



# About FSHD

Facioscapulohumeral  
Muscular  
Dystrophy



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## Facioscapulohumeral

(fā-sh(ē)-ō- skap-y<sup>ə</sup>-lō-'hyūm-(<sup>ə</sup>-)r<sup>ə</sup>l)



**Facioscapulohumeral muscular dystrophy is a heritable muscle disease, often called FSH or FSHD. Progressive weakening and loss of skeletal muscle are its major effects. It has significant medical and health impacts on individuals, families, and society. (It is also known as Landouzy-Dejerine disease, after the two French neurologists who first documented it in the late 19th century.)**

## What is FSHD?

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 torso.

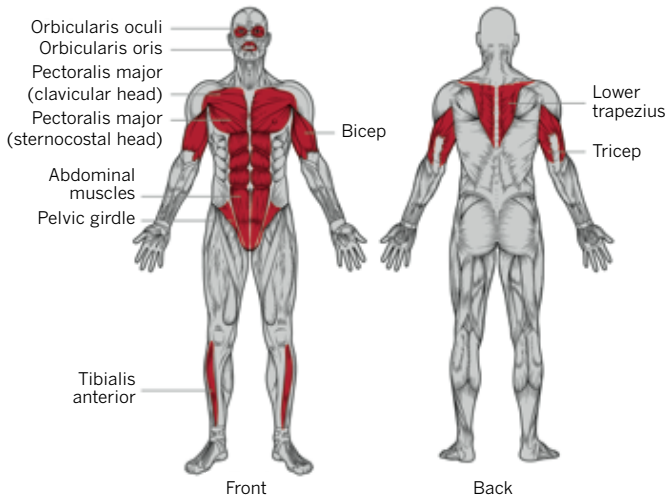
The symptoms can develop at any age, from infancy through advanced age. Many patients recall being unable to whistle, smile, or close the eyelids as a child. The majority of males are diagnosed by age 20, and females by age 30. About 4 percent of cases are diagnosed in children under the age of 5. These early-onset or infantile-onset (iFSHD) patients are at greater risk of having more severe symptoms and added health complications.

Although the progression of FSHD is variable, it is usually relatively slow. Asymmetry is a hallmark of FSHD. Most patients will observe that one arm (or shoulder blade, or lower leg) is weakened, while the other remains stronger. The reason for this asymmetry is unknown.

Early weaknesses of the muscles around the eye (difficulty closing the eye) and mouth (difficulty smiling, puckering the lips, or whistling) are distinctive for FSHD. Facial weakness in combination with weaknesses in the muscles that stabilize the shoulder blades, which result in "winging" of the scapula, is often the basis of the physician's initial diagnosis of FSHD.

As the disease progresses, the lower and upper leg muscles are often affected. About 20 percent of FSHD patients overall will become dependent on a wheelchair or scooter. Weakness in the abdominal muscles can cause a protuberant abdomen and lumbar lordosis ("sway back"). The lower abdominal muscles are usually weaker than the upper abdominal muscles. This results in a movement of the navel toward the head upon flexing the neck. Doctors call this a positive Beevor's sign; it is not seen in many other diseases and is a physical characteristic very specific to FSHD.

## FACIOSCAPULOHUMERAL DYSTROPHY



**This diagram shows the muscles that are typically affected in FSHD. Reprinted from Trends in Molecular Medicine, 2015 May; 21(5):295-306, Lek A, et al. Copyright 2015, with permission from Elsevier.**

## SYMPTOMS OR SIGNS

can (but don't always) include:

- inability to whistle;
- inability to sip through a straw;
- eyes that don't close fully during sleep;
- difficulty with sit-ups and pull-ups;
- shoulder blades that "wing" out;
- difficulty raising arm above shoulder height;
- foot drop (foot dorsiflexion weakness);
- difficulty walking, climbing stairs, or rising from a seat;
- falling;
- weak lower abdominal muscles, protuberant abdomen, "Beever's sign";
- curved spine (lordosis).

**Individuals with FSHD, particularly with more advanced or severe cases, can also experience:**

- episodes of "malaise" or "burning pain" in muscles;
- severe pain from changes in posture and strain on remaining muscles;
- chronic fatigue;
- respiratory insufficiency (potentially life threatening);
- symptomatic hearing loss;
- Coats' disease (symptomatic retinal vascular disease), though this is rare.

