Muscular Dystrophy Canada
let's make muscles move

Facioscapulohumeral Dystrophy (FSHD)

What are the symptoms of facioscapulohumeral dystrophy?

As the name of the disorder suggests, symptoms often first affect the face (facio is Latin for face), shoulders (scapulo), and upper arms (humerus). These symptoms are known as "classic" or "Landouzy-Dejerine" FSHD symptoms. Progressive weakening of the muscles in these areas is a major part of this disorder, though weakness is not limited to these muscles. Muscle weakening is not always equally balanced on the left and right sides of the body. The reason for this unbalanced weakening is currently unknown. FSHD is progressive, which means the symptoms worsen over time. Most people first experience muscle weakness in their teens. The disorder may gradually limit their personal or occupational activities as they enter middle age and beyond. Weakness around the eyes and mouth are often some of the first symptoms of FSHD. People living with FSHD may be unable to purse or pucker their lips, or turn up the corners of their mouth when smiling. Eye weakness is also evident, as someone with FSHD may not be able to close their eyelids tightly, leading to dryness and eye problems.

What is facioscapulohumeral dystrophy?

Facioscapulohumeral [fah - see - oh - ska - pew - low - hue - murr - all] dystrophy (FSHD) is a disorder affecting the skeletal muscle, and is the third most common dystrophy after Duchenne and Myotonic dystrophies. It is an autosomal dominant disorder and is currently classified as two distinct types, 1A and 1B. FSHD occurs in the Canadian population at an estimated frequency of about one in twenty thousand. It affects males and females.

The area around the shoulder blades is one of the primary areas of muscle weakness in FSHD. There is a gradual loss of stability around the shoulders. The shoulder blades, which would otherwise be fixed in place, may not get the leverage necessary to lift or pull. These bones (scapula) may lift or "wing" as they move. Early observable symptoms include inability to throw objects or lift the arms over the head.

There may be an unequal weakening of the biceps, triceps, deltoids, and lower arm muscles giving an unbalanced visual effect. As well, abdominal and hip muscles can weaken leading to an exaggerated curvature of the lower spine. It should be noted, however, that weakness could occur over the entire body as the disorder progresses. Involvement of muscles of the foot, ankle, hips and abdomen are common. These muscle losses frequently result in distorted gait patterns, increased incidence of falls and stress on remaining functioning muscle. This may lead to pain, inflammation and joint and spine problems. Measured or limited activity is important to maintain range of motion and strength of remaining

functioning muscle. Use of orthotic braces as well as an individual physiotherapy routine may help to limit stress on healthy muscle. Periods of muscle inactivity can rapidly reduce the ability to use those muscles in the future. Occasionally mobility is affected to the extent that a wheelchair is required. Muscles of the hands and wrists may also be affected.

Other rare symptoms include hearing loss, visual impairment, difficulty swallowing and problems with respiration, in particular, carbon dioxide retention during sleep. Most people with FSHD live a normal lifespan. There has been no documented link of FSHD to learning disabilities or mental impairment.

The symptoms listed above describe the classic forms of FSHD. As researchers often note: "The clinical presentations in patients with FSHD associated short fragments on chromosome 4q35 are not restricted to the classic FSHD form, but constitute a variety of clinical manifestations." (Krasnianski M, Arch Neurol. 2003 Oct; 60(10): 1421-5.) In other words, like many disorders, FSHD can appear in ways that are unique or unexpected. How rapidly the disability progresses and the extent of muscle loss differs considerably, even among family members. The degree of severity in a parent can not predict the extent to which the child will be affected.

What causes facioscapulohumeral dystrophy?

FSHD is the result of a genetic deletion. Often, other kinds of neuromuscular disorders are caused by a genetic mutation or deletion directly affecting the protein or enzyme in the muscle or nerve. But the mutations in FSHD work somewhat differently. In FSHD the genetic deletion occurs in a part of the chromosome that affects how other genes around it are controlled. Scientists and researchers are still debating about which genes are affected by this deletion, but whichever gene (or genes) it happens to be, the deletion leads to a change of how other neighboring proteins and enzymes are made, and ultimately to the symptoms of FSHD.

Dr. Rabi Tawil of the University of Rochester, New York writes in 2004: "Despite the identification of a causal deletion on chromosome 4q35 over a decade ago, the molecular pathophysiology of FSHD remains unclear. The deleted repeats, though clearly associated with FSHD, do not contain expressed genes. The FSHD-associated deletions must, therefore, influence the expression of one or more

What is the age of onset of facioscapulohumeral dystrophy?

Symptoms can appear anytime from infancy to late adulthood, however this disorder generally appears in early adolescence. In perhaps five percent of cases, a young child or infant develops symptoms.

How is facioscapulohumeral dystrophy diagnosed?

