CONDITION

Familial dysautonomia

Familial dysautonomia is an inherited condition that disrupts nerves controlling sensation and automatic body functions.

WHAT IT IS
Rare inherited condition affecting sensory and automatic nerve function

BODY SYSTEM
Nervous, breathing, digestive, heart and circulation systems

MAIN SYMPTOMS
Blood pressure swings, vomiting crises, poor balance, reduced pain sensation

WHAT CAUSES IT
Inherited ELP1 gene changes

HOW IT IS DIAGNOSED
Genetic testing finding two pathogenic ELP1 variants

MAIN TREATMENT
Symptom control, respiratory support, nutrition, and rehabilitation

CAN IT BE CURED?
No - inherited nerve damage has no cure

IS IT AN EMERGENCY?
Sometimes - breathing trouble, choking, or prolonged vomiting need urgent care

LONG-TERM OUTLOOK
Lifelong condition with progressive breathing, vision, and mobility problems

Overview

Familial dysautonomia, often shortened to FD, is a rare inherited condition affecting the nervous system. It is also classified as hereditary sensory and autonomic neuropathy type III.

The autonomic nervous system controls functions that normally happen without conscious effort. These include blood pressure, breathing, digestion, sweating, tears, and body temperature. Sensory nerves carry information about pain, heat, cold, taste, and the position of the body.

In FD, these nerves do not develop or work normally. Symptoms usually begin in infancy, although their type and severity differ from one person to another. The condition continues throughout life and may cause additional problems over time.

FD matters because the body may not respond normally to illness, injury, low oxygen, or changes in blood pressure. Someone may have a serious injury without much pain or may develop low oxygen without appearing very short of breath. Regular specialist care helps identify these problems early.

Symptoms

Early signs may include difficulty feeding, weak sucking, coughing or choking with feeds, poor weight gain, and reduced muscle tone. Babies may produce few or no tears when crying. Tears may increase somewhat with age, but dry eyes often remain a problem.

Common symptoms during childhood and adulthood include:

Some people have autonomic crises. These are episodes involving severe nausea or repeated retching, sweating, red or blotchy skin, agitation, and a sharp rise in blood pressure. Illness, pain, emotional stress, surgery, or lack of sleep may trigger a crisis. Sometimes no clear trigger is found.

Symptoms can be misleading. Reduced pain does not protect the body from injury, and reduced breathlessness does not mean oxygen levels are safe.

- Rapid or extreme changes in blood pressure and heart rate
- Dizziness or fainting when standing
- Poor balance, an unsteady walk, and reduced coordination
- Reduced ability to feel pain, heat, or cold
- Reduced taste and fewer visible taste buds on the tongue
- Dry eyes and damage to the eye surface
- Swallowing problems, reflux, or slow movement of food through the stomach
- Repeated coughing, chest infections, or breathing problems during sleep
- Abnormal sweating and difficulty controlling body temperature
- Curvature of the spine, known as scoliosis
- Joint injuries or fractures that cause less pain than expected

Causes and risk factors

Familial dysautonomia is caused by harmful variants in the ELP1 gene. This gene helps nerve

cells develop and survive. Changes that greatly reduce normal ELP1 protein particularly affect sensory and autonomic nerves.

FD follows an autosomal recessive inheritance pattern. This means a person has inherited one altered ELP1 copy from each biological parent. Parents who each carry one altered copy usually do not have the condition.

When both parents are carriers, each pregnancy has its own chance of producing a child with FD, a child who is a carrier, or a child who inherited neither altered copy. A genetic counsellor can explain these chances without making assumptions about family plans.

FD occurs most often in people with Ashkenazi Jewish ancestry because certain ELP1 variants became more common within that population over many generations. However, people from other backgrounds can also have the condition. Ancestry alone cannot confirm or exclude it.

Having an affected sibling or known ELP1 variants in the family increases the chance of FD.

Diagnosis

Symptoms and examination findings may suggest familial dysautonomia. These can include reduced tears, absent or reduced reflexes, an unusually smooth tongue, reduced response to pain, blood pressure instability, and feeding or breathing problems.

The diagnosis is confirmed by molecular genetic testing. A blood or saliva sample is tested for two pathogenic, or harmful, ELP1 variants-one on each copy of the gene. Broader testing may be needed if a person does not carry the ELP1 variants most often found in people with Ashkenazi Jewish ancestry.

Genetic testing also helps distinguish FD from other inherited nerve conditions and other causes of autonomic symptoms. Relatives may be offered testing for known family variants.

After diagnosis, tests are used to assess the condition rather than confirm it. These may include swallowing studies, breathing and sleep tests, eye examinations, spine imaging, kidney tests, hearing tests, and checks of blood pressure while lying down and standing.

Treatment and management

There is no cure that corrects the inherited nerve changes. Treatment focuses on controlling symptoms, preventing avoidable injury, and supporting breathing, nutrition, movement, and daily life.

Care is usually coordinated across several specialties. The plan may involve neurology,

respiratory medicine, gastroenterology, cardiology, kidney care, ophthalmology, orthopaedics, rehabilitation, nutrition, and clinical genetics.

Breathing care may include airway-clearance techniques, treatment of respiratory infections, sleep monitoring, and breathing support when needed. Swallowing assessment can identify food or liquid entering the airway, called aspiration. Changes to food texture, feeding methods, or feeding-tube support may be considered when swallowing is unsafe or nutrition is poor.

Blood pressure management must balance two problems: low pressure when upright and high pressure when lying down or during a crisis. Measures may include careful hydration, changes in position, physical supports, and medication. Because a treatment that raises standing pressure may worsen pressure while lying down, monitoring is important.

