



# Facioscapulohumeral muscular dystrophy (FSHD)

'I have been diagnosed with FSHD for around 13 years. I was overwhelmed with this bombshell, my future life was completely re-written and I suppose a grieving process was initiated. Reading the factsheet it is important to remember all possible symptoms are listed but this does not mean that everyone will get all the symptoms. Please get in contact with Muscular Dystrophy UK, they can put you in touch with someone with the condition who can answer any questions you may have in a less clinical way. Sometimes it just helps to talk to someone without having to explain how it affects you, someone who just 'gets it.'

Liz Williams, who has FSHD

## What is FSHD?

FSHD is a muscle-wasting condition caused by a genetic mutation, which switches on a gene that shouldn't normally be switched on.

The name describes the areas where FSHD usually causes weakened muscles:

- ▶ 'facio' = facial
- ▶ 'scapulo' = shoulder blade
- ▶ 'humeral' = upper arm.

Sometimes the lower legs can also be affected. The term 'peroneal' refers to this area of the body.

Scapulohumeral and scapulooperoneal syndromes are conditions that have similar clinical symptoms to FSHD. Landouzy-Dejerine and facioscapulooperoneal muscular dystrophy are two names for FSHD that are not used often nowadays.

## How common is it?

FSHD is one of the most common forms of muscular dystrophy. Experts estimate that between three and five people out of every 100,000 have FSHD. In the UK it is estimated that between 2,000-2,500 people have FSHD.

## How is FSHD diagnosed?

The DNA mutation causing FSHD can be recognised from a blood sample in most cases. The sample will need to be forwarded to one of a few molecular genetic laboratories able to analyse it for this mutation.



For more information about diagnostic and genetic tests, please see our [Diagnostic tests](#) factsheet.

## How will FSHD affect my muscles?

The earlier in life the weakness appears, the more severe it will eventually be. It is hard to predict how arm or leg weakness will progress. Although in more than 50 percent of cases the legs are affected to some degree, where this happens in early adulthood, it is unlikely that person will ever need a wheelchair.

One fairly common feature of FSHD is an asymmetry of weakness: where one side of the body is more affected than the other (particularly early on). This is often evident in the shoulders. It is also usually the right side which is first affected in right-handed people.

The degree of weakness or disability can vary quite widely between different affected members in the same family, but it can show even greater variation between people in different families. For some, it can result in weakness not only of facial muscles and shoulders/upper arms, but also of additional combinations from the neck, forearms, wrists, fingers, hips, legs, ankles and the back muscles.

About 10 to 20 percent of people with FSHD eventually require a wheelchair. By contrast, up to one third remain unaware of symptoms at least into old age, although they may well have subtle signs of FSHD only noticeable to a doctor/specialist. The majority of people with FSHD come somewhere between these two extremes.

Early weakness at the ankles can cause 'foot drop'. In over 50 percent of people some degree of weakness at the knees or hips develops by middle age. Together with weakness in the back muscles, this can result in a typical backward-leaning and high-stepping style of walking.

In general, the people most severely affected tend to be the first members of a family to be diagnosed, and where the symptoms of weakness are evident from early childhood.

## Can any other problems be anticipated?

In some cases where symptoms of FSHD start in early childhood, learning difficulties and epilepsy are possible.

Hearing loss is common but may not be symptomatic of the condition.

Some people experience conjunctivitis and ulceration of their cornea because of limited blinking and an inability to close their eyes properly, during the day and when sleeping. Using artificial tears and protecting the eyes during sleep may help. Specific problems with blood vessels at the back of the eye (retinal vasculopathy) can occur, and although this rarely causes visual problems, it may be useful to have periodic eye checks.



Muscle pain is quite a frequent complaint in FSHD, often in its early stages. This may relate to inflammation within the muscles, which seems to occur more in FSHD than other muscular dystrophies. A physiotherapist can recommend mild exercises to help alleviate the pain. Treatment with simple painkillers and anti-inflammatory drugs is common, but how much relief this gives can vary.

Lung function is usually normal. However, in a minority of patients, weak breathing muscles may result in respiratory failure. This causes shortness of breath and nocturnal hypoventilation (morning headaches, feeling sleepy in the daytime, not feeling refreshed in the morning, dizziness). Patients with respiratory failure can be more likely to get chest infections. Patients with swallowing difficulties might also be at risk of aspiration pneumonia.

