CONDITION

Facioscapulohumeral muscular dystrophy

An inherited muscle condition that mainly weakens the face, shoulders, and upper arms.

WHAT IT IS
Inherited muscle condition causing patterned, progressive weakness

BODY SYSTEM
Muscles

MAIN SYMPTOMS
Facial weakness, shoulder-blade winging, upper-arm weakness, foot drop

WHAT CAUSES IT
Inherited genetic changes allowing abnormal DUX4 activity

HOW IT IS DIAGNOSED
Genetic testing of chromosome 4 and related genes

MAIN TREATMENT
Rehabilitation, symptom control, mobility and breathing support

CAN IT BE CURED?
No - genetic cause cannot yet be reversed

IS IT AN EMERGENCY?
Sometimes - sudden severe breathing or choking problems

LONG-TERM OUTLOOK
Usually slowly progressive, with severity differing widely

Overview

Facioscapulohumeral muscular dystrophy, or FSHD, is an inherited condition that causes muscle weakness. Its name describes the areas often affected: the face, shoulder blades, and upper arms.

FSHD is a type of muscular dystrophy. Muscular dystrophies are genetic conditions in which muscles become weaker over time. In FSHD, weakness often starts during the teenage years, but it can begin in childhood or later adulthood.

The pattern is different from person to person. One side of the body may be weaker than the other. Some people have mild symptoms that change very slowly. Others develop more widespread weakness affecting their trunk, hips, or lower legs.

FSHD usually does not affect thinking, sensation, or control of the bladder and bowel. Most

people have a typical lifespan, although uncommon breathing complications can be serious. Early assessment can help with mobility, pain, fatigue, breathing, hearing, and eye problems.

Symptoms

The first signs may be subtle. You might have difficulty whistling, drinking through a straw, closing your eyes firmly, or making certain facial expressions. Some people sleep with their eyes partly open because the eyelid muscles are weak.

Weak muscles around the shoulder blades can cause scapular winging . This means one or both shoulder blades stick out from the back. It may become more obvious when you lift your arms or push against a wall. Reaching above shoulder height may become difficult.

Other possible symptoms include:

Muscle weakness is often uneven. For example, one shoulder or one ankle may be affected years before the other.

Some people develop hip and thigh weakness. A smaller number eventually need a wheelchair for some or all mobility. Severe weakness beginning in early childhood is more likely to affect several parts of the body.

Hearing loss can occur, particularly in people whose symptoms began early. Changes in the small blood vessels at the back of the eye are also possible. These usually cause no symptoms, but severe retinal changes can threaten vision.

Breathing muscle weakness is uncommon. Its risk is higher when weakness is extensive, the spine is curved, or a wheelchair is needed.

- Weakness in the upper arms, often greater on one side
- Sloping shoulders or collarbones that appear unusually straight
- Weak stomach and back muscles, affecting posture
- An exaggerated inward curve of the lower back
- Difficulty lifting the front of the foot, called foot drop
- Tripping, falls, or difficulty climbing stairs
- Muscle or joint pain
- Fatigue, especially after physical effort

Causes and risk factors

FSHD happens when genetic changes allow a gene called DUX4 to become active in muscle cells. DUX4 is normally switched off in adult muscles. When it is active, it produces a protein that can damage muscle cells.

There are two main genetic types.

In FSHD1 , a repeated section of DNA on chromosome 4 is shorter than usual. This change must occur alongside a chromosome pattern that permits DUX4 activity.

In FSHD2 , the repeated section is not shortened in the same way. Instead, changes in genes that normally keep this region switched off allow DUX4 to become active. The gene most often involved is called SMCHD1 .

FSHD is commonly inherited in an autosomal dominant pattern. This means a person with the relevant genetic change can pass it to a child. Each pregnancy has its own chance of inheritance. However, having the genetic change does not predict exactly when symptoms will begin or how severe they will be.

A person may also be the first in a family to have FSHD. This can happen because of a new genetic change or because a parent has very mild or unrecognised symptoms. Some people have mosaicism , meaning the genetic change is present in only some of their cells.

Diagnosis

A clinician may suspect FSHD from its characteristic pattern of weakness. The examination usually looks at facial movement, shoulder-blade position, arm strength, posture, walking, and ankle movement. Your personal and family history also helps.

Genetic testing is used to confirm the diagnosis. Testing for FSHD1 looks at the repeated DNA region on chromosome 4 and whether it has the pattern that allows DUX4 activity. If this does not confirm FSHD but the clinical pattern remains suggestive, testing may examine DNA methylation and genes linked to FSHD2. Methylation is a chemical marking that helps switch genes on or off.

Other tests may include:

Creatine kinase may be normal or only mildly raised in FSHD. A muscle biopsy, which removes a small tissue sample, is now less commonly needed. It may be considered when genetic results do not explain the symptoms or another muscle condition is possible.

Genetic counselling can help you understand the testing process, uncertain results, family implications, and reproductive choices.

- A blood test for creatine kinase, an enzyme released by muscle
- Electrical testing of muscles, called electromyography
- Breathing tests to measure lung function
- Hearing or eye assessment when appropriate

Treatment and management

There is currently no cure that reverses the genetic cause of FSHD. Treatment focuses on maintaining function, controlling symptoms, and identifying complications. Research into treatments that reduce DUX4 activity or protect muscle is continuing.

