FAQs about FSHD

Short frequently asked questions with clear answers about FSHD

Prepared by the FSHD diagnosis working group, Spierziekten Nederland, March 2025.

*Information about children with FSHD is shown on a **blue** background.*

WHAT IS FSHD?

1. What is FSHD?

FSHD is an inherited muscle disease. The full name is facioscapulohumeral muscular dystrophy. People with this disease have a defect in their DNA. This defect causes the protein DUX4. This protein damages skeletal muscles. The muscles become smaller and weaker.

2. Why the name FSHD?

The disease was first described in 1886 by two French doctors (Landouzy & Dejerine). In the patients they saw, muscle weakness began in three areas:

- the face (facio),
- the shoulders (scapulo)
- and the upper arms (humeral).

That is why it is called facioscapulohumeral muscular dystrophy.

3. How common is FSHD?

FSHD is one of the most common inherited muscle diseases in the world. About 2,000 people have FSHD in the Netherlands. Around the world, there are about 870,000 people.

4. How many types of FSHD are there?

There are two main types:

- About 95% of people have FSHD type 1 (FSHD1).
- About 5% have FSHD type 2 (FSHD2).

Some people have FSHD with no known cause. Symptoms are roughly the same for all types of FSHD.

5. How does FSHD present in children?

In children, FSHD often begins with weak muscles in the face and shoulders.
This can cause fewer facial expressions. Children sometimes struggle to close their eyes.
The shoulder blades may also stick out (scapula alata).
Sometimes the muscles in the lower legs also become weaker. Children then get a drop foot and trip over more. The symptoms are different for each child. Some children have trouble with movement at an early age. In others, this happens later.
Children with severe FSHD sometimes have other symptoms too, such as:

- worse hearing;
- problems with their eyes;
- learning difficulties;
- or epilepsy.

6. What causes FSHD?

Almost all cases of FSHD1 are caused by the absence of certain pieces of DNA at the end of chromosome 4 (genetic location: 4q35). Because this chromosome is shorter, the muscles produce a more stable form of a certain protein: DUX4. This protein is bad for certain muscle cells (skeletal muscles). Muscle cells break down.
Not all muscles are affected at the same speed. Some muscles are more vulnerable than others. So, it is often easy to identify the pattern of muscle weakness.

In FSHD2, the abnormality is not on chromosome 4, but on chromosome 18. This also causes the production of the protein DUX4. So, the cause is different, but the symptoms are the same.

7. How do you know if you have FSHD?

If FSHD runs in your family, you will often recognize certain symptoms before they really bother you. You will usually notice them because your muscles become weaker. Movement will become more difficult. Many people also tire more quickly or have pain. They also often suffer from fatigue and/or pain. About two out of three people with FSHD suffer from it.

8. At what age do symptoms begin?

The first symptoms usually appear between 10 and 20 years of age. Sometimes they begin at pre-school age or not until after the age of 50.

9. What symptoms can you get?

With FSHD, the following muscles in particular become weaker:

- muscles in the face (near the eyes and mouth);
- shoulders and upper arm muscles;
- stomach muscles;
- and leg muscles.

The stomach muscles get weaker. You trip over more and are wobbly when you walk.
Getting up out of a chair or climbing stairs becomes more difficult. You also find it harder and harder to use your arms properly. Pain and fatigue are often a big problem.

The disease usually begins in the face and shoulders.
The abdominal muscles and the muscles that lift the feet are also often affected early.
It is difficult to predict whether the thighs and hip muscles will also become weaker. In half of people with FSHD, those muscles continue to work well.

Young children with FSHD find it hard to walk for longer periods of time. They often have chronic pain and tire easily. As a result, they may experience a poorer quality of life. Weak facial muscles mean that children also sometimes have difficulty speaking. Their voices sound softer or different. Speech problems are common in young children with FSHD.
Children often need a wheelchair and other aids if they have a lot of muscle weakness at a young age and this muscle weakness increases.

10. How is FSHD diagnosed and by whom?

A neurologist will examine you to see whether you have FSHD. To make the diagnosis, he or she will first perform a physical examination. The doctor will also check whether other family members have FSHD. Additional tests may then be done, such as:

- a blood test to measure the muscle enzyme CK. This enzyme is released when muscles are damaged.
- DNA test on a blood sample.

Sometimes other tests are needed, such as:

- electromyography (EMG) (to measure how active the muscles are);
- a microscopic examination of a piece of muscle tissue (biopsy);
- an MRI scan of the muscles;
- a muscle ultrasound.

GENETICS

1. Is FSHD genetic?

FSHD1 is an autosomal dominant genetic disorder. This means that if one parent has FSHD, their child has a 50% chance of also getting FSHD.
With FSHD2, it is more complicated. This involves two genetic factors that are passed on to the child separately. So, the chance of passing on FSHD2 to a child is slightly less than 50%.