Autonomic crises are treated according to their trigger and severity. Care may include a quiet setting, fluids, control of nausea or retching, and medicines that reduce excessive autonomic activity. A written crisis plan can help families, schools, and emergency teams respond consistently.

Lubricating eye treatments help protect dry eyes. Regular eye examinations are important because reduced corneal sensation may make scratches less noticeable.

Physical and occupational therapy may support balance, strength, posture, equipment needs, and safer daily activities. Scoliosis may need monitoring, bracing, or surgery. Skin, joints, and feet should be checked regularly because injuries may not hurt normally.

Complications

Swallowing problems can allow saliva, food, or stomach contents to enter the lungs. Repeated aspiration may cause pneumonia and long-term lung damage. Weak breathing responses and sleep-related breathing problems can also lead to low oxygen.

Blood pressure swings may cause fainting and falls. Repeated high blood pressure, combined with difficulty regulating fluids, can contribute to kidney and heart problems.

Reduced pain and temperature sensation increase the risk of unnoticed burns, cuts, fractures, pressure injuries, and joint damage. A swollen joint or inability to use a limb may be more important than the amount of pain.

Progressive scoliosis can affect posture, mobility, and breathing. Balance difficulties may increase as a person gets older.

Some people develop optic neuropathy, which is damage to the nerve carrying visual information from the eye to the brain. This can cause gradual and sometimes substantial sight

loss.

FD can shorten life expectancy, particularly when severe breathing, aspiration, kidney, or autonomic complications occur. Monitoring and modern supportive care have helped more people reach adulthood, but serious complications remain possible.

Prevention

Familial dysautonomia itself cannot be prevented through lifestyle changes. It is caused by inherited ELP1 variants present from conception.

Carrier screening can show whether someone carries a common ELP1 variant, especially before or during pregnancy. If variants are known in a family, genetic counselling can explain reproductive options, including prenatal testing and testing of embryos created through in vitro fertilisation. These are personal choices rather than requirements.

Some complications may be reduced through regular respiratory, swallowing, eye, kidney, spine, and blood pressure monitoring. Avoiding smoke exposure, checking food and drink temperatures, using protective footwear, and inspecting the skin can lower specific risks.

Living with it and what to expect

FD changes over time. Feeding and growth problems often dominate infancy. During childhood, recurrent respiratory infections, poor coordination, blood pressure changes, and scoliosis may become more noticeable. In adolescence and adulthood, mobility, vision, kidney function, and breathing may require closer support.

Not every symptom steadily worsens. Some remain stable for long periods, while others appear during illness or after a period of relative stability. Autonomic crises may become less frequent in some adults, but blood pressure and breathing regulation can still be difficult.

Many people attend school, take part in family and community life, and pursue work or interests with suitable support. Learning ability is not necessarily impaired, although missed schooling, vision changes, fatigue, illness, or attention difficulties may affect education.

Daily routines often include medicines, eye care, airway clearance, feeding support, physical therapy, or equipment for mobility. Planning for procedures and hospital visits is especially important because pain, oxygen levels, blood pressure, and temperature may behave differently than expected.

Regular follow-up is usually lifelong. Appointments commonly monitor breathing, swallowing,

nutrition, blood pressure, kidney function, vision, joints, and the spine. A portable medical summary and emergency plan can explain the diagnosis and usual responses to illness.

When to get medical help

Call emergency services for:

Seek prompt medical assessment for a new cough, fever, noisy breathing, reduced oxygen level, unusual sleepiness, or a clear change in breathing. These may be signs of aspiration or infection even when breathlessness is mild.

Repeated vomiting, much less urine, a very dry mouth, or worsening dizziness may indicate dehydration. A red eye, new light sensitivity, or change in vision also needs prompt assessment because eye injuries may not be painful.

Arrange routine follow-up for gradual changes in walking, posture, swallowing, eyesight, sleep, blood pressure patterns, or weight. These are not usually emergencies, but they may require changes to ongoing support.

- Severe difficulty breathing, blue or grey lips, breathing pauses, or unresponsiveness
- Choking followed by ongoing breathing difficulty
- A prolonged autonomic crisis with repeated retching, confusion, collapse, or inability to keep down fluids
- A serious burn, head injury, major fall, or possible fracture, even with little pain
- Sudden weakness, loss of consciousness, or another abrupt neurological change

Common questions

Is familial dysautonomia the same as dysautonomia?

No. Dysautonomia is a broad term for problems involving the autonomic nervous system. Familial dysautonomia is one specific inherited form caused by harmful ELP1 variants.

Can familial dysautonomia be cured?

There is currently no cure that restores normal sensory and autonomic nerve development. Treatment can control many symptoms, support daily function, and reduce some complications.

Can someone with familial dysautonomia feel pain?

Most people can feel some pain, but their response is reduced or unusual. Heat, fractures, eye

injuries, or deep wounds may cause less discomfort than expected. Visible injury or loss of function should not be ignored because pain is mild.

Why is familial dysautonomia linked to Ashkenazi Jewish ancestry?

A small number of ELP1 variants became more common in this population through a founder effect. This happens when a gene variant carried by a relatively small ancestral group becomes more frequent in later generations. FD can still occur in people with other backgrounds.

Does familial dysautonomia affect intelligence?

FD does not automatically cause intellectual disability. Education may still be affected by recurrent illness, hospital stays, vision problems, attention difficulties, or fatigue. Support should be matched to the individual's needs.

Can other family members be tested?

Yes. Once the ELP1 variants in a family are known, relatives can have targeted carrier or diagnostic testing. Genetic counselling can explain what a result means for the person tested and for future pregnancies.

Sources

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