If the breathing function is affected, and if supplemental oxygen is required during a respiratory crisis, it must be carefully controlled and carbon dioxide levels monitored.

Non-invasive ventilation (NIV) may be required. Assisted coughing with chest physiotherapy and breath-stacking techniques with an AMBU bag helps to clear fluids in the lower airways during acute chest infections. The same techniques can help prevent problems when breathing may be difficult. A cough assist machine can also help

## Does FSHD affect life-expectancy?

Generally speaking, life-expectancy is not affected. The exception could perhaps be in the most severe cases, where not being able to move about much increases the risk of chest infections. Some recent reports suggest increased risk of heart rhythm disorders, but only in a few cases, and these respond to medication. Adults with FSHD should see their GP (or hospital doctor) every few years for a simple heart check.

## Treatment

### Are there ways to improve muscle strength?

There are no cures for FSHD or specific drug treatments. Regular exercise (especially hydrotherapy) helps to keep people moving and manage pain. It is essential to keep your weight down (through diet if necessary) to reduce stress on already weakened muscles. If you're exercising to increase muscle strength, build your exercises up gradually.

### Can orthoses help?

The use of orthoses (devices that support the feet) can be helpful for people with FSHD. They frequently improve the most common features, such as foot drop and shoulder weakness. After your physiotherapist assesses you, ask to be referred to the Orthotics team. They will guide you which orthoses will help you.



## Can surgery help?

The scapular muscles, which attach the shoulder blades to the chest, are often very weak with FSHD and can make lifting the arms difficult. The operation of 'scapular fixation' (fixing the shoulder blades to the ribs at the back) has enabled some people to regain more use of their arms.

Because people with FSHD may not be moving their arms or legs much, this could make these muscles weaker. It is advisable to have a combined assessment from a neurologist and an orthopaedic surgeon first.

If the eyes become inflamed because they remain open at night and artificial tears alone don't help it is possible to have surgery to bring the eyelids closer.

## What is the risk of anaesthetics?

There is no known risk, but some patients with FSHD can experience increased sensitivity to sedatives, inhaled anaesthetics and drugs that relax the muscles (neuromuscular blockers). Make sure the anaesthetist is aware of your diagnosis before any operation so they know how to look after you after the operation. This is especially so for people whose lungs don't work as they should.

Local anaesthetics and nitrous oxide are safe, for example in minor dental procedures.

It is also helpful to carry an FSHD alert card, and a care plan, both of which contain information to alert emergency and other healthcare professionals of the specific issues that affect you. These are available for free from Muscular Dystrophy UK – call the Freephone helpline on 0800 652 6352 or email [info@muscular dystrophyuk.org](mailto:info@muscular dystrophyuk.org).

## What causes FSHD?

Every human has 23 pairs of chromosomes that contain DNA. FSHD is caused by a genetic mutation (sometimes called a 'fault') that removes some of the DNA on chromosome 4. This chromosome contains lots of repeated pieces of DNA called D4Z4 repeat units, arranged like a train of identical carriages.

In someone with FSHD, the number of D4Z4 repeat units is reduced, like a train having too few carriages. This causes a gene called DUX4 to be unnecessarily switched on and produce DUX4 protein. DUX4 protein is thought to contribute to muscle wasting, inflammation and damage inside the muscle cells of someone with FSHD.

Scientists are trying to find out more about the function of DUX4. This will lead to a better understanding of FSHD and will help in the development of potential treatments.

To some extent, knowledge of the number of D4Z4 DNA repeats can indicate how severe the condition is and how it is expected to progress.



## DNA on chromosome 4



**DUX4 switched off**  
Person without FSHD



**DUX4 switched on = muscle damage**  
Person with FSHD

Figure 1: FSHD is caused by a loss of DNA on chromosome 4, which leads to the DUX4 gene being switched on



## How is FSHD inherited?

Our DNA is stored in structures called chromosomes. Every chromosome is part of a pair (one from your mother and one from your father).