Like many other types of neuromuscular disorder, diagnosis is made through a physical examination of reflex and sensory responses. Laboratory tests such as an electromyogram (EMG) which measures the electrical activity of muscle cells and muscle biopsies may be used to confirm the diagnosis. In addition, the physician may do a Creatine Kinase (CK) level test to determine if muscles are breaking down and releasing the enzyme creatine kinase into the blood. Other tests may include a Nerve Conduction Velocity (NCV) test measuring how fast signals are moving from the brain to the muscle.

An accurate genetic test is available for the most common type of FSHD, type 1A. The gene for type 1B FSHD has not yet been precisely located and thus a DNA test is not available. Such precise testing ensures that FSHD is not confused with other disorders such as polymyositis or nervous system diseases. A prenatal test for FSHD is also available.

genes at a distance from the site of the deletion." (Tawil, Curr Neurol Neurosci Rep. 2004 Jan; 4(1): 51-4.) A simple analogy to this might be to think of genes like roads and highways, and proteins and enzymes like the destinations. In many types of genetic disorders, gene mutation cause changes in the roads that sometimes make it difficult or impossible to reach particular destinations. In FSHD, it's as if the roads are all perfectly intact, but the signs and directions to get to our destination have been removed; the cell is unable to make the proteins and enzymes missing in FSHD without these directions.

Researchers continue to look at possible connections between their current understanding of the disorder and those so-called 'road signs.' In 2004, researchers such as Dr. Tupler of the University of Massachusetts Medical School have put possible explanations forward. These road signs -- repeated sequences of DNA labeled 'D4Z4' -- are under close scrutiny for their role in FSHD. Dr. Tupler writes: "Recently, 4q35 genes located upstream of D4Z4 have been found to be inappropriately overexpressed specifically in FSHD muscle. An element within D4Z4 has been shown to behave as a silencer that provides a binding site for a transcriptional repressing complex. These results suggest a model in which deletion of D4Z4 leads to the inappropriate transcriptional derepression of 4q35 genes, resulting in disease." (Tupler R, Cell Mol Life Sci. 2004 Mar; 61(5): 557-66.) In other words: an FSHD mutated gene has fewer copies of D4Z4. Fewer D4Z4 means that the muscle cells have less control of enzyme production. Too many enzymes are produced which, in turn, causes the symptoms of FSHD.

Is it contagious?

No. Genetic disorders are not contagious.

What research is being done?

One of the keys to treating FSHD will be to completely understand the gene and how it works within the cell. Currently, researchers are seeking to complete this understanding by studying what the gene does within the cell, what products it helps to control, and how it can be manipulated to reverse the negative effects of FSHD. This kind of understanding may lead to future strategies to help people living with FSHD and other genetic disorders. You can find out more information about recent FSHD research by contacting Muscular Dystrophy Canada, or through personal research.

Is there a cure or treatment?

There are still no treatments for this disorder that can halt or reverse the symptoms and muscle weakness. There are treatments and devices, which can help, ease the pain and discomfort associated with the disorder.

These include:

- drugs such as albuterol, clenbuterol, and oxandrolone, which are being studied for their muscle building effects. These kinds of treatments are important early on, researchers noting that "irrespective of type of muscular dystrophy, administration of clenbuterol may be beneficial in early stage of the disease." (Oya Y, Rinsho Shinkeigaku. 2001 Oct; 41(10): 698-700.) Other studies cautiously indicate that "albuterol did not improve global strength or function in patients with FSHD, [but] did increase muscle mass and improve some measures of strength." (Kissel JT, Neurology. 2001 Oct 23; 57(8): 1434-40.)
- anti-inflammatory drugs may be prescribed to reduce associated inflammation
- surgical procedures to stabilize the shoulder blades. Surgery may "produce significant benefits, though these have to be balanced against postoperative immobilisation, need for physiotherapy and potential complications." (Mummery CJ, Cochrane Database
- Syst Rev. 2003; (3): CD003278.)
- braces, girdles, or special bras to help compensate for weakened muscles moderate exercise, especially swimming, with supervision by a physical therapist

Where can I get more information?

Due to the variable nature of the disorder, its slow progression and sporadic periods of very rapid decline it is often difficult to access information and medical attention when needed. An FSHD Peer Support or Contact group enables individuals and families to access a wealth of information and assistance before a need arises in order to better 'manage' their disability. Muscular Dystrophy Canada can provide helpful information that includes contact with a network of people with similar concerns.

More detailed information is also available on the Muscular Dystrophy Canada website at www.muscle.ca.

How can I help?

Muscular Dystrophy Canada conducts year-round fund raising campaigns to support our diverse programs. Your gift will help the Association provide the dollars necessary to assist individuals living with neuromuscular disorders, and fund much needed medical research and educational information. Please make a gift through our National office or any Regional or Community Muscular Dystrophy Canada office.

All Muscular Dystrophy Canada Information Sheets are available on our website: www.muscle.ca

Ce feuillet d'information est aussi disponible en français.