2. Can FSHD also develop spontaneously?

Yes. Sometimes FSHD develops without any family history of it. This is called a new mutation.

3. What is mosaicism?

Sometimes FSHD develops in someone because of a mutation (change in genetic material) that has occurred after fertilization, during cell division in the early embryonic stage.
All cells that are created from that mutated cell then contain the same mutation. The other cells do not have the mutation. This is called mosaicism.
How many symptoms a person gets depends on:

- how many cells have the defect,
- and which cells contain the defect.

The chances of passing the mutation on to a child are then between 0% and 50%, depending on when the mutation occurred.

4. If I have no symptoms, can I still have the genetic mutation?

Yes, you can have the genetic mutation without having any symptoms.
With FSHD1, there is then a 50% chance of you passing it on to your child.
With FSHD2, this is harder to predict because it involves two genetic factors that are both 'inherited' independently.

5. How will the disease progress in my children?

This is hard to say. The disease can progress very differently from person to person, even within the same family. However, there is a link between repeat length in the DNA and disease severity. Disease symptoms are often more severe if the 'repeat' is lower.

6. Can I prevent my child from getting FSHD?

A test can be done during your pregnancy. This is called a prenatal test.
This is only possible for FSHD type 1 at the moment. For more information in Dutch, read the brochure Kinderwens bij een Spierziekte from Spierziekten Nederland.

7. Is embryo selection possible for FSHD?

Yes, this is possible for FSHD1. It is not yet available for FSHD2.
This test is called: PGT (Pre-implantation Genetic Test) or embryo selection. This test is done before you become pregnant, with IVF (in vitro fertilization). This is only available in the Netherlands if at least two family members have FSHD1, for example:

- you and a parent,
- or you and your child.

Even after a successful PGT procedure, we recommend an additional test during your pregnancy, e.g., chorionic villus sampling or amniocentesis.
For more information in Dutch, visit the website of the PGT center at MUMC+ (Maastricht): www.pgtnederland.nl.

8. Is FSHD equally common in men and women?

Yes, FSHD is just as common in men as it is in women.

TREATMENT

1. Do muscle strength exercises help with FSHD?

You may exercise your muscles as long as this is not too strenuous or extreme. But it usually does not help much with FSHD. However, aerobic exercise, such as walking or cycling, is good. This can help you feel less tired.

For children with FSHD, research is being carried out to see whether advice on movement and exercises for shoulder muscles can help. Movement is always important, including for children with FSHD.

2. Do people with FSHD have pain?

Pain is common with FSHD. The weakened muscles quickly become overworked. Other muscles then have to work harder and may also start to hurt.

Children with FSHD sometimes have less energy and a lower quality of life due to pain.

3. Does FSHD have anything to do with headaches?

Yes, it can. Morning headaches can occur if you do not get enough oxygen at night due to weak respiratory muscles. Muscles in the shoulders and neck can also cause tension headaches.

4. Can you reduce pain with supplements or homeopathy?

No. Pain with FSHD is often due to overworked muscles.
There are no known supplements or homeopathic remedies that are proven to help with pain from FSHD. You can, however, use regular painkillers (such as paracetamol), on your doctor's advice if applicable.

At the rehabilitation center, we will look at where the pain is coming from.
Then treatments may help, such as:

- physiotherapy;
- rest;
- or aids.

5. Will surgery help?

Surgery is sometimes performed for weak facial muscles. We know too little so far about the outcome to draw conclusions.
There is also surgery to stabilize the shoulder blade. This allows you to lift your arm better, but you usually cannot put any strain on it. It may look nicer, though.
This operation is not yet common in the Netherlands because little is known about the long-term outcome.

If you have any questions about this, please contact the FSHD Center of Expertise.

6. Is anesthesia risky?

Not usually. But it is important to tell the anesthetist that you have FSHD.
There may be risks if you have:

- a bent back;
- weak respiratory muscles;
- or problems with your lungs.

You should discuss this with the anesthetist before the operation. Spierziekten Nederland has an SOS card for this.

7. Is abdominoplasty possible with FSHD? Is a back brace a good idea?

It makes no sense to have abdominoplasty with FSHD because the stomach muscles are weakened and cannot be 'tightened'. A lumbar back brace (special support for the lower back) can sometimes help. For example, it may help you to stand and walk for longer. But a back brace can also be awkward for activities where you have to move your torso a lot. So, it depends on the situation.

8. Can I have laser treatment on my eyes?

People with FSHD often have dry eyes. This is because they cannot close their eyes as well. Dry eyes make laser eye surgery risky. So,

always discuss this with an ophthalmologist (specialist eye doctor) first.

