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Brief Report

# Facioscapulohumeral Muscular Dystrophy

**Sheila Mathew***

Doctor of Nursing Practice, Dominican College, New York, USA

***Corresponding author:** Sheila Mathew, Doctor of Nursing Practice, Dominican College, 470 Western Hwy S, Orangeburg, New York 10962, USA. Tel: +1-8455961945. Email: smathew107@hotmail.com

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## Introduction

The muscular dystrophies are a group of more than 30 genetic diseases characterized by progressive weakness and degeneration of the skeletal muscles that control movement. Some forms of MD are seen in infancy or childhood, while others may not appear until middle age or later. The disorders differ in terms of the distribution and extent of muscle weakness, age of onset, rate of progression, and pattern on inheritance (www.ninds.nig.gov).

One type of muscular dystrophy that I have chosen to discuss is Facioscapulohumeral Muscular Dystrophy (FSHD). FSHD is a genetic muscle disorder in which the muscles of the face, shoulder blades and upper arms are among the most affected. FSHD is most commonly found in adults and is classified among one of the progressive muscular dystrophies. Age of onset varies from infantile to late-life but typically occurs within the second decade [1]. Unlike other dystrophies, pathology is typically asymmetrical, as a muscle on one side of the body may undergo severe degeneration, while the same muscle on the opposite side may remain unaffected. Other commonly affected muscles are those of the shoulder, causing the scapula to protrude from the back ("Winging Scapula"). Muscles of the hip and lower leg can become weak and lead to difficulty walking [2]. FSHD is inherited in an autosomal dominant pattern. As a result, each son or daughter of a person with FSHD has 50% chance of inheritance). To determine FSHD several events need to concur: 1) there must be a contraction of the D4Z4 repeat array on 4q35; 2) the contraction is associated with chromatin relaxation and hypomethylation of the region; 3) DUX4 is transcribed; and 4) the transcript must be polyadenylated. This can only occur when contractions affect 4qA type chromosomes [3].

There are two forms of FSHD. The first form is FSHD1, this is a genetically heterogeneous disorder, has genetic bases and are unique and involves both genetic and epigenetic alteration. The second form is FSHD2 which has an undistinguishable clinical phenotype display and has a more complex pattern of inheritance and a distinct genetic defect [3]. Around 5-10% of patients with features of FSHD do not harbor a contraction of the 4q35 D4Z4 array and often have complex pattern of inheritance, which are those who have FSHD1. Chromatin studies in FSHD2 patients showed the presence of D4Z4 chromatin relaxation leading to DUX4-fl expression [3]. Both FSHD1 and FSHD2 result from inappropriate expression of DUX4 in different tissues. In FSHD1, this happens because the contracted D4Z4 repeats cannot form repressive heterochromatine and in FSHD2, one of the effector genes required for methylation (and repression of DUX4) is haplosufficient. The most frequent cause of FSHD is contraction of the polymorphic D4Z4 macrosatellite repeat array in the subtelomere of chromosome 4 at 4q35 resulting in FSHD1. This mutation explains >95% of the adult cases and all known infantile cases. Healthy individuals have 8 or more D4Z4 repeats on each 4q35 copy, whereas patients with FSHD1 have 1-10 repeats on one copy of the 4q35 chromosome region and a disease permissive allele 4A on the chromosome 4q subtelomere [4]. FSHD 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 (www.FSHSociety.org).

Studies of American, Brazilian, Dutch, Italian, and Chinese populations show sexual dimorphism in disease severity. Men are usually affected earlier in life, and more severely affected than women. Although one study in Korea FSHD subjects showed that women were more affected than men (178). In a Brazilian group 114 FSHD subjects were more clinically affected sons than daughters were reported among the offspring of asymptomatic women who had associated 4qA genetic disruptions as discussed later (299) [1]. With ongoing research, there is still a lack of information on the prevalence, natural history and clinical management of early onset FSHD. A cohort study on FSHD children was conducted

in Netherlands. Children age (0-17) with genetically confirmed diagnosis of FSHD1 or FSHD2.

However, children without genetic confirmation were included in the study if parents and or their children who were clinically suspected to have FSHD. Children were included in the study after obtaining informed consent [4]. Children were assessed at baseline and at 2 year follow up. The primary objective of the study was to assess the clinical, genetic, and epigenetic features of children with FSHD to optimize clinical management. The second objectives, to define a new comprehensive definition of early onset FSHD, to provide prevalence estimations of early onset FSHD in Netherlands, to establish a well-characterized baseline cohort for prospective follow up and recruitment for future clinical trials, and to assess 2-year progression rate in children with FSHD and develop a prognostic model. The iFocus study was a prospective observational study performed at the department of pediatric Neurology of the Radboud University Medical Center. Results from the iFocus study will provide insights into the clinical and genetic spectrum of children with FSHD. These insights are vital for adequate symptomatic management and clinical trial-readiness. In addition, the study aimed to provide additional insight in the epigenetic and environmental disease modifying factors [4].