**Lordosis (sway back) is caused by weakness in the core muscles.**

**Many patients say if there is one thing they wish they could still do, it's to smile.**

**Muscle loss is typically asymmetrical.**

FSHD can also have the following non-muscular manifestations: high-frequency sensorineural hearing loss in both ears, respiratory insufficiency, abnormalities of blood vessels in the back of the eye, and non-symptomatic cardiac arrhythmias.

In more than half of people with FSHD, high-frequency sensorineural hearing loss occurs in both ears; this should be checked in children and adults experiencing hearing difficulties.

Approximately half of FSHD cases also involve abnormalities of blood vessels in the back of the eye, but these lead to visual problems in less than 1 percent of cases. Since these abnormalities are not exclusive to FSHD, one must bear in mind that their presence alone in someone at risk for having FSHD is not sufficient for a diagnosis of FSHD.

Respiratory insufficiency is a more common problem, especially among patients who have become scooter or wheelchair dependent. These patients should have an annual consultation with a pulmonologist to monitor respiratory function and blood carbon dioxide.



## What causes FSHD?

FSHD is genetic in origin, caused by a complex combination of changes in an individual's DNA. It is inherited and is not contagious.

FSHD Type 1 (also called FSHD1, FSHD1A, or FSHMD1A) is the more common form of FSHD, accounting for approximately 95 percent of cases.

FSHD is thought to result from the abnormal expression in muscle of a gene called DUX4. Normally, DUX4 is expressed only in early embryogenesis and in the cells that develop into sperm. But when expressed in muscle, DUX4 appears to be toxic. Other genes may also be involved in the disease process.

The DUX4 gene is encoded in a unit called D4Z4, which is repeated in tandem near the end of chromosome 4 (at a location called 4q35). In unaffected individuals, the D4Z4 array on chromosome 4 is "hypermethylated" (has many methyl molecules attached) and is compacted, preventing DUX4 expression.

In individuals with FSHD Type 1, this D4Z4 repeat array is reduced from a normal range of more than 10 contiguous units to a range of between one and 10 contiguous units. The contraction of the D4Z4 region on chromosome 4 by itself is not sufficient to cause FSHD. Adjacent to the D4Z4 region lies a region that comes in two alleles (variants) called 4qA and 4qB. Only 4qA contains a polyadenylation site allowing a stable production of the DUX4 gene. Individuals who have a 4qB "non-permissive" allele are unaffected, even if the D4Z4 region is shortened. With FSHD, in addition to having 4qA, the contracted D4Z4 region is depleted in methyl groups (is under- or hypomethylated).

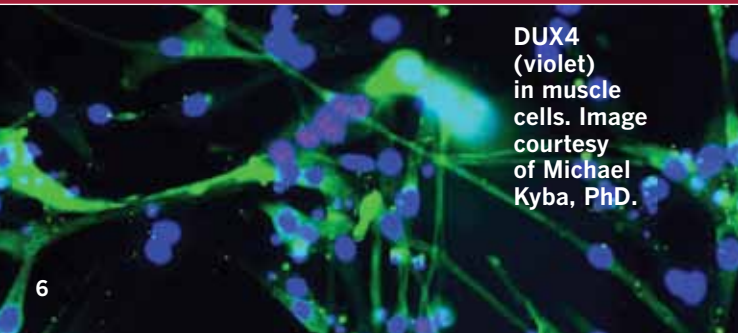
It is the combination of these three factors that results in the full expression of FSHD symptoms.

Researchers have found individuals who have the contracted D4Z4 region together with the 4qA allele but who have no symptoms, or extremely mild symptoms. These "non-manifesting" individuals can pass along FSHD to their children. The difference between being non-manifesting and having FSHD symptoms appears to lie in the degree of methylation of the D4Z4 units. Non-manifesting individuals have several times higher methylation than do individuals with FSHD symptoms, although less methylation than people with a normal number of D4Z4 repeats.