In FSHD, one copy of chromosome 4 is faulty. Hence there is a 50:50 (one in two) chance of each child of an affected parent to inherit the faulty copy, resulting in FSHD. The children also have an equal chance of inheriting the good copy (resulting in no risk for these individuals or their future children having FSHD). This pattern of inheritance is called 'autosomal dominant'. For more information about the pattern of inheritance, please see our [Inheritance](#) factsheet.

## Is there always a family history of FSHD?

A person diagnosed with FSHD, particularly in early childhood, may not have inherited it from either of their parents (called a novel mutation). More often, however, a person diagnosed with FSHD will have inherited the faulty gene from one of his or her parents.

It may be that there is a family history that had not been recognised before. This could be because the symptoms of other family members had been very mild, or they had been misdiagnosed.

In some cases where FSHD develops in young children, who appear to be the first ones in a family, one of the parents can show the same FSHD mutation in some of their cells but not in others. This situation in the parent may not give them any symptoms. But this does mean that further children of theirs would potentially be affected. It's recommended that both parents provide blood samples for DNA study if they wish to know whether future children are likely to have the condition.

In other cases, genetic testing may determine whether or not a young adult is affected. Family members or couples seeking further information should ask for a referral to their local Clinical Genetics Service.

## Are men and women affected equally?

On average, men with FSHD tend to show more weakness and from a slightly earlier age than women. The reason for this is not yet clear. Within large families, and excluding the most severe cases, women are more likely to be less severely affected and could be unaware they have inherited the condition. By the age of 30, nearly all males with FSHD show symptoms, but only two-thirds of females do.

## Will all family members be affected equally?

Children inheriting the faulty gene are likely to be affected from a similarly young age and at least as severely as their affected parent. In large families, daughters with FSHD might be less severely affected than their fathers.

The age when symptoms begin, and the severity of FSHD seem to relate broadly to the number of D4Z4 repeats on chromosome 4. This usually remains fixed within a family.



Therefore there will be some families where FSHD will always tend to be quite severe and others where it will always be relatively mild.

Some people (particularly men) with average or mild presentations of FSHD, may, if they are the first cases in a family, have a mixture of normal and FSHD-type cells. Their children, who will inherit the FSHD mutation in all their cells, will develop signs of FSHD earlier and more severely.

## At what age does FSHD usually become noticeable?

This is dependent on the number of D4Z4 repeats on chromosome 4. Generally, the fewer D4Z4 repeats, the earlier the age when FSHD starts and the greater the severity.

In large families where several members have FSHD, a person usually first becomes aware of muscle weakness in their teenage years or early adulthood. He or she may experience difficulty in raising one or both arms, or may notice prominent shoulder blades or the weakening or wasting of their upper arm muscles.

In the more severe cases, difficulty moving the facial muscles, particularly around the mouth, can be evident by early childhood. This can be followed by weakness in the shoulder and upper arm. These children may experience progressive weakness of the legs by their teenage years, and may need a wheelchair.

By contrast, in the families less severely affected, people inheriting the condition may remain unaware of symptoms until late in adulthood.

## If I have no symptoms, can I still carry the genetic fault and pass it on to my children?

If the person has been affected from childhood, it is very unlikely that an adult relative (say a brother or sister) who is unaware of any symptoms, could 'carry' the faulty gene or pass the condition on to their children. The parents of the affected child are an exception, as they could be 'carrying' the mutation but in only some of their cells. They could pass the condition on to more than one child.

For people from families where several relatives or a parent have FSHD, DNA testing is the only way to find out if they too are carrying the faulty gene. In these situations, many people potentially affected may be affected only mildly, and are unaware of the signs that would show a specialist they have FSHD.

Up to one-third of women carrying the milder mutations for FSHD, and a probably much smaller proportion of men, may not show any definite signs of the condition. Therefore, the answer to this question can only be given reliably by DNA testing.



## Can one of my children have inherited FSHD and another not?

If the apparently unaffected child is several years beyond the age at which the affected one first presented with symptoms, it is likely that they have not inherited the condition.

This is particularly so if:

- ▶ the affected child is the first person in the family to show signs of FSHD, and
- ▶ DNA testing has shown that their condition has arisen from a new DNA mutation not present in a DNA sample from either parent.

