FSHD Global Research Foundation Ltd

Management of

Facioscapulohumeral DYSTROPHY

A guide for allied health professionals

Allied health practice points

Diagnosis of FSHD involves clinical and genetic investigations

People seeking a diagnosis for possible genetic conditions should be seen by a genetic counsellor.

Management of FSHD is complex and involves a multidisciplinary team

A variety of health professionals including physiotherapy, occupational therapy and exercise physiology are necessary for optimal management.

Management should be goal oriented and individualised

Goals may include maintaining mobility or preventing falls. Management plans must be tailored to individual needs and take into account different patterns of weakness.

Pain is a common presentation of FSHD

Management options may include bracing, strapping, hydrotherapy or massage. Individualise plans based on responses.

Exercise is an important component of management

Exercise plans should be tailored to individual needs and patterns of weakness and reviewed regularly. Exercise should be started soon after diagnosis.

Mobility aids and orthotics are essential to maintain independence

Assessment of mobility needs should happen regularly to help people with FSHD maintain independence and identify any mechanical issues that may result in falls.

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant muscular dystrophy that affects an estimated 3,500 people in Australia (prevalence 1217: 100,000).

While FSHD is considered a rare disease it is likely that you, or someone in your practice, has encountered someone with FSHD or with a family history of FSHD.

At the moment there is no approved treatments for FSHD and no cure. However, this does not mean that there is nothing that can be done to help people with FSHD maintain quality of life.

This resource is for allied health professionals working with people with FSHD. In the absence of treatments or a cure you are vitally important to people with FSHD by providing essential help in managing progressive muscle weakness. This resource may help you by providing up to date information on the condition and the latest thinking about what works for people with FSHD.

FSHD: symptoms & prognosis

FSHD, like all the muscular dystrophies, is associated with progressive muscle weakness and loss of function. The condition is named for the typical pattern of muscle weakness observed in FSHD; face, shoulder and upper limb.

However, for many people with FSHD, the effect is not limited to these areas. Many people experience weakness in the trunk and lower limbs.

In addition, while the cardinal sign of FSHD is facial weakness leading to an inability to express emotions, some people may not exhibit any facial weakness.

FSHD can manifest at any point in a person's life from infancy through to late adulthood, and the severity and rate of deterioration is highly heterogeneous. FSHD that manifests in infants and children is usually particularly severe. The reason for the highly variable presentation of FSHD is not currently known.

Most people with FSHD will require mobility aids to complete activities of daily living. However, the rate of progression is highly variable. Some people may be reliant on mobility aids soon after diagnosis while others may not require support for many years after the first symptoms are noticed.

Unfortunately for people with FSHD this means that there is no prognosis that can be given at diagnosis. Vigilance is required to ensure that problems with walking are identified early to prevent falls and further loss of mobility.

FOR MORE INFORMATION ON THIS RESOURCE PLEASE CONTACT THE FSHD GLOBAL RESEARCH FOUNDATION ADMIN@FSHDGLOBAL.ORG , (02) 8007 7037, FSHDGLOBAL.ORG

FSHD: genetic cause and diagnostic pathway

There are two sub-types of FSHD that are genetically distinct, but clinically indistinguishable.

FSHD Type 1, which accounts for over 95% of cases is caused by mutations on chromosome 4. This mutation is a contraction of a D4Z4 repeat region. People without FSHD have between 11 and 100, people with FSHD have less than 11. These regions act like genetic silencing elements in regions of the genome where expression of genes is undesirable. In FSHD the contraction of the D4Z4 region leads to the production of RNA for a protein called DUX4. The presence of a poly-adenylation signal in the permissive allele allows the RNA to be translated into DUX4 protein. This protein is normally expressed during development. However, when expressed in mature muscle tissue it causes a cascade of epigenetic changes in muscle cell expression which leads to cell death and eventual loss of muscle function. The exact mechanism for DUX4 mediated damage is still to be elucidated.

FSHD Type 2 (around 5% of cases) is associated with mutations in a gene called the Structural Maintenance of Chromosomes Hinge Domain Containing 1 (SMCHD1). This gene acts as a transcriptional silencer preventing expression from the D4Z4 domain. Mutations in this gene cause a reduction in protein and a resultant reduction in inhibition that is independent of the number of repeats. The resultant pathology is caused by DUX4 expression and is therefore clinically identical.

