CONDITION

Chylomicronemia syndrome

Learn how chylomicronemia syndrome causes extremely high triglycerides, how it is diagnosed, and how diet, medicines, and follow-up can reduce complications.

WHAT IT IS
Group of disorders causing extreme triglyceride elevation

BODY SYSTEM
Blood and circulation

MAIN SYMPTOMS
Upper abdominal pain, nausea or vomiting, eruptive xanthomas, repeated pancreatitis

WHAT CAUSES IT
Inherited or genetic plus acquired factors

HOW IT IS DIAGNOSED
Repeated fasting lipid panels

MAIN TREATMENT
Low-fat diet, contributing-factor management, triglyceride-lowering medicines

CAN IT BE CURED?
No, managed with diet and medicines

IS IT AN EMERGENCY?
Sometimes, with severe or worsening abdominal pain

LONG-TERM OUTLOOK
Treatment reduces risk, but repeated pancreatitis can cause lasting complications

Overview

Chylomicronemia syndrome is a group of disorders in which very large amounts of chylomicrons remain in the blood. Chylomicrons are particles that carry triglycerides, a type of fat, from food through the bloodstream. They normally disappear from the blood within several hours after eating.

In chylomicronemia, the body cannot clear these particles normally. Fasting triglyceride levels often remain at or above 1,000 milligrams per deciliter, or 11.3 millimoles per liter. Levels may vary, especially when the condition is affected by diet, alcohol, diabetes, illness, pregnancy, or medicines.

The main health concern is acute pancreatitis , which is sudden inflammation of the pancreas. Pancreatitis can cause severe abdominal pain and may require hospital treatment. Preventing

pancreatitis is a major goal of care.

There are two main forms:

Distinguishing these forms matters because they do not always respond to the same treatments. FCS often responds poorly to standard triglyceride-lowering medicines, while treating contributing conditions can greatly improve MCS.

- Familial chylomicronemia syndrome , or FCS, is usually caused by inherited changes affecting the pathway that clears chylomicrons. It is rare and may begin in childhood, adolescence, or early adulthood.
- Multifactorial chylomicronemia syndrome , or MCS, results from a combination of genetic susceptibility and other factors that raise triglycerides. These may include diabetes, excess weight, alcohol, pregnancy, certain illnesses, and some medicines.

Symptoms

Some people have no clear symptoms until a blood test finds very high triglycerides. Others develop symptoms when triglyceride levels rise further or pancreatitis occurs.

Possible features include:

Eruptive xanthomas are deposits of fat in the skin. They may appear suddenly and can improve when triglyceride levels fall. Lipemia retinalis is usually found during an eye examination and does not always affect vision.

Pancreatitis commonly causes steady, moderate to severe pain in the upper abdomen. The pain may spread to the back and may occur with repeated vomiting, fever, or abdominal tenderness. New or worsening abdominal pain needs prompt medical assessment because symptoms alone cannot confirm the cause.

- Episodes of upper abdominal pain, sometimes with nausea or vomiting
- Repeated acute pancreatitis
- Small, yellow or reddish-yellow skin bumps called eruptive xanthomas , often on the buttocks, back, arms, or legs
- A pale or creamy appearance of blood vessels in the retina, called lipemia retinalis
- An enlarged liver or spleen
- Tiredness or difficulty concentrating, particularly when triglycerides are extremely high

Causes and risk factors

An enzyme called lipoprotein lipase , or LPL, normally breaks down triglycerides carried by chylomicrons. Several other proteins help LPL work or move it into the correct position along blood vessels.

FCS usually occurs when a person inherits harmful changes in both copies of a gene involved in this pathway. Genes associated with FCS include `LPL`, `APOC2`, `APOA5`, `GPIHBP1`, and `LMF1`. Because inheritance is often recessive, parents may carry one altered gene without having the syndrome. A family history of severe triglycerides may therefore be absent.

MCS is more common than FCS. A person may inherit several genetic differences that each have a small effect on triglycerides. Chylomicronemia can then develop when one or more additional factors overwhelm the body's ability to clear triglyceride-rich particles.

Important contributing factors include:

This list is not complete. A prescribed medicine should not be stopped without speaking with the clinician who manages it. In many cases, another treatment can be considered or the medicine can be adjusted safely.

- Diabetes that is not well controlled
- Insulin resistance, metabolic syndrome, or obesity
- Regular or heavy alcohol use
- An underactive thyroid, called hypothyroidism
- Kidney disease, including nephrotic syndrome
- Pregnancy, especially later in pregnancy
- Diets high in fat, added sugars, or refined carbohydrates
- Certain medicines, including oral estrogens, corticosteroids like prednisone or hydrocortisone , retinoids, some antipsychotics like olanzapine or risperidone , and some treatments for HIV

Diagnosis

Diagnosis begins with a lipid panel, usually after fasting. Because triglycerides can change substantially, clinicians may repeat the test. Blood samples with marked chylomicronemia can appear cloudy or milky.