FSHD Type 2 (also called FSHD2, FSHD1B, or FSHMD1B) is the term used to describe the 5 percent of FSHD cases that test negative for FSHD Type 1 (meaning that they are not associated with a loss of D4Z4 repeat units on chromosome 4).

Eighty-five percent of FSHD2 cases are caused by the inheritance of two independent genetic variations: mutation of the Structural Maintenance of Chromosomes flexible Hinge Domain containing 1 gene (SMCHD1) on chromosome 18, combined with having the same "permissive" 4qA allele on chromosome 4 that is associated with FSHD1. Individuals with FSHD2 have extreme loss of methylation of the D4Z4 units. The size of the D4Z4 region of the 4qA (i.e., DUX4-producing) chromosome in FSHD2 is most often between 11 and 16 units, which is at the lower end of the repeat size spectrum of unaffected individuals.

FSHD "Type 3" refers to the 1 percent of cases that lack the FSHD1 and FSHD2 genetic mechanisms, and is an active area of research.



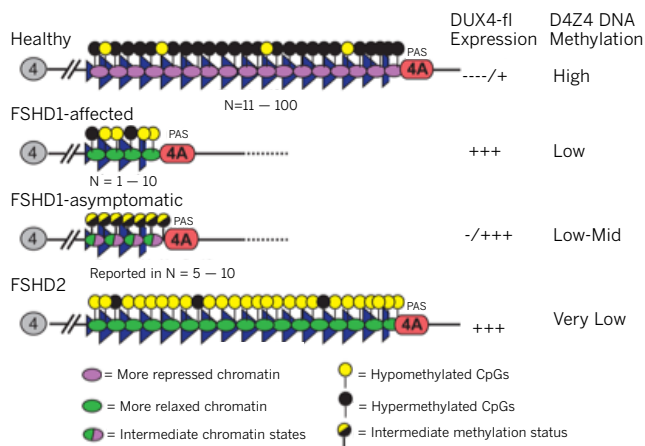
DUX4  
(violet)  
in muscle  
cells. Image  
courtesy  
of Michael  
Kyba, PhD.

## IN SUMMARY:

**Shortened D4Z4 + 4qA + hypomethylation of D4Z4 = FSHD Type 1**  
┌────────── chromosome 4 ─────────┐

**SMCHD1 mutation + 4qA + hypomethylation of D4Z4 = FSHD Type 2**  
┌── chromosome 18 ─┐ ┌── chromosome 4 ─┐

## FSHD is linked to the 4qA subtelomere and the epigenetic status of the 4q35 D4Z4 array



**FIGURE.** The D4Z4 repeat region at location 4q35 on chromosome 4 differs markedly among healthy, FSHD1, and FSHD2 individuals. Healthy individuals have numerous D4Z4 repeats which are highly methylated (black dots). FSHD1-affected individuals have few repeats, and these are hypomethylated (yellow dots). FSHD1 non-manifesting, or unaffected, individuals also have few repeats, but these have higher methylation (half-filled dots). FSHD2 individuals have many D4Z4 repeats, like healthy individuals, but they are severely hypomethylated. Figure courtesy of Peter Jones, PhD.

## How is FSHD diagnosed?

The first step in diagnosing FSHD is a visit with a doctor for a physical exam. An initial diagnosis is based on the pattern of muscles affected. The doctor will ask a series of questions about the patient's family history and medical history.

The doctor may order tests to determine whether the symptoms are a result of FSHD. Tests may also rule out other problems that could cause muscle weakness, such as surgery, toxic exposure, medications, or other diseases. These tests may include the following:

