Facioscapulohumeral Muscular Dystrophy (FSHD)

Including 2 related types:

- Early Onset FSHD
- Infantile FSHD

Processing a new diagnosis

A diagnosis of FSHD can be a huge shock for yourself, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at **https://youtu.be/A9XM71knDVw.**

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on **Psychology** for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on **Counselling** for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on **Social Work** for more information.
- A genetic counsellor can help you understand how FSHD is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Visit our page on **Genetic Counselling** for more information.
- Your state or territory neuromuscular organisation can provide support, advice and information about living with DM. Visit our page on **state and territory neuromuscular organisations** for more information.
- Your GP can talk to you about a mental health plan and how you can use this to help you access the support and services you need. Visit our page on **Wellbeing** for more information.
- Other people and families living with FSHD have also experienced the emotional rollercoaster that comes with a diagnosis of FSHD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with FSHD in The Loop Community on our **Forum**.

How to talk to your child or others about a new diagnosis

Having a conversation with a loved one about a diagnosis of FSHD, is not easy. In fact, it will probably be one of the hardest talks you will have. But, like all difficult conversations, it is important and necessary.

There are many reasons why people may not want to have this conversation, mostly centred on wanting to protect their loved ones and themselves.

But avoiding the topic is not helpful. People may have noticed differences and changes that have occurred and/or heard the condition being discussed.

So it is crucial that you have a role in your loved one learning about their/your diagnosis. This will enable you to support them as they process the information and to be on hand to answer any questions they may have. If you are the person FSHD it will provide you with support and someone to talk to. Together, you can learn about the journey ahead

It will also allow you to:

- Provide answers to questions in an age appropriate way
- Correct any misinformation they have heard or read
- Show them that you have faith in their ability to handle difficult conversations.

There is no right time to start this conversation. Research suggests the earlier you talk to your loved ones about your/their condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don'ts:

- Do be positive but also realistic.
- Do reassure them that no one has done anything wrong and this is not their fault.
- Do tell them they/you will do many wonderful things in their/your life – they/you may just do them differently.
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to.
- Don't avoid answering questions and don't shut the conversation down.

If you are having difficulty starting the conversation, just ask a couple of questions such as 'How are you feeling today?' or 'Do you know why you/they are having difficulty with your/their muscles?'

Remember it's okay to be upset during the conversation but try to avoid breaking down as this will only cause greater distress. And if speaking with a child, remember children are resilient and they generally handle information of this nature far better than adults do.

Understanding FSHD and how it's diagnosed

Facioscapulohumeral muscular dystrophy (FSHD) is one of the most common forms of muscular dystrophy (a group of diseases that cause your muscles to become progressively weaker) and can affect both males and females. The most common symptoms are progressive weakening of facial, shoulder and upper arm muscles and it can affect both children and adults.

FSHD Global Research Foundation states that "FSHD affects 1 in every 7500 Australians – around 3000 people. It is estimated to affect around 1 million individuals globally."

What causes FSHD?

FSHD is a genetic condition, meaning it is caused by a mutation (a change or fault) in a person's DNA (the parts of our cells that hold our genes and tell our cells what to do).

FSHD is hereditary, meaning the genetic mutation is passed from parents to children. However about one in three people with FSHD have no prior family history of the disease.

What are the types of FSHD?

There are two types of FSHD: the more common type 1 (FSHD1) and the less common type 2 (FSHD2) which generally have the same signs and symptoms, but are caused by different genes. They can both affect adults and children.

However, FSHD is a complex genetic condition, and the severity of the disease has to do with other factors controlling genetic expression. It is likely that more subtypes of FSHD will be identified as knowledge of FSHD improves.

When FSHD affects infants and children, they are sometimes classified as "early-onset FSHD" or "infantile FSHD". Early-onset FSHD is much rarer, and tends to be more severe. Many children with the condition experience a faster and more severe decline in muscle function. Hearing and sight complications are also more common.

