

What is...

Facioscapulohumeral Muscular Dystrophy

Facioscapulohumeral muscular dystrophy (FSHD) is a **genetic muscle disorder** in which the muscles of the face, shoulder blades, and upper arms are among the most affected.

Researchers have described two types of facioscapulohumeral muscular dystrophy: **type 1 (FSHD1)** and **type 2 (FSHD2)**. The two types typically have the same signs and symptoms and are distinguished by their genetic cause.

FSHD1 is caused by changes in a region of chromosome 4 called **D4Z4** that result in the abnormal activation of a gene called **DUX4**. Mutations in a gene called **SMCHD1** are the underlying cause of FSHD2.

FSHD has an estimated prevalence of **1 in 20,000** people. About **95 percent** of all cases are FSHD1; the remaining **5 percent** are FSHD2.

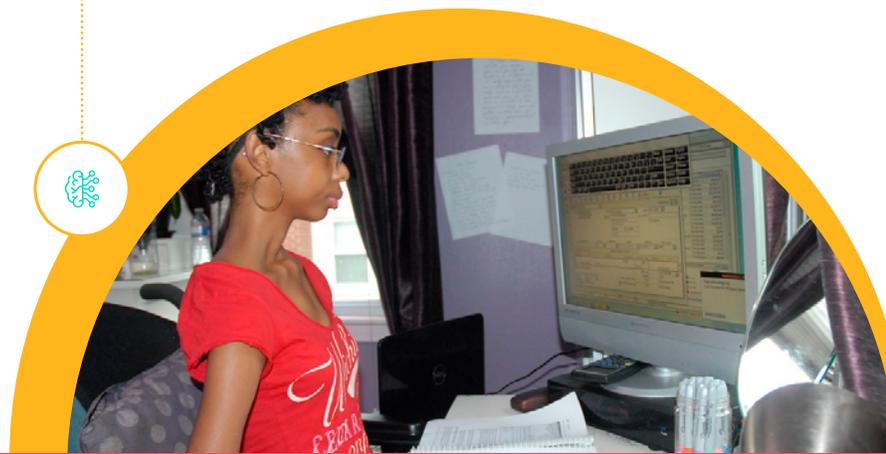
FSHD rarely affects the heart or respiratory system. It doesn't cause learning disabilities or other cognitive impairments, nor does it affect sensation, ability to control the bladder and bowels, or sexual function.

FSHD usually begins **before age 20**, but it can begin **as early as infancy** and **as late as the 50s**.

In most people with FSHD, **the disease progresses very slowly**, and most people affected by the disease have a **normal lifespan**.

Initial symptoms include **weakness and atrophy** of the muscles around the eyes and mouth, shoulders, upper arms, and lower legs. Later, weakness can spread to abdominal muscles and sometimes hip muscles.

There is no cure for FSHD, but medications and therapy can help manage some symptoms and potentially slow the course of the disease.



What are the symptoms of FSHD?

FSHD mainly affects skeletal muscles, along with vision and hearing. Rarely, the heart or lungs may be affected.



What should I know about FSHD?