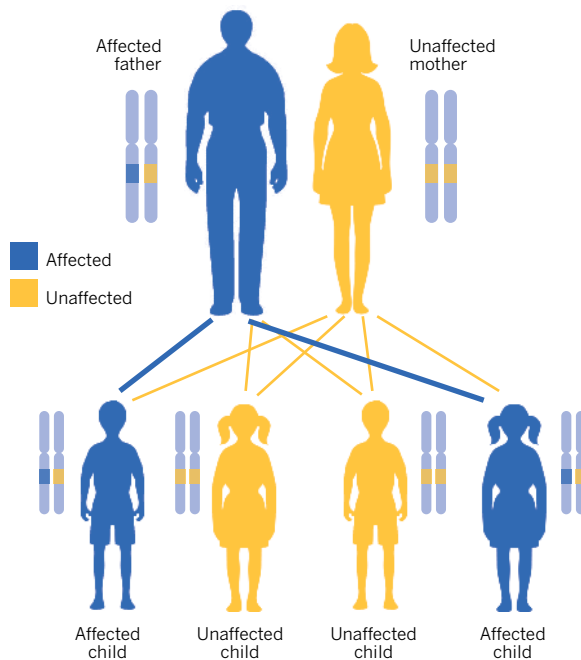
- Blood tests** to measure levels of serum creatine kinase (CK), an enzyme that is released into the bloodstream when muscle fibers are deteriorating, and serum aldolase, an enzyme that helps break down sugars into energy. Elevated levels of either of these enzymes can indicate a problem with muscles and a need for additional testing. However, a normal CK level does not rule out FSHD.
- Neurological tests** including electromyography (EMG) to rule out other nervous system disorders, identify patterns of muscle weakness and wasting, test reflexes and coordination, and detect muscle contractures.
- Muscle biopsies**, which involve the removal of muscle tissue using a biopsy needle or during a simple surgical procedure. The tissue is then examined under a microscope. In FSHD, a muscle biopsy might reveal several abnormalities, but none are uniquely characteristic for the disease, or the muscle might even appear normal. To confirm a diagnosis of FSHD with certainty, a genetic test is needed.
- A genetic test** involves taking a small sample containing the patient's cells (blood, saliva, skin, etc.) and sending it to a specialized laboratory where the DNA is extracted and analyzed. See the next section for further details.



Individuals who test negative for FSHD1 may be referred for testing for FSHD2. The FSHD2 test detects mutations in the SMCHD1 gene on chromosome 18, the presence of the 4qA allele on chromosome 4, and the amount of methylation of the D4Z4 region on chromosome 4.

*FOR MORE INFORMATION on laboratories that offer genetic testing, please refer to the FSHD Society's website ([www.fshdsociety.org/genetic-testing](http://www.fshdsociety.org/genetic-testing)). The Society does not endorse any test or laboratory. Individuals should consult their own physician and genetic counselor about taking the DNA diagnostic test.*

## AUTOSOMAL DOMINANT



*U.S. National Library of Medicine*

## How does genetic testing work for FSHD?

Genetic tests for FSHD1 are commercially available. Genetic testing for FSHD2 is not yet widely available. The tests are highly reliable for most cases. Your doctor can order a small blood sample to be drawn and sent to a testing laboratory. The laboratory extracts DNA for the test from the white blood cells.

The FSHD1 genetic test detects the deletion of D4Z4 repeat units on chromosome 4, described earlier. Although several factors may occasionally complicate the test, confirmation of this deletion is 98 percent reliable as a presumptive diagnosis of FSHD1.

**Every person has 23 pairs of chromosomes (DNA packages) in each cell. Half of the genes in each pair come from the father and half from the mother. In autosomal dominant inheritance, it takes only one copy of a disease-causing gene (blue) to cause a disease. If one parent has a disease-causing gene, each child has a 50% chance of inheriting that gene and having the disease.**

## How does a person inherit FSHD?

About two-thirds of individuals with FSHD inherit the disease from an affected parent. If one parent has the FSHD genetic mechanism, that parent has a 50-50 chance of passing the disease on to each child of either sex.

Seemingly unaffected family members can carry the mutation. This fact was discovered after genetic testing was done on multiple family members after one member was diagnosed with FSHD. It is not known whether these “non-manifesting” individuals will develop symptoms as they grow older. The discovery of non-manifesting cases means that a child could inherit FSHD even if both parents appear to be unaffected, if one parent carries the mutation but does not have symptoms. Only genetic testing of both parents can determine if this is the case.

With several very rare exceptions, if individuals do not have a positive FSHD1 or FSHD2 genetic test, they cannot pass the disease on to their children.

