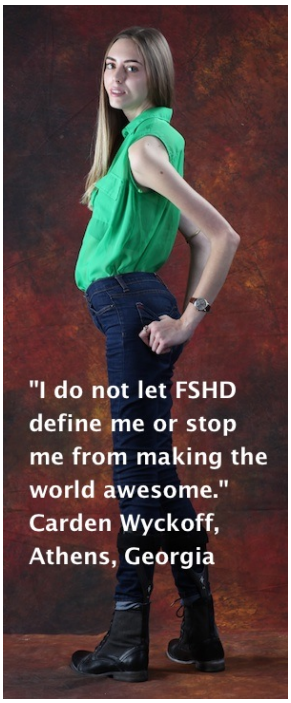
Facioscapulohumeral Muscular Dystrophy (FSHD) Fact Sheet



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 1 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. 95% of patients develop noticeable muscle weakness by the age of 20. Approximately 20% 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% chance of passing the genetic defect to each child. The majority of cases of FSHD 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.
- 30% 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!