How is FSHD diagnosed?

The information below is from FSHD Global Research Foundation Australia's publication **Facioscapulohumeral Dystrophy: A guide for people living with FSHD.**

"*Diagnosis of FSHD may involve a combination of:*

- *Physical examination to assess muscle weakness*
- *Blood, neurological and imaging investigations to confirm a muscular disorder and rule out other causes of weakness and*
- *Molecular investigations to determine whether a disease-causing genetic change is present.*

There are a number of ways to confirm the diagnosis of FSHD. The simplest is through a blood test where your genetics can be assessed. The only test that can diagnose FSHD and rule out other causes of muscle weakness is a genetic test.

Progressing straight to a genetic test may be appropriate for some people such as those with the typical symptom pattern and family history of FSHD.

For others it may be appropriate to perform some other tests before having a genetic test. If you have the typical pattern of symptoms expected of FSHD and a first degree relative (a person's parent, sibling or child) with genetically confirmed FSHD then proceeding to a genetic test is probably unnecessary. Family history and evidence of symptoms is sufficient to confirm an FSHD diagnosis"

Unfortunately it can be a long journey to getting a diagnosis of FSHD due to the non-specific and varying symptoms people can experience. It is important to remember you and your family can seek support and advice from your GP, specialist and/or your state or territory neuromuscular organisation during this time.

The genetics of FSHD

FSHD is a complex condition and researchers are still trying to fully understand the genetics that cause it. Nearly all cases of FSHD involve a genetic mutation, a change or fault in your DNA). This mutation switches on a gene called DUX4 that shouldn't normally be switched on. When this gene is activated, it produces a certain protein, called DUX4 protein. DUX4 protein is thought to contribute to muscle wasting, inflammation and damage inside the muscle cells of someone with FSHD.

FSHD Global Research Foundation's website has explanations and diagrams that assist in explaining the genetic cause of FSHD. There are diagrams that show chromosome 4 with no FSHD, one with FSHD1 and one with FSHD 2. These are complex diagrams and you may need to seek your GP's or specialist's advice to help explain and understand these diagrams.

Long-term outlook

FSHD usually begins before age 20, but it can begin as early as infancy and as late as after 50 years of age. The progression of FSHD is highly variable. However, most people with FSHD develop noticeable muscle weakness by their 30s. Most people with FSHD have a normal life expectancy. Although there is no cure for FSHD, medications and therapy can help manage some symptoms and potentially slow the course of the disease.

Although we know a lot about the adult form of FSHD the infantile form, and how it progresses, is less well understood.

While no treatment exists that slows the progression of FSHD, management of its symptoms can greatly improve quality of life. See the Living With Your Condition section for more information on symptoms.

Living with FSHD usually means life, and the future, looks different from what you had planned. However children and adults with FSHD can lead fulfilling, rewarding lives just like everyone else. School, friendships, sports and recreation, arts, university, work, having a family and travel are all possible when you have FSHD.

For more information about living with FSHD, overcoming some of the day-to-day challenges and where to get the right support, visit:

- **Living Life**
- **Counselling**
- **Social Work**
- **Psychology**

Understanding and planning for changes

Learning about FSHD and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person's journey with FSHD will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

What are the main symptoms of FSHD?

The main symptoms of FSHD can vary from person to person but generally FSHD is associated with progressive weakening of the muscles in the face, shoulders and upper arms.

Weakening of facial muscles can make it difficult to drink from a straw, whistle or smile.

Weakness in muscles around the eyes can prevent the eyes from closing fully while a person is asleep, which can lead to dry eyes and other eye problems. Lubricants or using an eye patch while sleeping may assist in managing this symptom.

The weakening of shoulder and upper arm muscles may make the shoulder blades 'wing" out and make it hard to raise arms over your head or throw a ball.

In most people with FSHD, weakness may be different between the left and right sides of the body. This is called asymmetrical weakness.