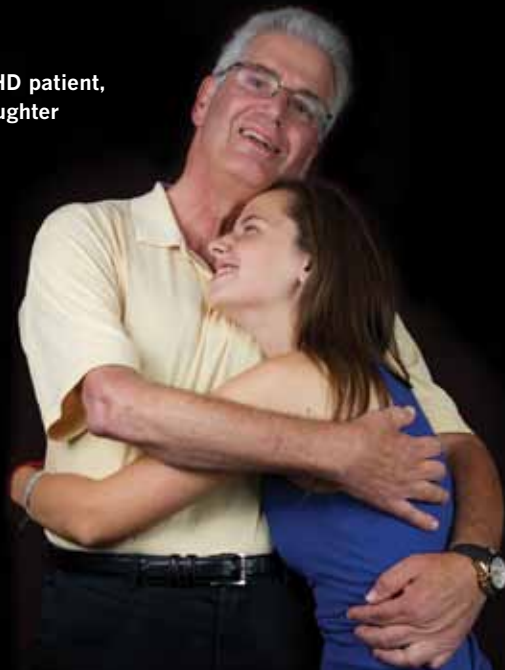
## If a family member has FSHD, could I have the FSHD mutation?

Yes. If you have a biological parent, sibling, or other blood relative who has the FSHD mutation, you have a risk of carrying the mutation, but only if at least one parent has the FSHD gene mutation. The number of D4Z4 repeat units, known as the FSHD deletion size, is stable across generations, so an affected parent will likely pass along the same number of D4Z4 repeats and 4qA haplotype to a child.

Oftentimes, an individual is diagnosed and does not know if a parent has the FSHD genetic mutation. Although a parent may not have signs of FSHD, only a genetic test can confirm whether or not the parent carries FSHD. The parent’s parents may be affected as well, and possibly uncles, aunts, and cousins.

Often, when a person is diagnosed, the disease is discovered to be present throughout the extended family tree and over many generations. It is important to be aware that there may be other family members who are affected but unaware that they may have FSHD or may be

Steven, FSHD patient,  
with his daughter



at risk for it. Professionals with knowledge of genetics and inheritance of FSHD can advise them regarding that risk.

## Should I seek a diagnosis even if I don’t have symptoms?

Adults at risk, even without obvious symptoms, may want to consult a physician or genetic counselor about seeking a diagnosis if they wish for reassurance.

Examinations by clinicians familiar with the disease are quite dependable when they detect an expected pattern of weakening muscles. However, the diagnosis may still be equivocal at younger ages and with some at-risk adults with mild or non-manifesting cases. This uncertainty can occur during years when there are important vocational, marital, and family planning choices to be made.

A genetic test can help alleviate much of this uncertainty. However, the test result does not fully predict how the disease will run its course in an individual. A genetic counselor can help you navigate the decision on whether to be tested and assist you and your family in processing the information from the test results.

## What are sporadic cases of FSHD?

Sporadic FSHD cases are those in which a patient's parents are both unaffected. Studies report that up to a third of FSHD cases are sporadic. Eighty percent of sporadic cases are the result of a new, spontaneous mutation (known as a de novo mutation). Once this mutation appears in an individual, each of his or her offspring has a 50 percent chance of inheriting FSHD.

Approximately 20 percent of reported sporadic cases result from having a seemingly unaffected parent who is a "germline mosaic," meaning that only the mother's or father's germ cells (egg or sperm) have the FSHD mutation, while the rest of the body is genetically normal. Once a child inherits FSHD through a germline mutation, all cells in the child's body carry the FSHD mutation, and the disease can be transmitted to subsequent generations.

## Is there a prenatal test for FSHD?

Yes. Using the same technology as the DNA test described above, prenatal testing is possible. Also, for women who choose to have in vitro fertilization, pre-implantation genetic diagnosis (PGD, DNA testing of embryos) is available.

An individual who is interested in a prenatal test or PGD for FSHD should consult a physician or contact the FSHD Society. The Society can provide further information about this subject.



## How many people have FSHD?

Often-cited figures for the prevalence of FSHD range from one in 14,000 to one in 20,000. However, due to increased experience with FSHD, population-based research, and improved genetic testing, this estimate may be low.

