CONDITION

Chanarin dorfman disease

Learn about Chanarin-Dorfman disease, a rare inherited disorder that affects fat storage and may involve the skin, liver, muscles, hearing, eyes, and nervous system.

WHAT IT IS
Rare inherited fat storage disorder

BODY SYSTEM
Skin, liver, muscles and nervous system

MAIN SYMPTOMS
Dry scaly skin, enlarged liver, muscle weakness, hearing loss

WHAT CAUSES IT
Inherited ABHD5 gene changes

HOW IT IS DIAGNOSED
ABHD5 molecular genetic testing

MAIN TREATMENT
Supportive symptom and organ care

CAN IT BE CURED?
No - no treatment corrects gene change

IS IT AN EMERGENCY?
Sometimes - vomiting blood or black stools

LONG-TERM OUTLOOK
Lifelong and variable, sometimes progressing to cirrhosis

Overview

Chanarin-Dorfman disease is a rare inherited disorder in which the body cannot properly break down certain stored fats called triglycerides. It is also known as neutral lipid storage disease with ichthyosis .

As a result, fat droplets collect inside cells throughout the body. The skin is almost always affected. The liver, muscles, ears, eyes, and nervous system may also be involved. The pattern and severity can vary widely, even among people from the same family.

A common skin feature is ichthyosis , meaning persistently dry, thick, or scaly skin. It often begins at birth or in early infancy. Some babies are born with a tight, shiny membrane covering the skin, known as a collodion membrane.

Liver involvement is also common. Fat may build up in the liver, causing an enlarged liver or abnormal liver blood tests. In some people, this remains stable. In others, it can lead to ongoing inflammation and scarring.

Chanarin-Dorfman disease is lifelong. There is no established cure, but treatment can relieve skin symptoms, support nutrition and mobility, and address complications. Regular monitoring is important because internal organ changes may develop even when a person feels well.

Symptoms

Symptoms may appear at birth, during childhood, or later. Not everyone has every feature.

Skin findings may include:

The liver may become enlarged because of fat accumulation. This may cause no obvious symptoms at first. Possible findings include abdominal fullness, abnormal liver enzyme results, or fatty liver seen on imaging. Less commonly, ongoing liver injury may cause jaundice, in which the skin or whites of the eyes turn yellow.

Muscle involvement can cause weakness, reduced endurance, muscle pain, or difficulty climbing stairs and rising from a chair. Weakness may progress slowly. Blood levels of creatine kinase, an enzyme released from muscle, may be elevated.

Other possible features include:

Symptoms can overlap with other inherited skin, liver, or muscle disorders. Having one or more of these findings does not by itself establish the diagnosis.

- Dry, scaly, or thickened skin over much of the body
- Redness and scaling that are more noticeable in skin folds
- Itching, discomfort, or cracking
- Reduced sweating and difficulty tolerating heat
- A collodion membrane at birth
- Hair thinning or patchy hair loss in some people
- Hearing loss, often related to the inner ear
- Cataracts or other eye problems
- Poor balance, coordination problems, or an unusual walking pattern
- Developmental delay or learning difficulties in some people

- Short stature
- Enlarged spleen
- Numbness or weakness related to peripheral nerve involvement

Causes and risk factors

Chanarin-Dorfman disease is caused by disease-related changes in the ABHD5 gene, which is also called CGI-58 . This gene provides instructions for a protein that helps cells release fatty acids from stored triglycerides. When the protein does not work properly, neutral fats remain trapped in small structures called lipid droplets.

The condition follows an autosomal recessive inheritance pattern. This means a person usually has a disease-causing change in both copies of the ABHD5 gene, one inherited from each biological parent.

Parents who each carry one altered copy generally do not have the disease. When both parents are carriers, each pregnancy has:

These chances apply separately to each pregnancy. Genetic counseling can help families understand test results, inheritance, and reproductive options.

The condition can occur in any population. It has been reported more often in some communities where marriage between biological relatives is more common. Having affected relatives or known ABHD5 variants in the family increases the likelihood of the disorder.

Diet or lifestyle does not cause Chanarin-Dorfman disease. However, alcohol, certain medicines, and other health conditions may add stress to an already affected liver.

- A 25% chance that the child will inherit both altered copies and have the condition
- A 50% chance that the child will inherit one altered copy and be a carrier
- A 25% chance that the child will inherit neither altered copy

Diagnosis

Diagnosis usually begins with a medical history and physical examination. A clinician may ask about skin changes since birth, muscle weakness, hearing, vision, development, liver problems, and similar findings in relatives.

One helpful test is a blood smear examined under a microscope. In Chanarin-Dorfman disease, certain white blood cells may contain visible fat droplets. This finding is called Jordan anomaly .

It strongly supports a neutral lipid storage disorder, but genetic testing is generally needed to confirm the specific cause.

Molecular genetic testing looks for disease-causing variants in both copies of the ABHD5 gene. Testing may use a single-gene test, a panel covering several genes associated with ichthyosis or lipid storage disorders, or broader testing such as exome sequencing.