- 1 The age of disease onset, progression, and severity of FSHD vary a great deal.
- 2 Usually, symptoms develop during the teen years, with most people noticing some problems by age 20, although weakness in some muscles can begin as early as infancy and as late as the 50s.
- 3 Weakness involving the facial muscles or shoulders is usually the first symptom, with facial muscle weakness often making it difficult to drink from a straw, whistle, or smile.
- 4 Weakness in muscles around the eyes can prevent the eyes from closing fully while a person is asleep, which can lead to dry eyes and other eye problems.
- 5 Weak shoulder muscles tend to make the shoulder blades (scapulae) protrude from the back, a common sign known as scapular winging. Weakness in the muscles of the shoulders and upper arms can make it difficult to raise the arms over the head or throw a ball.
- 6 Weakness in the muscles of the lower legs can lead to a condition called foot drop, which affects walking and increases the risk of falls.
- 7 When muscle weakness is prolonged, it can lead to freezing of joints in one position, called a contracture. In FSHD, contractures are most likely to occur in the ankles.
- 8 Muscular weakness in the hips and pelvis can make it difficult to climb stairs or walk long distances.
- 9 When the muscles surrounding the spine weaken, the column is pulled out of alignment. The misalignment often takes the form of lordosis, where the spine curves in to an excessive degree and the stomach sticks out. It also can take the form of scoliosis, in which the spine curves to the side, like an S. The scoliosis that sometimes occurs in FSHD usually isn't severe.
- 10 Additional signs and symptoms of FSHD can include mild high-tone hearing loss. When FSHD starts in childhood, loss of hearing may be more profound than in adult-onset FSHD.
- 11 Abnormalities involving the light-sensitive tissue at the back of the eye (the retina) may occur. In addition, eye muscle weakness can keep the eyes from closing completely during the night, causing dryness and injury.
- 12 Inflammation of muscles — an attack by certain types of cells of the immune system — occurs in some muscular dystrophies and can be extensive in some people with FSHD.
- 13 In most people with FSHD, weakness may be asymmetrical, differing between the left and right sides of the body.
- 14 Pain in FSHD may also result from the way weakened muscles pull bony structures, such as the spine and shoulder blades, out of alignment.
- 15 Rarely, facioscapulohumeral muscular dystrophy may affect the heart (cardiac) muscle or muscles needed for breathing.

How is FSHD treated?

No treatment currently exists to halt or reverse the effects of FSHD, but there are treatments and devices that can help alleviate many of the symptoms.

Physical therapy may help to retain muscle strength and function, enhance mobility, and help prevent falls.

Occupational and speech therapy can help maintain daily living skills.

Low-intensity aerobic exercise may be recommended to help maintain mobility. Any exercise regimen should be initiated under the guidance of a physician and customized to accommodate the individual's disease symptoms, age, and cardiovascular status.

Anti-inflammatory drugs known as nonsteroidal anti-inflammatories, or NSAIDs, are often prescribed to improve comfort and mobility.

Massage or warm, moist heat may help with the discomfort associated with FSHD.

Ankle/foot orthoses can improve mobility and prevent falls in individuals with foot drop.

Devices such as **back supports, corsets, girdles, and special bras** for people with FSHD can help compensate for weakening muscles in the upper and lower back.

Surgical fixation of the scapula to the chest wall may improve range of motion in the arms.

Standard therapies, including **amplification**, are appropriate treatments for hearing loss.

Ventilatory support such as bi-level positive airway pressure (BiPAP) may be necessary for those who experience hypoventilation.

Use of lubricants can help prevent drying of the eyes in individuals whose muscle weakness causes them to sleep with their eyes partially open. In some cases, use of an eye shield or patching the eyes during sleep may be necessary to alleviate dryness.

Please talk to your medical provider to obtain more information on these treatments.



MDA Glossary

Atrophy

A decrease in the size and mass of muscle tissue

Contracture

A shortening of muscles or tendons around joints that can limit mobility

Exposure keratitis

Dryness of the cornea caused by an inability to effectively close the eyelids

Hypoventilation

When breathing is too shallow or slow to meet the body's needs, resulting in an increase in carbon dioxide levels

Lordosis

An abnormal exaggerated curve in the lower (lumbar) region of the spine

Muscular dystrophy

A term that refers to a number of diseases that cause progressive loss of muscle mass, resulting in weakness and, sometimes, loss of mobility

Mutation

A flaw in the DNA code

Scapula

Shoulder bone, or shoulder blade

Scapular winging

When the muscles that hold the shoulder blades in place weaken, the shoulder blades stick out and rise up toward the neck as they move; the protruding bone resembles a wing

Scoliosis

A curvature in the spine that occurs when weakened muscles are unable to hold the spine straight

Read more about FSHD at mda.org. If you're looking for one-on-one support, give our MDA Resource Center a call at 800-572-1717.



Designated a Top-Rated Charity by the American Institute of Philanthropy, MDA is the first nonprofit to receive a Lifetime Achievement Award from the American Medical Association for "significant and lasting contributions to the health and welfare of humanity."



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