

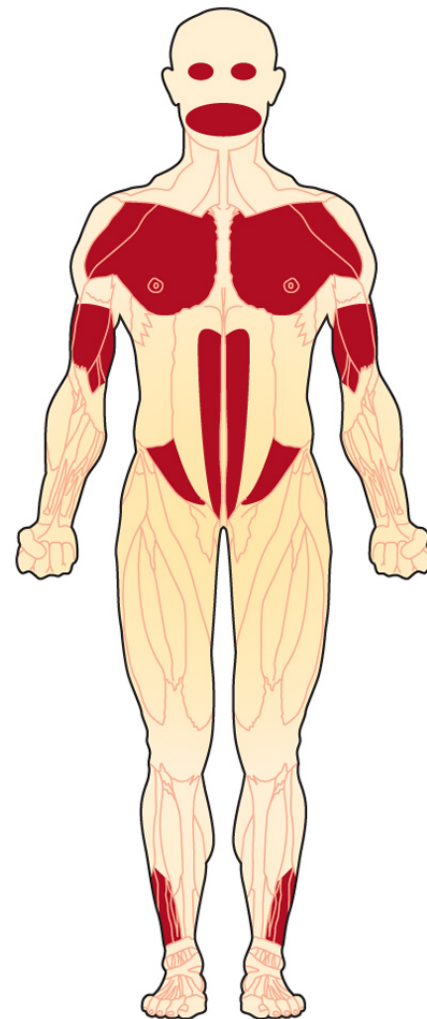
# Facioscapulohumeral Muscular Dystrophy (FSH, FSHD)

## 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.

The long name comes from *facies*, the Latin word and medical term for face; *scapula*, the Latin word and anatomical term for shoulder blade; and *humerus*, the Latin word for upper arm and the anatomical term for the bone that goes from the shoulder to the elbow.

The term *muscular dystrophy* means progressive muscle degeneration, with increasing weakness and atrophy (loss of bulk) of muscles. In FSHD, weakness first and most seriously affects the face, shoulders, and upper arms, but the disease usually also causes weakness in other muscles. FSHD is the third most common type of muscular dystrophy, behind Duchenne and Becker muscular dystrophies and myotonic dystrophy. The estimated prevalence of FSHD about 4 cases per 100,000 individuals.<sup>1,2,3,4,5</sup>



## What are the symptoms of FSHD?

In around 90% of FSHD patients, symptoms usually begin before age 20, with weakness and atrophy of the muscles around the eyes and mouth, shoulders, abdominal muscles, upper arms, and lower legs, usually with asymmetric involvement.

Some experts divide FSHD into adult-onset and infantile-onset forms. The adult-onset (which includes FSHD that begins in adolescence) is far more common. In either type of FSHD, facial weakness can start in childhood. Occasionally, other FSHD symptoms appear in early childhood.

Infantile-onset FSHD generally runs a more pronounced course with regard to muscle weakness, and sometimes also affects hearing and vision. Preliminary evidence suggests that the infantile-onset form is associated with a larger piece of missing DNA. For more, see [Signs and Symptoms \(/disease/facioscapulohumeral-muscular-dystrophy/signs-and-symptoms\)](https://disease/facioscapulohumeral-muscular-dystrophy/signs-and-symptoms).

## What causes FSHD?

FSHD may be inherited through either the father or the mother, or it may occur without a family history. The most probable cause of FSHD is a genetic flaw (mutation) that leads to inappropriate expression of the double homeobox protein 4 gene (*DUX4*) on chromosome 4, in the 4q35 region.<sup>6</sup> The segment is not part of any particular gene, but it nevertheless seems to interfere with the correct processing of genetic material.

For more about the genetic causes of FSHD, see [Causes/Inheritance \(/disease/facioscapulohumeral-muscular-dystrophy/causes-inheritance\)](https://disease/facioscapulohumeral-muscular-dystrophy/causes-inheritance). For more about how the missing chromosome 4 segment may cause FSHD, see [Research \(/disease/facioscapulohumeral-muscular-dystrophy/research\)](https://disease/facioscapulohumeral-muscular-dystrophy/research).

## What is the progression of FSHD?

FSHD usually progresses very slowly and rarely affects the heart or respiratory system. Most people with the disease have a normal life span. However, disease severity is highly variable.

## What is the status of research in FSHD?

In 2009, MDA-supported researchers found that pieces of a gene called *DUX4* are abnormally activated in FSHD-affected cells, leading to production of potentially toxic proteins. Blocking the erroneously activated genes or the proteins made from them seems a likely pathway for treatment. For more, see [Research \(/disease/facioscapulohumeral-muscular-dystrophy/research\)](https://disease/facioscapulohumeral-muscular-dystrophy/research).

Richard E. donated \$50 🙏  
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