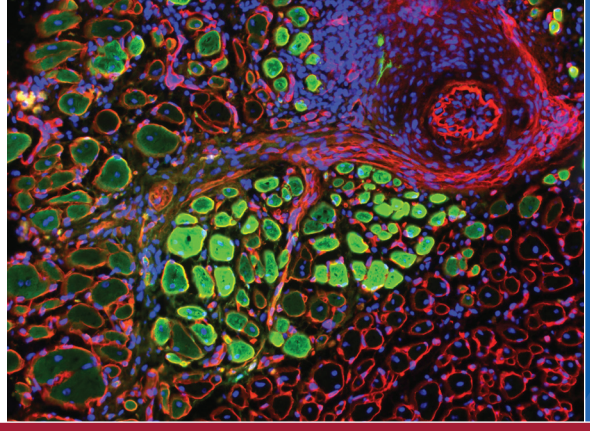
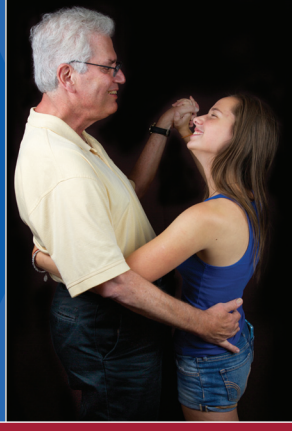
FSH SOCIETY
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY





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Supporting research and patient outreach for Facioscapulohumeral Muscular Dystrophy (FSHD), the most prevalent form of muscular dystrophy. Progress on FSHD could benefit a wide range of other areas of medicine, from cancer and diabetes to muscle regeneration and repair.

Who We Are

Founded in 1991 by patients, the FSH Society is the world's largest grassroots network of facioscapulohumeral muscular dystrophy (FSHD) patients, their families, and research activists. FSHD is the most common genetic myopathy affecting men, women, and children. Marked by progressive degeneration of skeletal muscle, it typically manifests in the face (facio), shoulders (scapula), and upper arms (humerus), as well as the legs, and can spread to any muscle, leading to profound disability. An estimated 870,000 people suffer from FSHD worldwide. About 30 percent of cases arise spontaneously in families with no prior history.

What We Do

We help patients and families through education and outreach; fund scientific research leading to treatments, guided by our world-class scientific advisory board; and advocate for increased government and industry funding for research and to encourage drug trials.

What We Have Accomplished

We established an FSHD research program when none existed, by recruiting scientists and persuading the federal government to allocate funding for FSHD research. Our efforts have resulted in:

- passage of the MD-CARE Act mandating federal action on all muscular dystrophies;
- the discovery of the genetic causes of FSHD;
- understanding of how the genetic defects lead to expression of a toxic gene;
- high-throughput drug screening to identify potential treatments;
- the development of FSHD animal models;
- induced pluripotent stem cell lines for drug screening and gene therapy;
- applications of genomic engineering aimed at blocking the genetic defect in FSHD;
- the development of biomarkers, imaging markers, and clinical trial endpoints.

Our Promise

As long as we are here, no patient need ever face this disease alone. And with generous donations from patients, family, friends, major donors, and sponsors, the FSH Society will keep working to accelerate research leading to treatments.

You Can Help

Make a gift to the FSH Society today! Thank you.

*The FSH Society is an independent 501(c)(3) non-profit and tax-exempt organization. It has earned its **eighth** consecutive Charity Navigator 4-star award and been named one of America's "**Exceptional Charities**" for outstanding performance.*

CHARITY NAVIGATOR
★★★★
Four Star Charity

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FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

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?

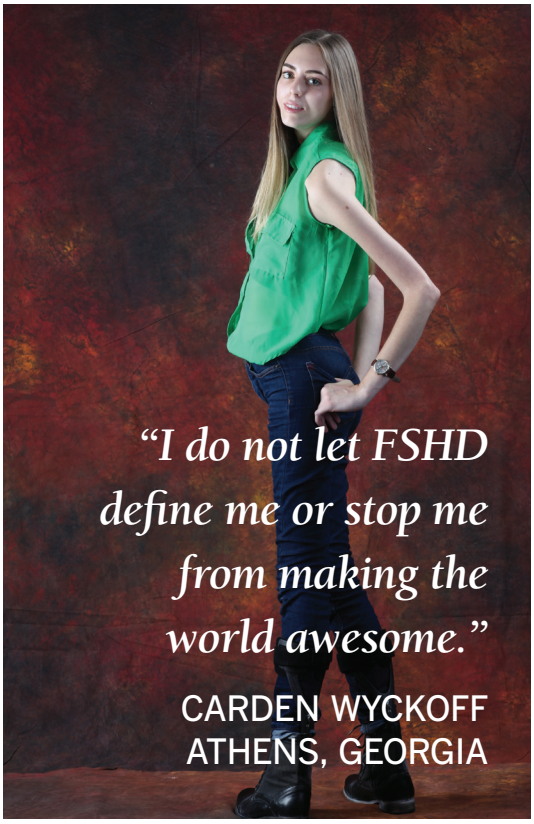
- FSHD causes a progressive loss of all skeletal muscle. Weakness is usually noticeable starting with facial, scapular/back, and upper arm muscles.
- Weakness in facial muscles is a hallmark of FSHD—early symptoms can include difficulty whistling or smiling and eyes not fully closing during sleep.
- 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 apprears 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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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!

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