." The aim is to improve functional ability in daily activities such as eating, drinking, reaching and lifting. These procedures are complex, and many factors must be considered.

More than 30 years ago I had one shoulder blade fixed by the "scapular fusion" procedure. A piece of bone was taken from my pelvis and used to fuse my shoulder blade to a rib. This resulted in far greater functional ability forward and above my head (for example, in eating, brushing my teeth and reaching things on shelves) but far less range of motion behind my back. At the time of my surgery the cast extended from my pelvis to my shoulders and remained on for 13 weeks. The recuperation period was difficult, with significant inactivity.

In today's procedures the cast is smaller and the recuperation period is shorter but still not easy. Scapular fusion and other fixation procedures have strong pros and cons, and I urge anyone considering these procedures to consider only a surgeon with significant experience, discuss the procedure and recuperation in detail with the surgeon, and speak with several FSHD patients who've had the procedure done recently by that surgeon. The recuperation period requires careful planning to minimize inactivity; before having this procedure, consult with a physical therapist and an occupational therapist with relevant experience.

22. Conclusion. A myriad of therapies, techniques and strategies are available to manage, cope with and mitigate the effects of FSHD. Neurologists, primary care doctors, orthopedists, physiatrists, physical therapists, occupational therapists, respiratory therapists, medical equipment dealers and other professionals each have an important role to play. They care, they are expert, they are dedicated, and they will listen, but they don't and can't know everything. Each FSHD patient is different, and nobody knows yourself as well as you do.

You must take the initiative! I hope this paper has given you some useful ideas about managing your FSHD.