9. Are there other eye problems besides dry eyes?

Sometimes a small abnormality develops in the blood vessels in the retina. This usually does not cause any symptoms.

Severe eye problems can occur in children with FSHD. These can lead to Coats disease. Children who are too young to say whether they can see well should see the ophthalmologist every year for a checkup.

10. Do hormones play a role in FSHD, such as during puberty or menopause?

There is no evidence that hormones affect FSHD. However, the disease does generally progress slightly more severely in men. Women lose muscle strength during menopause anyway. This sometimes makes it seem like the disease is progressing faster. But some women actually notice less decline during that time.

Adolescents with FSHD may develop a bent back (scoliosis) during their growth spurt. Adolescents may also have more difficulty accepting their disease. So, (extra) attention in care and good guidance is important.

11. Can you get breathing problems with FSHD?

The lung function of people with FSHD usually remains good. It may only be significantly reduced if you have a severe form of FSHD, are confined to a wheelchair and have severe scoliosis (curvature of the back). In that case, you may need help with breathing at night or to use a technique to help you cough better (such as air stacking).
The risks are also greater if you have another lung disease.
So, lung function should always be checked in people who are always in a wheelchair.

12. What is air stacking?

Air stacking is a technique in which air is blown into the lungs through a mask or mouthpiece. It helps you to cough better.
You can find more information in Dutch at www.vsca.nl/thuisbeademing.

13. Does FSHD go away on its own?

No, FSHD does not go away on its own.

14. Do children with FSHD sometimes have hearing problems?

Yes, about half of all children with FSHD hear less well, especially high-pitched sounds. Usually this is not so bad that help is needed. Children who are too young to say whether they can hear well should have their hearing tested every year.

15. Is there a link between FSHD and poor bowel function or bowel movements?

No. FSHD itself does not cause poor bowel function or bowel movements. But people with FSHD often move less. This can cause constipation. Weak stomach muscles can also play a role.

16. Does FSHD make your feet or legs get cold faster?

Yes, less movement and less muscle mass lead to less heat production. Fortunately, there are many aids to help you stay warm.

17. Is there a link between FSHD and sleep apnea?

Sleep apnea means that you sometimes stop breathing while you are asleep.
Not much research has been done yet, but it seems that sleep apnea is more common in people with FSHD.
If you sleep poorly, are often tired, are sleepy during the day, or wake up with a headache, talk to your doctor about this. A sleep study may be necessary.

LIVING WITH FSHD

1. What does the future look like with FSHD?

How many symptoms someone with FSHD has varies quite a lot from person to person. The loss of muscle strength and the consequences of that are usually the main focus. Many people with FSHD also have pain and are often tired. This can sometimes make daily tasks difficult.

Children with FSHD

Children with FSHD may have a lower quality of life due to physical limitations. They may also feel insecure about their appearance, and be afraid of social exclusion and uncertain about the future. Young people with FSHD really want to live like other young people. The fear of being excluded sometimes causes them to do things that are actually too hard for them.

2. What problems can you have if your facial muscles are weak?

- Swallowing: some people choke a lot. A speech therapist can help by teaching you some tricks.
- Chewing: chewing can be difficult, especially when you are tired. In that case, choose foods that are easier to chew.
- Biting your lip or cheek: having weak muscles in the mouth means that sometimes you accidentally bite your lip or cheek. Sometimes a permanent lump develops, which can be painful. Talk to your (rehabilitation) doctor about this.
- Eye irritation: if you cannot close your eyes properly, this can cause them to become dry or irritated. Eye drops or ointment may help. Sometimes eyelid surgery helps. Talk to your doctor.
- Communication problems: if you cannot use facial expressions well, people around you may not understand you properly. Try 'subtitling' yourself. In other words, explain the things you say in more detail. Always talk about this with the people around you.
- Difficulty speaking: your voice may become monotone and your pronunciation may become less clear when you are tired.

Communication with children

With children with FSHD, it is important to be aware that the face, which is essential for communication between the child and parents (or others), does not work as well. So, always check that you understand each other.

3. At what age do symptoms usually begin?

Although not everyone develops symptoms, symptoms usually begin between the ages of 10 and 20. But they can also begin at pre-school age or not until after the age of 50.

4. Should I tell my insurance company that FSHD runs in the family?

Yes, you must always answer these kinds of questions honestly.
The same applies to questions from the CBR (the Central Office for Motor Vehicle Driver Testing, which issues driver's licenses). If you have FSHD and you think it may affect your ability to drive, you should contact the CBR yourself.