Other symptoms may include:

- Fatigue (tiredness)
- Pain in the shoulder, neck, lower back, and lower legs
- Weakness in lower leg muscles which may lead to a condition called foot drop. This may lead to tripping and increase the risk of falls.
- Contractures where joints stiffen in one position, most likely if FSHD affects the ankles.
- Muscular weakness in the hips and pelvis which can make it difficult to climb stairs or walk long distances.
- Changes to your posture and alignment of your spine, due to weakening of muscles in your back. This can take the form of lordosis, where there is an increased inward curve of your lower back (also known as swayback). It also can take the form of mild scoliosis, in which the spine curves to the side, like an S.
- Hearing loss, particularly of sounds that are higher pitched. This is not a common complication and mostly affects children who have FSHD.
- Vision loss due to changes to blood vessels in the back of the eye (retina)
- Rarely, FSHD may affect the heart (cardiac) muscle or breathing (respiratory) muscles, causing symptoms such as shortness of breath, sleepiness during the daytime and chest infections.

Progression and monitoring

FSHD usually progresses very slowly and only rarely causes serious health complications. Most people with the disease live full, independent lives with a normal life span.

Breathing difficulties and problems with the heart are not common complications of FSHD. However, regular checks of your heart and lung function are recommended as early symptoms can go unnoticed and may only be picked up with specialised tests.

You may also need regular checks of your vision, particularly the retina (the area at the back of your eye) and general eye health.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at **https://youtu.be/_6oJ9Mc0UO8.**

Life stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our **Living Life** section.

Where to find more information about living with FSHD

Facioscapulohumeral Muscular Dystrophy (FSHD) fact sheet
From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

What is Facioscapulohumeral Muscular Dystrophy? A Guide for Patients and Families
From Muscular Dystrophy Association, United States, November 2019. This guide includes information such as:

- What are the symptoms of FSHD?
- What should I know about FSHD?
- How is FSHD treated?
- A glossary of terms.

Facioscapulohumeral Dystrophy: A guide for people living with FSHD
From FSHD Global Research Foundation, Australia, 2016. This guide includes important information, including:

- Understanding FSHD
- Genetic mechanism of FSHD
- Diagnosing FSHD
- What should your care team look like
- Getting the most out of healthcare
- Options for management.

Information About FSHD

Treatment and management

There is no cure for FSHD but medications and therapies can be used to help manage some of the symptoms of FSHD and the impact of these symptoms on your life. Being a rare disease, it is often not well understood by the general medical community and you may find you need to work in partnership with, and help educate, your healthcare team. Treatments include:

- Anti-inflammatory drugs, known as nonsteroidal anti-inflammatories or NSAIDs, are often prescribed to control pain and improve mobility.
- Massage or warm, moist heat may also help with the pain associated with FSHD.
- **Physiotherapy** may help to retain muscle strength and function, enhance mobility, help you manage pain, and prevent falls.
- **Occupational therapy** can help with maintaining daily living skills, including the use of equipment and aids for home, work, study and recreation.
- **Speech pathology** can help with any difficulties you're having with speaking and swallowing.
- Low-intensity aerobic exercise and **hydrotherapy** (exercising in water) may be recommended to help maintain mobility. Any exercise program should be discussed with a doctor before starting and should be tailored to your symptoms, age and general health.
- The use of foot/ankle orthoses, to help lift your toes when walking, can help to prevent falls.
- Use of **equipmen**t such as a walker, manual wheelchair or power wheelchair can be helpful if you're finding it difficult to walk independently, tire easily or are at risk of falling.
- Shoulder surgery may be an option to attach the shoulder blades to the rib, making your shoulders more stable. However, this type of surgery does not always result in significant improvement and it is important that you have considered all the risks and benefits with your health professional before deciding on surgery.
- Hearing aids can be used for hearing loss.
- Lubricants or using an eye patch while sleeping may assist in preventing and managing dry eyes.
- Devices such as back supports, braces and special bras can help support weakened muscles in the upper and lower back.
- A bi-level positive airway pressure (BiPAP) machine can be used for weakness of breathing muscles.

