Facioscapulohumeral Muscular Dystrophy

➢Facioscapulohumeral Muscular Dystrophy (FSHD) is a genetic disorder that involves progressive weakening and atrophy starting in the skeletal muscles in the face (facio), shoulder area (scapulo) and upper arms (humeral).

➢Two major types of FSHD are recognized, FSHD1A and FSHD1B. FSHD1A accounts for some 95% of FSHD cases and involves a known mutation. The FSHD1B mutation has not been identified.

➢FSHD is the third most common dystrophy after Duchenne muscular dystrophy (DMD) and Myotonic dystrophy (DM). FSHD occurs in about one in twenty thousand Canadians.

➢ Symptoms: Distinctive features include early weakness of the muscles of the eye & mouth (affecting the smile, pucker, and whistle), with weakness in the shoulder muscles.

➢ Eventual involvement of the muscles of the foot, ankle and hips frequently result in distorted gait patterns and about 20% of cases will eventually require a wheelchair.

➢FSHD symptoms may be so mild that adults may be unaware they have FSHD until a family member with more severe symptoms is diagnosed, and 5% of cases carry the mutation but do not develop any symptoms (but can still pass FSHD on to their children).

➢ The area around the shoulder blades is a primary area of muscle weakness in FSHD (shoulder weakness is the presenting symptom in more than 80% of patients). There is a gradual loss of muscle stability around the shoulders. The shoulder blades, which would otherwise be fixed in place, may not get the leverage necessary to lift or pull causing the scapula to lift or "wing" out as they move. Early observable symptoms include the inability to throw objects or lift the arms over the head.

➢The severity of symptoms experienced is linked to the extent of the genetic mutation associated with FSHD1A. There are two major classes: patients with severe mutations have earlier onset and are severely affected while patients with milder mutations show wide variations in onset and clinical presentation.

➢Most people with FSHD live a normal lifespan. Males tend to be more severely affected than females. As FSHD progresses, speech may be affected. Mental impairment and epilepsy is observed in about 40% of early onset (childhood) FSHD cases (especially when associated with large deletions).

➢ A few cases (less than 5%) may experience heart problems; atrial arrest, bundle branch block, and dilated cardiomyopathy have been reported.

➢ A few cases (less than 10%) may experience respiratory problems.

➢ Onset: In perhaps 5% of cases, a young child or infant develops symptoms. Generally, onset of muscle weakness is from the teens or early twenties into the thirties.

➢Diagnosis: Some symptoms of FSHD might be hard to see in mild cases. In typical adult-onset cases, clinical diagnosis is usually straightforward with evidence of major facial and shoulder weakness.

➢Genetic Testing: DNA tests can show the presence and size of the deletion in about 95% of cases of FSHD1A (including prenatal diagnosis).

➢Cause: The FSHD1A mutation involves a deletion of genetic information (referred to as "D4Z4" repeats) on chromosome number 4. It appears the mutation does not involve the structure of a gene, but rather acts by disrupting the transcriptional control mechanisms that govern one or more unidentified genes, possibly leading to an overproduction of certain enzymes and proteins. Ultimately these changes lead to the symptoms of FSHD1A. Genetic anticipation and germ line mosaicism are seen.

➢Inheritance: FSHD is inherited in an autosomal dominant fashion although many cases (30%) may spontaneously appear due to new mutations occurring in the early stages of embryonic development.

➢Treatments: There are currently no treatments for FSHD that can halt or reverse the symptoms and muscle weakness. Each symptom displayed reflects an underlying issue and each needs to have a treatment plan developed to best address the concern on an individual basis.

➢ANESTHESIA WARNING: A higher rate of complications is generally associated with general anesthesia in patients with neuromuscular disorders. Even mildly affected patients may have complications. Patients with FSHD or a family history of FSHD need to discuss this with their surgeons.

Neuromuscular disorders and what people commonly call muscular dystrophies are very complex and often devastating conditions. Some strike children, others do not display symptoms until well into adult life. Most are progressively disabling, getting worse over time and some are fatal, often in childhood. Some affect primarily muscles, others can affect a wide spectrum of health, involving personality and intellectual functioning, the eyes, the bones, digestion and diabetes to list just a few. There are over 100 different types of disorders and variations listed under this umbrella. Many disorders have genetic causes, but in many cases, just how these genetic mutations cause the symptoms we see is not yet understood. Every day, there is progress in understanding these disorders, but at the present time, there are more theories than facts. Sadly there are no direct treatments yet.

Please feel free to contact Muscular Dystrophy Canada if you have suggestions or questions about this page. Our website is: http://www.muscle.ca/ or e-mail us at info@muscle.ca

Thank you.