A physiotherapist can assess strength, flexibility, posture, balance, and walking. A suitable programme may include gentle stretching and low- to moderate-intensity aerobic activity. Exercise should be adjusted to your abilities. Severe pain, prolonged exhaustion, or a major loss of function after activity are signs that the level may need review.

An occupational therapist can suggest ways to make daily tasks easier. Adaptations may include changes to seating, workspaces, bathing, dressing, or kitchen equipment.

Other management options include:

Surgery to secure a shoulder blade to the chest wall may help carefully selected people lift an arm more effectively. It does not restore muscle strength and can reduce shoulder movement in other directions. Assessment by a team experienced in FSHD is important before considering it.

If eyelid weakness prevents full closure, eye care may be needed to prevent dryness or injury. This is different from the retinal blood-vessel changes that can occur in FSHD.

- An ankle-foot brace for foot drop or ankle instability
- Walking aids, powered mobility, or a wheelchair when useful
- Pain assessment and treatments suited to the cause
- Support for fatigue, sleep, and mental wellbeing
- Hearing aids when hearing loss affects daily life
- Breathing support if respiratory muscles become weak

Complications

Weakness may lead to falls, reduced mobility, and loss of independence with some activities. Poor shoulder mechanics can contribute to pain. Weak stomach and back muscles may affect posture or lead to spinal curvature.

Foot drop can make the toes catch on the floor. An orthosis, which is a brace supporting the foot and ankle, may improve safety and walking efficiency.

Breathing complications are not common, but they matter when present. Weak breathing muscles may cause shallow breathing during sleep. Possible signs include morning headaches, disturbed sleep, daytime sleepiness, poor concentration, or breathlessness when lying flat.

Severe retinal blood-vessel changes can cause fluid leakage or vision loss. Hearing loss may affect speech development in young children if it is not recognised.

Swallowing problems are usually mild, but some people cough during meals or have difficulty managing certain foods. Repeated choking or chest infections need assessment.

Prevention

FSHD cannot currently be prevented in someone who has the genetic changes that cause it. Lifestyle choices do not cause FSHD, and neither the affected person nor their parents are responsible for it.

Genetic counselling can explain the chance of passing the condition on. People planning a pregnancy may choose to discuss options such as testing during pregnancy, testing embryos created through in vitro fertilisation, donor eggs or sperm, adoption, or pregnancy without genetic testing. These are personal choices rather than required steps.

Falls and secondary problems can sometimes be reduced through appropriate footwear, braces, mobility aids, home adaptations, and review of changes in strength.

Living with it and what to expect

FSHD usually progresses slowly over years. Symptoms may seem stable for a long time and then become more noticeable. The speed and pattern cannot be predicted from another family member's experience, even when they share the same genetic change.

Many people continue to work, study, raise families, drive, and take part in social and physical activities. Some need changes to their schedule or environment. Others use mobility equipment to conserve energy rather than because they cannot walk at all.

Facial weakness may affect expression even when your emotions are unchanged. Explaining

this to family, friends, teachers, or coworkers can reduce misunderstandings.

Regular follow-up is used to review mobility, pain, falls, daily activities, and any changes in breathing. Hearing and eye monitoring may be particularly important for children and people with early or more severe symptoms.

Planning ahead can help you stay independent. This might include workplace adjustments, accessible housing, mobility support, or decisions about driving. Emotional support can also be useful when symptoms change or when family members are considering genetic testing.

When to get medical help

Arrange a routine appointment if you develop new or gradually worsening weakness, frequent tripping, shoulder-blade winging, or difficulty lifting your arms. If you already have FSHD, report changes in walking, falls, pain, hearing, vision, sleep, swallowing, or ability to manage daily tasks.

Seek prompt medical advice for:

Call emergency services for severe difficulty breathing, blue or grey lips, choking that does not clear, loss of consciousness, or another immediately life-threatening problem.

- New breathlessness, especially when lying flat
- Morning headaches or marked daytime sleepiness
- Repeated coughing or choking while eating
- Sudden or rapidly worsening vision changes
- A clear decline in walking or repeated falls

Common questions

Is FSHD fatal?

FSHD is not usually life-limiting. Most people have a typical lifespan. Rare severe breathing weakness can affect health and needs monitoring and treatment.

Will I eventually need a wheelchair?

Not everyone does. Some people remain able to walk throughout life, while others use a wheelchair for distance, energy conservation, or full-time mobility. Your current symptoms cannot predict the exact course.

Can FSHD skip a generation?

It can appear to skip one because some people with the genetic change have very mild or unnoticed symptoms. Genetic testing and counselling can help clarify the pattern in a family.

Is exercise safe with FSHD?

Appropriate exercise is generally encouraged. Aerobic activity and gentle strengthening may support fitness and function, but the programme should match your abilities. Activity that causes severe pain or prolonged exhaustion needs review.

Does FSHD affect intelligence?

FSHD does not usually affect intelligence or thinking. Fatigue, poor sleep, pain, or hearing loss can still make concentration and learning more difficult.

Is FSHD the same as ALS?

No. FSHD is an inherited muscle condition linked to abnormal DUX4 activity. ALS mainly damages the nerve cells that control muscles and usually follows a different course.

Sources

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