A study published in 2014 found that the prevalence of FSHD is about one in 8,000 in the Dutch population. Applying the Dutch study results to the U.S. population of 320 million and the world population of 7 billion in 2015 yields an estimated 40,000 Americans and 870,000 individuals worldwide with FSHD. Actual prevalence may be even greater because FSHD is frequently undiagnosed or misdiagnosed, and some national health statistics (notably in the U.S.) have not tracked the number of FSHD patients.

FSHD occurs in all racial groups and with equal frequency in both sexes.

## When do symptoms appear?

Although the FSHD genetic mechanism is present from conception, weaknesses are generally not noticeable until the second decade of life. Muscle weakness can be found in most affected individuals by age 20 in males and by age 30 in females. However, in some individuals the symptoms can be so slight that they can go unrecognized well into advanced age. Conversely, in some children symptoms can be quite pronounced and severe from the first few years of life.







Infantile FSHD patients, Vanessa and Alyssa, with their mother

It is also not uncommon for FSHD symptoms to be mistaken for an injury or other disorder such as polymyositis (muscle inflammation). For some people, FSHD is diagnosed fairly quickly, while for others it can take many years from initial symptoms to a confirmed diagnosis. This depends on many factors, including the individual's access to doctors and other healthcare providers knowledgeable about FSHD.

### FSHD in children

In early-onset or infantile FSHD (iFSHD), an infant or child under the age of 5 develops symptoms. About 4 percent of symptomatic FSHD cases are of the infantile type. In iFSHD there are facial weaknesses during the first two years of life in addition to other typical muscle weaknesses of FSHD. Some of these children also have moderate to profound bilateral sensorineural hearing loss and sight-threatening retinal abnormalities (Coats' disease). It is important to routinely check hearing and vision if your child is affected by FSHD.

Early-onset and infantile cases of FSHD often pose special challenges arising from severity of the symptoms, and schooling and socialization issues. The FSHD Society provides helpful information, including a brochure for schools, and coordinates a private Facebook group for parents of children with iFSHD. For a brochure or further information, please contact the FSHD Society.

### What is the prognosis of FSHD?

Predicting the course and outcome of the disease—the prognosis—has its certainties and uncertainties. There is certainty that some skeletal muscles will weaken and waste throughout life and that this can, and often does, cause limitations on personal and occupational activities. FSHD appears not to diminish the intellect. The heart and internal (smooth) muscles are generally spared.

There are uncertainties. The rapidity and extent of muscle loss differ considerably among FSHD patients—even among members of the same family. Some report few difficulties throughout life, while others need a cane, walker, or wheelchair as walking becomes too difficult or impossible. The degree of severity in a parent with FSHD cannot accurately predict the extent of disability that may develop in his or her child.

Muscle and movement are an important part of the full expression of much of life. Often, there are losses difficult to define in clinical terms. The accompanying losses often eclipse the clinically defined symptoms and are a significant part of the FSHD prognosis.

### PROGNOSIS BASED ON GENETIC DIAGNOSIS

NUMBER OF D4Z4 REPEAT UNITS	SEVERITY OF SYMPTOMS
1 – 3	Severe. One unit is generally early onset.
4 – 6	Moderate.
7 – 10	Mild, with prognosis dependent on the degree of D4Z4 methylation.
Normal units + FSHD2	Prognosis is dependent on the degree of D4Z4 methylation.



**Many FSHD patients, like Haviva, find ankle-foot orthoses to be beneficial for foot drop.**

## **Are treatments and aids available for FSHD?**

There is no treatment or cure yet for FSHD. There are, however, things that can be done to alleviate its effects, including meeting with knowledgeable health practitioners.

Neurologists are often the primary physicians in muscle disease clinics, since muscles do their work through stimulation by nerves. If your primary care doctor notices muscle weakness, he or she should refer you to a neurologist who specializes in muscle diseases.

Physiatrists are physicians who work with chronic neuromuscular conditions. Periodic visits with a neurologist or physiatrist are useful to monitor the progress of FSHD and to obtain referrals to other professionals and services.

An orthopedist (a physician concerned with the skeletal system and associated muscles, joints, and ligaments) can offer advice about mobility issues and other functional problems of the muscular/skeletal system.