When a patient's medical history, physical exam, and muscle testing indicate FSHD, blood creatinine kinase (CK) and DNA genotyping tests are conducted. CK is a prevalent metabolic enzyme expressed in cardiac and skeletal muscle tissue and is detected in blood in response to muscle damage or muscular dystrophy [1]. However, the range of CK levels varies in men and women and is only slightly to moderately elevated in FSHD and is therefore not a reliable diagnostic of FSHD. Genetic testing to determine if you have the DNA deletion for FSHD can be done through blood or tissue sample. There are three possible results; the first is a negative result which means that a person has not inherited FSHD. The second result is a positive result which means that a person has inherited a DNA deletion for FSHD. The accuracy of this result is close to 100%. A positive result does not necessarily mean that a person has any physical signs of FSHD, nor does it indicate at what age a person will begin to show signs of FSHD. A positive result usually means that at some point in that person's lifetime, he or she will develop at least mild symptoms of FSHD. A third result is uncertainty or the 'gray area' in FSHD testing. In these situations, the DNA deletion is so small that it is not certain whether the symptoms are related to FSHD [2].

There are a number of ways one can use in determining if they have FSHD. A Magnetic Resonance Imaging (MRI) has become a dominant diagnostic and monitoring tool for FSHD. Skeletal muscle MRI is noninvasive, low risk, and can be used to monitor disease progression [1]. Whole body MRI (WBMRI), are being used to obtain a more complete picture of muscle involvement in FSHD. A Manual Muscle Test (MMT) which originally developed in 1912, during a Polio outbreak is used to qualify muscle strength loss by rating force generated against external resistance. In 2010, a MMT was developed that was specifically modified for FSHD which included an evaluation scale for qualitative muscle strength and functionality of facial muscles, scapular girdle, upper limb muscles, pelvic girdle muscles and abdominal muscles.

Another method used to identify FSHD is Electromamyogram (EMG). EMG's distinguish between muscle and neurological diseases. Electrical impulses are diminished in affected muscles in subjects. A six-minute walk is also a muscle functional test that measures distance that subjects can walk within six minutes. It is used to test strength and cardiopulmonary reserve. Blood tests as well as genetic tests are also ways in which FSHD can be detected. Lastly, muscular biopsies are also another affective way in providing information on FSHD disease pathology and progression. In a muscular biopsy, small tissue samples are extracted and used for testing. However, it is noted that biopsies only allow analysis of small samples of tissue that may not be representative of the muscle as a whole as disease associated pathologies may be present in small present sections of the given muscle [1].

There is a big difference between symptomatic testing and asymptomatic testing. For an individual who does have symptoms for FSHD, testing is part of a diagnostic evaluation. For a person without symptoms, there are many issues to think about prior to being tested. Genetic counseling is an important part of the asymptomatic testing process. This involves providing information about implications of the testing by someone with expertise in genetic testing. In addition, costs for genetic testing will vary. Usually the cost of testing (DNA blood test, pre and posttest counseling and neurological examination) is under $1,200. Many insurance companies will cover the cost of testing. Having a positive test result can also lead a person to make difficult decisions in life, whether or not they should get married or have children. Testing is not offered to children below the age of 18 except in cases where a child may be having signs of FSHD. When children become legal adults, they can make their own choice about testing [2].

Through continued research of FSDH, researchers as well as doctors hope to find methods as well as different strategies of how they can help patients with FSHD and provide the best possible outcome for not only the lives of their patients but their families as well. While there is no etiological therapy available to date for FSHD patients, the role of exercise in maintaining or improving muscular force and/or functional ability is still controversial because of the lack of controlled studies and because vigorous exercise could worsen muscular weakness inducing rhabdomyolysis [1]. Several studies have shown that both strength training and aerobic exercise

in FSHD have at least a short-term beneficial effect. A one-year observational study was done that demonstrated safety, feasibility, and effectiveness of neuromuscular electrical stimulation strength training in FSHD1 patients. Physical therapy should promote a non-sedentary lifestyle, joint flexibility training to avoid retractions and improve muscle strength and aerobic capacity by mild aerobic exercising [1]. FSHD is a very serious and detrimental disease which should not be taken lightly; therefore, we must strive to one day find a cure which can save the lives of many people around the world. As a DNP, I would like help and create a treatment plan for therapeutic treatment for patients with FSHD.

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