Care recommendations for FSHD

The FSHD Global Research Foundation has developed the **Clinical Consensus on Diagnosis and Management of FSHD.** This document provides detailed, evidence-based (based on the findings of the best available current research) advice on how to diagnose and manage FSHD. The Consensus also provides advice about what an effective neuromuscular care team might look like to provide the best level of care to someone living with FSHD.

Items outlined in the consensus include:

- diagnosis and genetic testing
- lifestyle interventions including aerobic exercise
- rehabilitation including referral to rehabilitation specialist, physiotherapist, occupational therapist and/or orthotist.
- surgery including referral to an orthopaedic surgeon with experience in neuromuscular conditions
- pain and options for management
- assessment and screening of bone density
- respiratory management and regular monitoring of respiratory function.

Although this document might seem long and daunting, it can be useful for discussions with your specialists, doctors, nurses and other health professionals. Try to familiarise yourself with the consensus and talk to your doctor or state/territory neuromuscular organisation about any topics you don't understand or have concerns about. There are also some resources that have been developed by FSHD Global Research Foundation to be used by doctors and allied health professionals, as well as a guide for people with FSHD. See the Useful FSHD Resources section below for links to these resources.

Support with family planning

People with FSHD may have concerns about starting a family because of the risks of passing the condition to their children. These issues should be discussed with a genetic counsellor as there are a number of options available. Visit our page on **Genetic Counselling** for more information.

Other helpful support services

The video is available at **https://youtu.be/BDaCNsd9R_s.**

- Your state or territory neuromuscular organisation can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing. Visit our page on **state and territory neuromuscular organisations** for more information.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them here. Visit our pages on **Psychology**, **Counselling**, **Social Work** and **Wellbeing** for more information.
- Living with FSHD may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on **Disability Support Workers** for more information.
- Other people and families living with FSHD have also experienced the emotional rollercoaster that comes with a diagnosis of FSHD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with FSHD in The Loop Community on our **Forum**.

Medical Alert Card

Muscular Dystrophy Queensland has developed a **Medical Alert Card for FSHD**. A Medical Alert Card can help you communicate your care needs in a medical emergency situation. If you would like a Medical Alert Card to keep in your wallet or purse, please call (07) 3243 9700 or complete the form on the Muscular Dystrophy Queensland **website**.

How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

Employers

Educators

Living Life: Education

Guides & Fact Sheets

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- Understanding FSHD
- Genetic mechanism of FSHD
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- What should your care team look like
- Getting the most out of healthcare
- Options for management.

Care Recommendations

Clinical Consensus on Diagnosis and Management of FSHD.
By the FSHD Global Research Foundation.

The FSHD Global Research Foundation (Australia) have developed clincial consensus resources for primary care (GP) and allied health professionals including:

- For general practitioners: **Clinical Consensus on Facioscapulohumeral Dystrophy: A resource for primary care**
- For allied health professionals, such as physiotherapists and occupational therapists: **Management of Facioscapulohumeral Dystrophy: A guide for allied health professionals**

Physical therapy for Facioscapulohumeral Muscular Dystrophy has been developed by FSH Society (US) to assist physical therapists (physiotherapists) and individuals with FSHD in developing a plan of care based on the best research evidence, clinician expertise, and patient preferences.

Registry

Australian Neuromuscular Disease Registry (ANMDR)
The **Australian Neuromuscular Disease Registry (ANMDR)** aims to establish a database of all patients, children and adults in Australia diagnosed with neuromuscular disease. By creating this database, they hope to form a network of patients, clinicians, researchers, and industry to further research into neuromuscular diseases, ease the process in finding and selecting participants for clinical trials, and create an additional support for patients and their families. Joining the Registry is entirely voluntary and Australian families affected by FSHD are encouraged to **register** or contact the Registry for more information via **anmdr@mcri.edu.au**.