

Early-onset Facioscapulohumeral Muscular Dystrophy 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 (also called infantile FSHD or iFSHD) is estimated to affect approximately 1 in 200,000 people, or around 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%) of cases of FSHD 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% chance of also having FSHD. However, in 20-30% 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!