Physical therapy, including light exercise, helps preserve flexibility. Swimming is especially helpful by making many movements easier. One should stay as active as possible, with rest breaks as needed during exercise and activities.

Moderate aerobic exercise combined with cognitive behavioral therapy has been shown in a clinical trial to reduce chronic fatigue in FSHD patients.

Physical and occupational therapists can help with suggestions for adaptations and physical aids that can often partially free an FSHD patient from some limitations of the disease. Foot drop can sometimes be managed with ankle-foot orthotics (AFOs) and knee-ankle-foot orthotics (KAFOs).

Patients may resist adaptations and aids, feeling that they are “giving in” to FSHD by using them, but these devices reduce the risk of falls and serious injuries that can lead to permanent loss of mobility. A cane or walking stick can be very helpful in avoiding falls and alerting others that you are at risk for falling. Adaptations and physical aids help to extend, rather than end, mobility and independence.

Many patients report significant pain, although others are spared. No specific treatments are available. Gentle stretches in the morning can alleviate pain from cramped muscles. Pain medication and mild physiotherapy are often prescribed with moderate results. Some patients have found significant relief through acupuncture. Relaxation and managing stress are reported to help with many chronic pain conditions.

Dietitians can help maintain a healthy diet and avoid unnecessary weight to reduce stress on already weakened muscles.



**FSHD patient Len wears a neck brace to compensate for weakened neck muscles.**



**This patient has had scapular fixation surgery on his right side, which restored his ability to raise his right arm. On the left side his scapula “wings” out and he is unable to raise the left arm above shoulder height. Photo courtesy of Leigh Ann Curl, MD.**

Speech and hearing therapists can help with limitations imposed by hearing loss and weakened facial musculature to improve speech and communication.

Surgery to attach the scapula (shoulder blade) to the ribcage can improve motion of the arms or relieve pain. Surgical methods to address foot drop and facial muscle weakness are also being developed. Only some patients are suitable candidates for surgery. Individuals who are considering such surgery should consult with their neurologist or physiatrist and an orthopedic surgeon (for scapular and leg surgery) or reconstructive plastic surgeon (for facial surgery). Choose only surgeons who have experience with the procedure and a deep understanding of FSHD and the demands of post-surgical physical rehabilitation. It is essential to discuss these procedures with individuals who have undergone the surgery.

*THE FSHD SOCIETY provides referrals to physicians and other professionals, as well as to fellow patients who are willing to discuss their experiences.*

*FOR MORE INFORMATION and to download our brochure on physical therapy and exercise, visit the “Understanding FSHD/Resources” section of our website.*

## **Can respiratory insufficiency occur in FSHD?**

Yes. Respiratory involvement can occur. Patients with moderate to severe FSHD should have their respiratory function evaluated during periodic clinic visits. Regular

monitoring of respiratory function is suggested, as one might experience insufficiency over a long period of time without presenting signs.

It's important to be aware that respiratory compensatory mechanisms can allow one to adapt to functioning with high levels of carbon dioxide (CO<sub>2</sub>) in the blood—at levels doctors would not expect to permit normal function. This is known as hypercarbia and is dangerous in the long term.

Discuss breathing tests with your doctor if you experience any of the following:

- Never feeling rested even after a good night's sleep.
- Morning headaches.
- Snoring loudly or in a different pattern than usual.
- Labored and interrupted breathing while lying down.
- Fatigue and daytime sleepiness.

The tests should include forced vital capacity and nocturnal oximetry tests. These are easy, non-invasive tests that don't require a hospital stay. Because many doctors, even experienced neurologists, don't associate FSHD with respiratory problems, your doctor may be reluctant to order respiratory tests. Insist on respiratory tests if you feel the symptoms described above.

Respiratory insufficiency can initially be managed with nighttime non-invasive pressure support, typically a BiPAP (Bi-level Positive Airway Pressure) machine. BiPAP or similar mechanical ventilation at night can increase oxygen and greatly improve sleep and energy level.

In more advanced or acute cases, patients may require the use of a volume-control and pressure-control ventilator for invasive and non-invasive ventilation.

You should be tested periodically to ensure that the settings on the machine are appropriate. Your doctor should consult a respiratory therapist (RT) as early as possible, and the RT should remain involved in monitoring your progress.