Figure 1. Example of a clinical diagnosis pathway modified from Tawil et. al. 2015

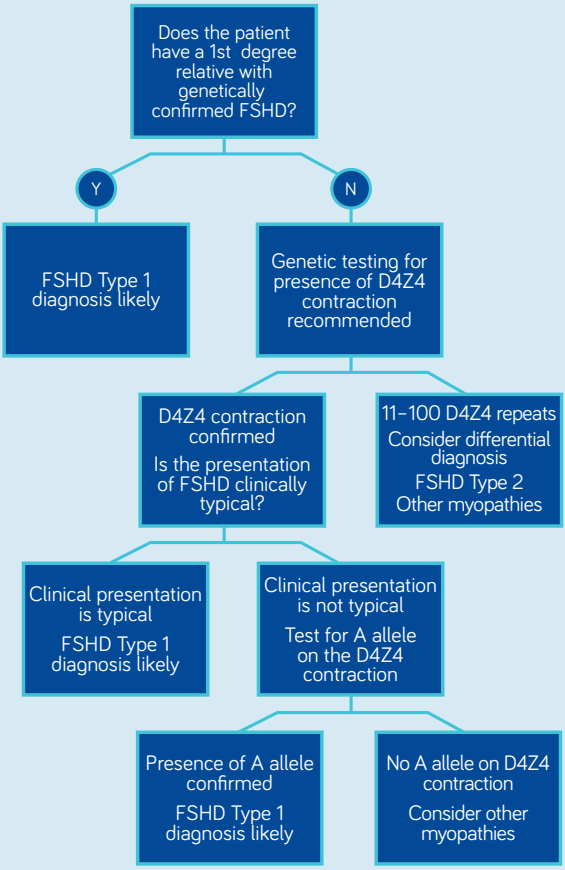


Figure 1

Management of FSHD

While there are no treatments for FSHD an effective rehabilitation program may help people with FSHD maintain quality of life.

An initial assessment of people with FSHD should include the following:

- Goals and expectations of the person with FSHD
- Assessment of muscle function
- Functional level of activities of daily living at home and in the community
- Pain history
- Home access issues
- Previous trial of exercise programs
- Current use of orthoses and adaptive devices

The evidence supporting the use of exercise programs for people with FSHD is inconclusive. A recent Cochrane review suggests that exercise is not harmful, however there was insufficient evidence to conclude benefit.

However, clinical experience suggests that exercise may prolong ambulation by maintaining muscle strength, improve tolerance and endurance and prevent the decline in gait parameters. Exercise may also help prevent obesity, deconditioning and prevent pain and fatigue.

A recent study demonstrated that aerobic exercise (15-30 minutes on a stationary bike three times a week for 16 weeks) combined with cognitive behaviour therapy increased physical activity and reduced fatigue.

An exercise prescription for FSHD can be challenging as muscle weakness tends to be asymmetrical and with large variations in rate of progression, age of onset and compensatory mechanisms each program needs to be individualised to achieve benefit.

Exercise programs should, for all people with FSHD:

- Be started early in the disease process
- Individualised
- Goal oriented (eg. maintaining independence, preventing falls)
- Practical

Each prescription should be customised to the disease history of the person with FSHD and include:

- Type of exercise
- Frequency
- Duration
- Intensity

Home-based exercise programs may be beneficial and regular review and modification of exercise programs are essential to ensure any progression of weakness is not compromising the effectiveness of the exercise program.

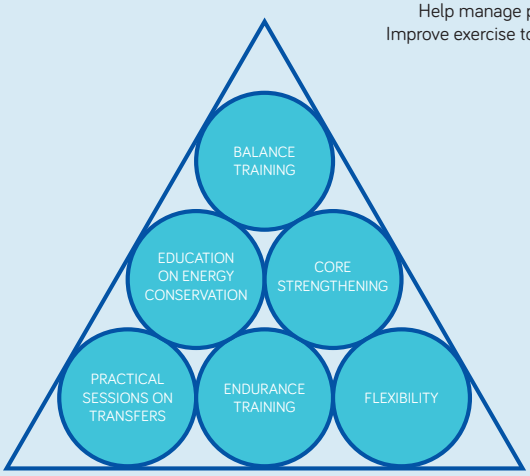
The building blocks of an effective exercise program

HYDROTHERAPY
General fitness
Help manage pain

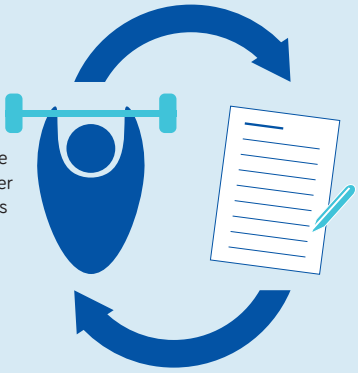
EDUCATION ON POSTURE
Help manage pain
Improve exercise tolerance

REGULAR STRETCHING
Encourage proper positioning
Prevent contractures

PILATES AND YOGA
Focus-strength
Balance
Flexibility and posture



Vigorous training to the point of fatigue or longer lasting muscle soreness should be avoided



Education regarding signs of overwork and weakness should be provided

Mobility aids and orthoses

Prescription of appropriate mobility aids and assistive devices can help maximise function and participation to maintain independence.

