

Facioscapulohumeral Muscular Dystrophy (FSHD) Fact Sheet

What is FSHD?

- FSHD is one of the most prevalent of the nine primary types of muscular dystrophy affecting adults and children.
- It affects approximately one in 8,333 people around the world, or over 870,000 worldwide. The actual frequency may be significantly higher due to undiagnosed cases.

What are the symptoms?

- FSHD causes a progressive loss of all skeletal muscle. Weakness is usually noticeable starting with facial, scapular/back, and upper arm muscles.
- Weakness in facial muscles is a hallmark of FSHD—early symptoms can include difficulty whistling or smiling and eyes not fully closing during sleep.
- Loss of muscular strength limits both personal and occupational activities. Ninety-five percent of patients develop noticeable muscle weakness by the age of 20. Approximately 20 percent of patients become unable to walk.
- Respiratory insufficiency, which can be life threatening, is also a symptom.

Who is affected?

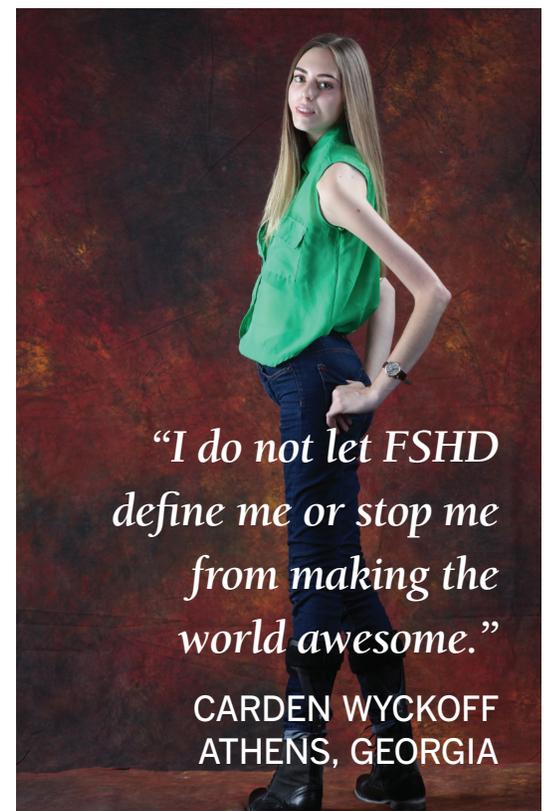
- FSHD occurs with equal frequency in both males and females and can affect children and adults of all ages and all racial groups.
- An affected parent has a 50 percent chance of passing the genetic defect to each child. The majority of FSHD cases are caused by a genetic deletion on chromosome 4.
- The age of onset is variable, as is the eventual extent and degree of muscle loss.
- Every person has the DUX4 gene that leads to FSHD. Usually, the gene is “bottled up” so it can’t cause harm, but when the bottle “breaks,” FSHD results.

- Thirty percent of new FSHD patients have no prior family history and are a result of a spontaneous genetic change. In this sense, every person has a risk of having a child with FSHD.

What are the treatments?

- Currently, there is no treatment to slow down or cure FSHD.
- Low-intensity aerobic exercise appears to be safe and potentially beneficial. This should be done under the supervision of a physical therapist.
- Genetic diagnostic and prenatal diagnostic tests are available for FSHD.
- Researchers hope to develop new drugs for FSHD over the next 3-5 years. There is hope!

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*“I do not let FSHD
define me or stop me
from making the
world awesome.”*

**CARDEN WYCKOFF
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Early-Onset Facioscapulohumeral Muscular Dystrophy (FSHD) Fact Sheet

What is early-onset FSHD?

- FSH muscular dystrophy (FSHD) is a genetic degenerative muscle disease with a wide range of severity and age of onset. When weakness is evident from birth or in early childhood, it is sometimes termed infantile FSHD.
- Early-onset FSHD is estimated to affect approximately one in 200,000 people, or about 30,000 individuals worldwide.
- FSHD occurs with equal frequency in males and females in all racial groups.

What are the symptoms?

- FSHD causes progressive wasting of skeletal muscle, often first noticed in facial, shoulder, back, and upper arm muscles, and can progress to any skeletal muscle.
- Early-onset FSHD is generally associated with early decline in standing and walking.
- Affected children may have low muscle tone, delayed development, and facial weakness leading to excessive drooling, impaired speech, and feeding difficulties.
- Orthopedic issues such as progressive curvature of the spine and winging of the shoulder blades are also more common and can lead to chronic pain and fatigue.
- Infantile FSHD can involve visual problems due to abnormal blood vessels in the eyes, and progressive sensorineural hearing loss. Less commonly, epilepsy and learning problems have been reported.
- Respiratory insufficiency may result from a combination of factors including muscle weakness and skeletal deformities. Early monitoring of heart and lung function is important to help with diagnosis and treatment, especially in case of any breathing concerns.

What causes FSHD?

- The majority (95 percent) of FSHD cases are caused by a genetic deletion on chromosome 4. Infantile FSHD is associated

with a smaller residual DNA fragment remaining from the deletion.

- Every person has the DUX4 gene associated with FSHD. Normally, the gene is suppressed, but the chromosome 4 deletion permits DUX4 to be expressed. DUX4 is thought to harm muscle. Other genes may also be involved.
- If a parent has FSHD, each child has a 50 percent chance of also having FSHD. However, in 20-30 percent of cases, FSHD is not inherited but results from a spontaneous mutation. In this sense, we all have a risk of having a child with FSHD.

What are the treatments?

- Currently, there is no treatment to slow down or cure FSHD. Early diagnosis is important, as interventions exist to address hearing loss and speech impairments, and possibly to prevent blindness.
- Thanks to recent scientific breakthroughs, researchers are optimistic that new drugs will begin to be tested over the next few years. There is hope!