For FSHD patients with respiratory insufficiency, in standard practice, trauma (ER, ICU), surgery, and anesthesiology settings, care should be taken not to suppress respiratory drive with narcotics unless it is a

situation of palliative care. If narcotics are necessary for pain control, it is very important that you notify the emergency responders or doctors about FSHD and any respiratory problems you might have or be at risk for. Carry a medical alert card with you at all times.

Oxygen supplementation can be detrimental to patients with undetected or mismanaged hypercarbic (high CO<sub>2</sub>) respiratory failure and lead to worsening CO<sub>2</sub> levels. Oxygen should generally not be administered unless BiPAP (Bi-level Positive Airway Pressure) or similar ventilatory support is also being used. Your physician and a pulmonologist can help you periodically monitor CO<sub>2</sub> levels in the office or pulmonary function lab in the hospital.

Along with respiratory care, it is important to know that in FSHD, cardiac studies show that cardiac arrhythmias and right bundle branch block (RBBB) can occur without cardiac symptoms and when echocardiography is usually normal. That said, individuals at risk for respiratory insufficiency or respiratory failure should talk with a physician about monitoring for symptoms of pulmonary hypertension and congestive heart failure.

## 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.

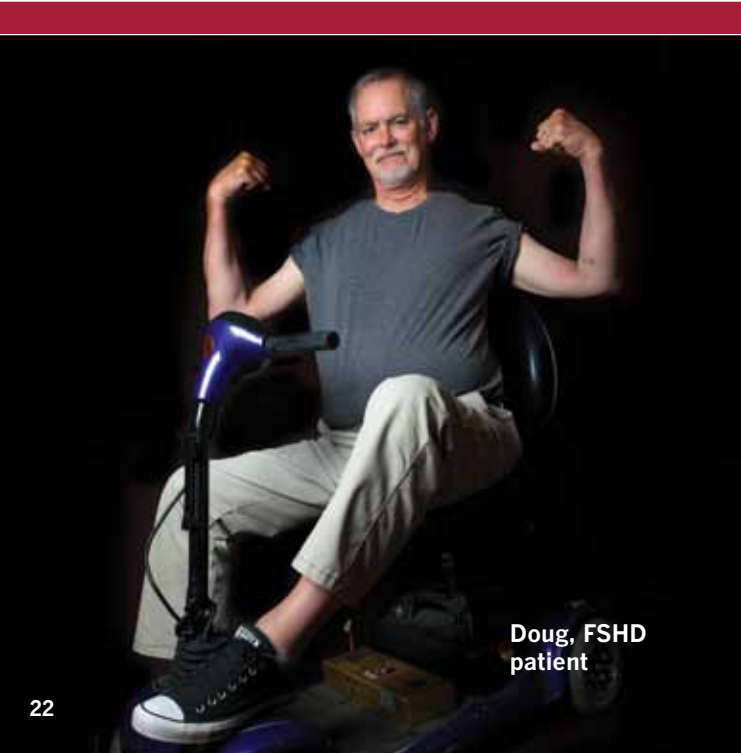
For more information or to support the work of the FSHD Society, visit [www.fshdsociety.org](http://www.fshdsociety.org).

## Or contact:

FSHD Society  
450 Bedford Street  
Lexington, MA 02420  
Phone: (781) 301-6060  
Email: [info@fshdsociety.org](mailto:info@fshdsociety.org)



*The FSHD Society is an independent 501(c)(3) non-profit and tax-exempt organization. It is the top-rated muscular dystrophy charity in the U.S. and has been named one of America's "Ten Charities Worth Watching" for outstanding performance. Copyright FSHD Society, 2019. We thank Howard Chabner, George Padberg, MD, and Jeffrey Statland, MD, for reviewing this brochure.*



## **FSHD Society, Inc.**

450 Bedford Street  
Lexington, MA 02420  
(781) 301-6060  
(781) 862-1116 (fax)  
[www.fshdsociety.org](http://www.fshdsociety.org)  
[info@fshdsociety.org](mailto:info@fshdsociety.org)



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LIGHTING THE WAY TO A CURE