

# 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 first 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 the **DUX4 gene**. **FSHD2** is caused by mutations in the **SMCHD1** gene on chromosome 8 that activate the **DUX4** gene.

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

FSHD affects the lungs in about 15% of individuals. The heart is rarely affected. 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**.



Often, the initial symptoms include weakness and atrophy of the muscles around the eyes and mouth, shoulders, upper arms, and lower legs. Weakness can spread to the abdominal and back muscles as well as the hips and upper legs.

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

# WHAT SHOULD I KNOW ABOUT FSHD?

- 1** The age of disease onset, progression, and severity of FSHD vary greatly.
- 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 the muscles around the eyes can prevent the eyes from closing fully while a person is asleep, leading 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 shoulders and upper arm muscles can make it difficult to raise the arms over the head or throw a ball.
- 6** Weakness in the muscles of the lower legs leads to foot drop, which affects walking and increases the risk of falls.
- 7** Prolonged muscle weakness can lead to a joint freezing in one position, called a contracture. Contractures are rare in FSHD, but when they occur, they are typically 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 can cause scoliosis, where the spine curves to the side. When the muscles in the back are weak, individuals have difficulty standing straight and the spine curves inward at the lower back, known as lordosis. Both scoliosis and lordosis can be mild in FSHD.
- 10** Additional signs and symptoms of FSHD can include mild to moderate high-tone hearing loss, mostly in childhood-onset FSHD. Loss of hearing in childhood-onset is typically more severe than in adult-onset FSHD.
- 11** Abnormalities involving the light-sensitive tissue at the back of the eye (the retina) may occur. In addition, eyelid muscle weakness can keep the eyes from closing, causing dryness and injury.
- 12** Muscle inflammation occurs in several forms of muscular dystrophy, including FSHD. This inflammation results in loss of muscle mass and strength.
- 13** In 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 weakened muscles pulling bony structures, such as the spine and shoulder blades, out of alignment.
- 15** Rarely, FSHD 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 some treatments and devices can help alleviate many of the symptoms.

**Physical therapy** may help to retain muscle strength and function, enhance mobility, and 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 upper and lower back muscles.

**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 with reduced lung capacity.

**Lubricants** can help prevent dry eyes in individuals whose muscle weakness causes them to sleep with their eyes partially open. In some cases, using an eye shield or eye patches during sleep may be necessary to alleviate dryness.

# WHAT ARE THE SYMPTOMS OF FSHD?

FSHD mainly affects the skeletal muscles. In some people with childhood-onset FSHD, it can affect vision and hearing. About 15% of people with FSHD have lung involvement, and it is rare to have heart involvement.

## Sensory perception

- Abnormalities of the retina
- Exposure keratitis
- Mild hearing loss



## Skeleton and muscle

- Muscle weakness
- Muscle atrophy
- Pain
- Inflammation
- Contractures
- Scoliosis
- Lordosis

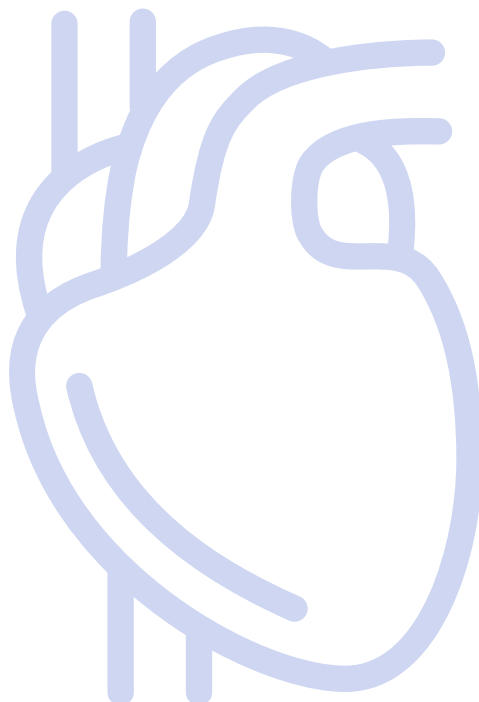


## Heart

- EKG abnormalities, rarely symptoms

## Respiratory system

- Reduced lung capacity



# 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**

Posture characterized by an inward curving of the lower back

## **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**

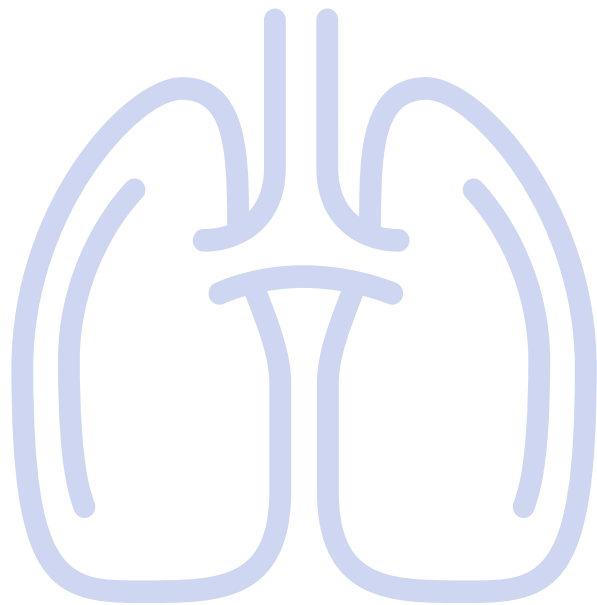
Shoulder bone, or shoulder blade

## **Scapular winging**

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

## **Scoliosis**

An abnormal sideways curvature in the spine that occurs when weakened muscles are unable to hold the spine straight





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