Additional tests help determine which organs are affected. These may include:

A clinician may use specialized imaging to measure liver fat or stiffness. Liver stiffness can be a sign of fibrosis, which means scar tissue. A liver biopsy is not required for every person, but it may be considered when the cause or severity of liver disease remains uncertain.

The diagnostic process also considers other causes of congenital ichthyosis, fatty liver, muscle weakness, and abnormal fat storage. Identifying the genetic cause can prevent repeated testing and allows relatives to consider targeted testing.

- Liver blood tests and tests of liver function
- Creatine kinase and other muscle-related blood tests
- Abdominal ultrasound or another form of liver imaging
- Hearing tests
- Eye examinations
- Neurologic and muscle assessments
- Developmental or learning evaluations when appropriate

Treatment and management

There is no established treatment that corrects the underlying ABHD5 gene change. Care is therefore directed toward protecting affected organs, reducing symptoms, and supporting everyday function. Because the disorder can involve several body systems, care may include dermatology, liver, genetics, neurology, nutrition, hearing, eye, and rehabilitation specialists.

Skin care is usually needed throughout life. Thick, fragrance-free moisturizers and ointments help hold water in the skin. They often work best when applied soon after bathing. Products that soften and loosen scale, called keratolytics, may be useful under medical guidance. Strong products can irritate the skin, especially in infants or on cracked areas.

Prescription retinoids, which are medicines related to vitamin A, may improve severe scaling in selected people. However, they can affect the liver, blood fats, bones, and pregnancy. Their use requires specialist assessment and laboratory monitoring, particularly when liver disease is

already present.

Dietary approaches have included limiting some long-chain dietary fats and using medium-chain triglycerides, which are handled differently by the body. Evidence for long-term benefit is limited, and this approach does not remove fat already stored in all tissues. A restrictive diet should not be started without a clinician and dietitian because children and adults still need enough calories, essential fatty acids, protein, vitamins, and minerals.

Liver care focuses on monitoring and reducing additional sources of injury. A clinician may review medicines, supplements, alcohol use, weight, blood sugar, and cholesterol. Medicines or supplements marketed for "liver cleansing" should not be assumed to be safe. Advanced liver disease is managed by a liver specialist. Liver transplantation has been reported in severe cases, but it does not correct the genetic condition in other organs.

Other supportive treatments may include:

Genetic counseling is an important part of care. It can help explain family testing and the limits and implications of genetic results.

- Physical therapy and appropriate exercise for strength and mobility
- Occupational therapy for daily activities
- Hearing aids or other hearing support
- Cataract treatment when vision is affected
- Educational or developmental services
- Treatment of itching, skin infection, pain, or other specific symptoms

Complications

Liver disease is one of the most important possible complications. Fat accumulation may progress to steatohepatitis, meaning fatty liver with inflammation. Continued injury can cause fibrosis and, in some people, cirrhosis. Cirrhosis is advanced liver scarring that can interfere with blood flow and liver function.

Skin scaling can lead to painful cracks and increase the chance of bacterial infection. Reduced sweating may make overheating more likely, especially during hot weather, fever, or vigorous activity.

Progressive muscle or nerve involvement may affect walking, balance, endurance, and independence. Falls may become more likely if weakness, poor coordination, vision problems, or hearing loss are present together.

Hearing loss can affect speech development, school performance, work, and communication if it is not recognized. Cataracts and other eye findings may reduce vision.

Routine monitoring is intended to identify these problems early. Not every person develops severe complications.

Seek prompt medical advice for new jaundice, increasing abdominal swelling, dark urine, very pale stools, rapidly worsening weakness, a sudden change in hearing or vision, or signs of an infected skin crack such as spreading redness, warmth, drainage, or fever.

Seek emergency care for vomiting blood, black or bloody stools, severe breathing difficulty, fainting, new confusion, extreme drowsiness, or sudden inability to move an arm or leg. These symptoms are not specific to Chanarin-Dorfman disease, but they require immediate evaluation.

Follow up

Follow-up should be individualized according to age, symptoms, and the organs involved. Even people with mild skin symptoms may need periodic liver and muscle assessment.

Routine visits may review skin care, nutrition, growth, strength, walking, hearing, vision, school or work needs, and emotional well-being. Blood testing may track liver enzymes, liver function, blood counts, muscle enzymes, glucose, and blood fats. The exact tests and timing depend on the person's medical history.

Liver imaging may be repeated when clinically appropriate. Hearing and eye checks can identify gradual changes that may not be obvious at home. Children may also need growth and developmental monitoring.

Before starting a new medicine, over-the-counter product, or supplement, patients should tell the clinician or pharmacist about their liver condition. Medical teams should also be informed before surgery because skin care, mobility, nutrition, and liver function may affect planning.

Relatives may wish to discuss carrier or targeted genetic testing after a familial ABHD5 variant has been identified. Genetic results should be interpreted with a genetics professional whenever possible.

Sources

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