Walking aids such as canes or wheeled walkers should be considered for all people with FSHD to help maintain mobility. Walking aids can assist with balance, compensate for weak muscles, reduce fatigue and/or pain, facilitate a safe walking pattern and help prevent falls.

It's important to note that walking aids may increase weight bearing on the upper limbs exacerbating fatigue and pain in this area. This may be of particular concern for people with FSHD who have significant involvement of their upper limbs and shoulder.

Routine use of lumbar supports is not recommended due to concerns that overuse may lead to deconditioning. However, short term use may help people with FSHD improve sitting posture and may alleviate acute episodes of back pain.

Foot drop is a common presentation in FSHD and can lead to falls due to poor toe clearance. Customised orthoses may help alleviate poor toe clearance and help with safe walking. The aim of the orthotic is to:

- Provide adequate toe clearance in the swing phase
- Decrease excessive hip and knee flexion and ankle plantarflexion during swing phase
- Decrease excessive knee extension/flexion in stance phase
- Restore heel strike
- Improve energy efficiency
- Improve confidence, safety and endurance

Prescription of an appropriate orthotic requires a comprehensive assessment of the goals and expectations of the person receiving the orthotic as well as an assessment of their muscle strength, joint range, knee control, limb function and presence of any joint pain. The choice between ankle-foot and ankle-knee is an individual one. Ankle-knee orthoses provide better stance control, improved gait safety, decrease fatigue and are helpful in managing knee pain. However, they have poor cosmetic appeal and are difficult to get on and off.

All prescriptions of orthotics should be followed by gait retraining and education.

About one in five people with FSHD will become wheelchair dependent by the age of 50. Transition to a chair often occurs because of fatigue associated with walking and an increased risk of falls. The choice to transition to a wheelchair may be gradual starting with the use of a back up chair. Customised prescription of a wheelchair should be a collaboration between the user, carer, clinicians, suppliers and funding bodies.

People with FSHD face a myriad of challenges that include long delays in diagnosis, lack of clarity on prognosis and the need to constantly cope with a body that changes often suddenly and without warning. However, there are a great deal of options for people with FSHD and as their main point of contact for rehabilitation services you are vitally important to them.

More resources

References: Tawil, R., Mah, J.K., Baker, S., Wagner, K.R., Ryan, M.M., The Sydney Workshop Participants., Clinical practice considerations in facioscapulohumeral muscular dystrophy Sydney, Australia, 21 September 2015. Neuromuscular Disorders 2016; 26: 462 - 471

Tawil R., Kissel, J.T., Heatwole, C., Pandya, S., Gronseth, G., Benatar, M., Evidence-based guideline summary: Evaluation, diagnosis and management of facioscapulohumeral dystrophy. Neurology 2015; 85:357-364

About the Foundation

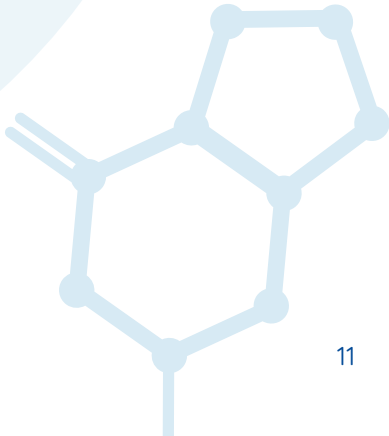
The FSHD Global Research Foundation focuses on finding treatments and a cure for the debilitating disease Facioscapulohumeral Muscular Dystrophy (FSHD). In doing so, we fund world-class medical research, awareness and education. We are also committed to complete transparency and accountability in our operations.

The Foundation was established in 2007 by Bill Moss AO, a well-known Australian businessman and philanthropist who has FSHD. Since then, we have been addressing the chronic lack of medical funding and awareness of FSHD, both in Australia and globally. Over the past 9 years, the Foundation has committed $8.3 million to fund 40 ongoing medical research grants in 9 countries; the USA, Canada, the Netherlands, Italy, France, Belgium, Spain, New Zealand and Australia.

The FSHD Global Research Foundation does not operate like an average not for profit. We allocate 100% of all cash tax deductible donations to current and future medical research grants. We are also transparent in doing so, offering all donors via the 'FSHD – Find the Cure' mobile app the opportunity to track exactly which research programs their money has been allocated and the latest milestones of those programs.

The main sources of our funding for FSHD research are individuals afflicted by FSHD, their friends, supporters, as well as corporate sponsors. All funds donated are invested through careful consideration, guided by our Scientific Advisory Boards, Board of Directors and International Research Committees, ensuring FSHD Global remains a leader in discovering the world's best science.

Notes



The FSHD Global Research Foundation is committed to advancing global medical research, education and collaboration to improve quality of life and ultimately find a cure for Facioscapulohumeral Dystrophy. Through transparency, accountability, good governance and pure passion we aim to achieve results as quickly as possible.

FSHD
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