5. How do I find the best care for FSHD?

There are many people who can help you cope with FSHD. It is good to get help from the following people:

- neurologist
- rehabilitation doctor
- physiotherapist
- occupational therapist
- dietician
- speech therapist
- social worker
- psychologist or remedial teacher

The FSHD Center of Expertise is located at Radboud university medical center (clinical) and Leiden University Medical Center (genetic and clinical). See also the Care Guide (in Dutch).

6. Does FSHD affect your life expectancy?
Basically, no. Life expectancy can sometimes be affected, but only if a severe form of FSHD develops in childhood.

7. Will I need a wheelchair later?
This varies from person to person. Some people have few symptoms or only mild symptoms; others have many symptoms. About 20% of people with FSHD use a wheelchair after the age of 50. It depends on your muscle weakness, but also on what you want to do in your daily life. A wheelchair makes activities possible that have become very difficult.

Most children with FSHD do not need a walking aid or wheelchair.

8. Can I be a donor if I have FSHD?
Yes, you can. For more information in Dutch, visit: www.transplantatiestichting.nl.

DRUG DEVELOPMENT

1. Is there already a drug for FSHD that slows, stops or cures the disease?
No, not at the moment.

2. When might there be a drug?
That is impossible to say. A lot of research is being done, in several countries. Some drugs are already being tested, but it may take 10 to 15 years more before a drug is ready and available to everyone.

3. What is a drug trial?
A drug trial is a study to see whether a new drug is safe (first phase) and whether it works well (second and third phases). The drug is tested first on a few people and then on larger groups. These trials are often done at multiple centers nationally and internationally at the same time.
FSHD drug trials are expected to take place first with adults, and – if proven safe – only then with children.

Drug trial with children
It is important to test drugs on children as well. That way doctors know whether they work, what the right dose is and if there are any side effects. If a drug has not been officially tested on children, it is often not covered by health insurance. Therefore, studies have to be carried out on children too so that they can use the drug safely and have access to it when it is needed.

4. How do I apply for future drug research (trials) in the Netherlands?
When a new drug trial begins, there is a call for participants. This call can come through:
- the FSHD registry;
- the doctor treating you;
- or Spierziekten Nederland.

If you register with the FSHD registry, you will be kept informed and researchers will be able to approach you more easily for participation in trials.

5. What is the FSHD registry?
This is a partnership between
- the FSHD Center of Expertise (LUMC and Radboud university medical center);
- the FSHD Foundation;
- and Spierziekten Nederland.

If you register, researchers can find you for participation in scientific research and drug trials: www.fshdregistratie.nl.

6. Who can participate in these trials?
Each trial has rules on who can and cannot participate. These rules are called inclusion and exclusion criteria.
Examples include:
- an official FSHD diagnosis (genetic),
- a certain age,
- a certain height-to-weight ratio (BMI),
- the stage of the disease,
- the FSHD type.

7. What is trial fitness?

Trial fitness means that you must be in good condition to participate in a trial. Sometimes you cannot participate if you:

- are overweight;
- use certain medications;
- or have other diseases.

8. Where can I find more information about participating in drug research?

You can watch a webcast in Dutch on this topic on the website of Spierziekten Nederland: www.spierziekten.nl. For more information, contact your own doctor, national patient organization or visit the website of FSHD Europe FSHD Europe - The European Voice for People with FSHD or the FSHD Society www.fshdsociety.org.

9. Will the cost of a new drug be refunded by basic health insurance?

Drugs for rare diseases are often very expensive. If a drug is approved by the European Medicines Agency (EMA), each country must decide for itself whether to refund its cost. In the Netherlands, this is often a slow and complicated process. Spierziekten Nederland is involved in this process and is committed to getting drugs refunded and available to as many people as possible as quickly as possible.

10. If you cannot participate in a trial, will you not get the drug later either?

Not always. When a new drug is approved, people look at the results of the trials. Only the group for whom the drug is proven to work is usually refunded for the cost of the drug later.

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MORE INFORMATION

This FAQs document is based on the Dutch version made by the FSHD patient group of Spierziekten Nederland in collaboration with multiple medical advisors of the FSHD Center of Expertise (Radboud university medical center and LUMC) and information from FSHD Europe FSHD Europe - The European Voice for People with FSHD and the FSHD Society www.fshdsociety.org. For more information, contact your own doctor, national patient organization or FSHD Europe (info@fshd-europe.info).

SPIERZIEKTEN NEDERLAND

FSHD DATABANK & REGISTRATIE

LUMC Leids Universitair Medisch Centrum

Radboudumc

FSHD STICHTING

FSHD Europe

CBF ERKEND GOED DOEL

Algemeen Nut Beogende Instelling ANBI

De FSHD Stichting is gerangschikt als ANBI.
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