However, if either parent is affected or carries the mutation, the only way to be sure is to have DNA testing. If a child has no signs of FSHD, requests for DNA testing would normally be refused until they are of an age (16 years old in the UK) to choose this for themselves.

## Is there any way of not passing the faulty gene on to my children?

Accurate pre-natal testing, performed by chorion villus biopsy (CVS) usually at 11 weeks pregnant, is now available to find out if the faulty gene has been passed on. (For more information, please see our [prenatal testing and diagnosis](#) factsheet). Couples considering CVS, should get in touch with their local genetics service.

## What about research?

Find out the latest news on FSHD research by visiting our website at <http://www.muscular dystrophyuk.org/news/news/research/>

To find out more about the research that Muscular Dystrophy UK is funding please visit: <http://www.muscular dystrophyuk.org/progress-in-research/research-projects/current-grants/>

If you have any questions about FSHD research and clinical trials, please email our research team at [research@muscular dystrophyuk.org](mailto:research@muscular dystrophyuk.org)

If you'd like to speak to someone who can relate to what you're going through, or give you practical advice, we can introduce you to a specially trained volunteer. You can talk to them confidentially, over the phone or on email. For more information, please email the information team at [info@muscular dystrophyuk.org](mailto:info@muscular dystrophyuk.org)

## Other related publications and resources:

- ▶ [Inheritance and muscular dystrophies](#)
- ▶ [An introductory guide for families with a child newly diagnosed with a neuromuscular condition](#)
- ▶ [Education guidelines](#)
- ▶ [Muscle biopsies](#)
- ▶ [Heart check](#)
- ▶ [Pregnancy and fertility](#)
- ▶ [Prenatal testing and diagnosis](#)
- ▶ [Carrier testing and reproduction: your options](#)



- ▶ [Exercise advice for adults with muscle wasting conditions](#)
- ▶ Muscular Dystrophy UK's TalkMD forum: [www.muscular dystrophyuk.org/talkmd/](http://www.muscular dystrophyuk.org/talkmd/)
- ▶ Joseph Patrick Trust: [www.muscular dystrophyuk.org/jpt](http://www.muscular dystrophyuk.org/jpt)
- ▶ American Academy of Neurology's Summary of Evidence-based Guidelines for Patients and their families: [www.aan.com/Guidelines/home/GetGuidelineContent/702](http://www.aan.com/Guidelines/home/GetGuidelineContent/702)
- ▶ UK FSHD patent registry: [www.fshd-registry.org/uk/](http://www.fshd-registry.org/uk/)
- ▶ Theadom A, Rodrigues M, Roxburgh R, Balalla S, Higgins C, Bhattacharjee R, *et al.* *Prevalence of muscular dystrophies: a systematic literature review.* Neuroepidemiology 2014; 43(3-4): 259-68.

## We're here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child's muscle-wasting condition, and let you know of progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences
- ▶ tell you about – and help you get – the services, equipment and support you're entitled to.

If you would like your GP or other health professional to have more information about FSHD, we have some relevant materials. We've developed an online training module for GPs, as well as one for physiotherapists working with adults with muscle-wasting conditions. Contact our helpline or email us to find out more.

If you have feedback about this factsheet or would like to request any references used to produce it, please email [info@muscular dystrophyuk.org](mailto:info@muscular dystrophyuk.org).

### Disclaimer

While every reasonable effort is made to ensure that the information in this document is complete, correct and up-to-date, this cannot be guaranteed and Muscular Dystrophy UK shall not be liable whatsoever for any damages incurred as a result of its use. Muscular Dystrophy UK does not necessarily endorse the services provided by the organisations listed in our factsheets.

## Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on **0800 652 6352** or [info@muscular dystrophyuk.org](mailto:info@muscular dystrophyuk.org) from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

## [www.muscular dystrophyuk.org](http://www.muscular dystrophyuk.org)

Version 3 / Date published: Nov 2000 / Original author: Dr P. W. Lunt, Consultant Clinical Geneticist / Updated: March 2017 / Updated by: Dr Chiara Marini Bettolo/ Date of review: March 2020  
Registered Charity No. 205395 and Registered Scottish Charity No. SC039445