A clinician will review previous triglyceride results, age at onset, pancreatitis history, diet, alcohol use, medical conditions, medicines, pregnancy status, and family history. The pattern

over time can help distinguish lifelong FCS from multifactorial disease.

Additional blood or urine tests may look for contributing conditions. These may include glucose or hemoglobin A1C for diabetes, thyroid testing, and tests of liver and kidney function. Other cholesterol measurements, including apolipoprotein B, may help a lipid specialist interpret which particles are present.

Genetic testing can support a diagnosis of FCS when the clinical pattern suggests it. However, testing may not find every disease-causing change, and a negative result does not rule out all inherited or multifactorial forms. Genetic counseling can help a person understand the meaning of a result and whether relatives should be tested.

A clinician may also use a validated clinical scoring system that considers factors such as repeated fasting triglyceride levels, age at onset, response to standard medicines, and the absence of common secondary causes. No single symptom or triglyceride result establishes the exact type of chylomicronemia.

Treatment and management

Treatment aims to lower triglycerides enough to reduce the risk of pancreatitis while maintaining adequate nutrition. The plan depends on whether chylomicronemia is familial or multifactorial, previous pancreatitis, other medical conditions, and available treatments.

A low-fat or very-low-fat eating pattern is central to managing FCS. Some people with FCS are advised to obtain no more than about 10% to 15% of daily calories from fat. This level of restriction should be planned with a dietitian or specialist because the body still needs essential fatty acids and fat-soluble vitamins.

Fat is spread across meals rather than eaten in one large amount. Alcohol is generally avoided because it can sharply raise triglycerides and increase pancreatitis risk. Added sugars, sugar-sweetened drinks, and highly refined carbohydrates may also worsen triglycerides.

Medium-chain triglyceride oil may provide calories without being packaged into chylomicrons in the same way as ordinary dietary fat. It is not suitable for everyone and should be used with professional guidance. Children, pregnant people, and anyone following a highly restricted diet need particular attention to growth and nutritional adequacy.

For MCS, management also focuses on correcting contributing factors. This may include improving diabetes control, treating hypothyroidism, reducing alcohol intake, supporting gradual weight loss when appropriate, or reviewing medicines that raise triglycerides.

Clinicians may use fibrates or prescription omega-3 fatty acids to lower triglycerides in appropriate patients. Statins like atorvastatin or simvastatin may be recommended when a

person also needs to reduce the risk of heart attack or stroke. These medicines often have limited triglyceride-lowering effects in FCS because they partly depend on a working LPL pathway.

In the United States, the Food and Drug Administration has approved olezarsen as an addition to diet for adults with FCS. Olezarsen reduces production of apolipoprotein C-III, a protein involved in triglyceride metabolism. It is given as an injection. Possible effects include injection-site reactions, joint pain, and lower platelet counts. A specialist can explain monitoring, expected benefits, and whether the treatment is appropriate.

Acute pancreatitis is treated in a hospital. Care may include intravenous fluids, pain control, nausea treatment, and temporary changes in food intake. Insulin may help when severe triglyceride elevation is linked to uncontrolled diabetes. Procedures that rapidly remove triglyceride-rich plasma are reserved for selected situations because they are not routine treatment for every episode.

Complications

Acute pancreatitis is the most important complication. Some episodes are mild, while others can affect nearby tissues or other organs. Repeated attacks may lead to chronic pancreatitis, long-term abdominal pain, difficulty digesting food, or diabetes.

Severe dietary restriction can also create problems if it is not carefully planned. Possible concerns include inadequate calories, essential fatty acid deficiency, and low levels of vitamins A, D, E, or K. Dietitian support and appropriate laboratory monitoring can help prevent these issues.

Cardiovascular risk differs between types of chylomicronemia. People with MCS may have increased risk of heart and blood vessel disease because they often have cholesterol-rich remnant particles, diabetes, high blood pressure, or other risk factors. FCS does not always carry the same cardiovascular risk pattern despite very high triglycerides. A clinician should assess the person's full risk rather than relying on the triglyceride level alone.

Living with a restrictive diet and concern about pancreatitis can affect work, school, travel, eating with others, and emotional well-being. These effects are valid parts of care and can be discussed with the treatment team.

Follow up

Routine follow-up usually includes fasting triglyceride measurements, review of abdominal

symptoms, and assessment of nutrition and contributing conditions. The clinician may also monitor glucose, liver and kidney function, thyroid function, or vitamin levels when relevant.

People taking medicine need follow-up for effectiveness and side effects. Those with FCS may benefit from care involving a lipid specialist, registered dietitian, primary care clinician, and genetic counselor. During pregnancy or when planning pregnancy, early coordination with an obstetric and lipid team is important because triglycerides can rise quickly.

Contact a clinician promptly for a new pattern of abdominal discomfort, repeated nausea, reduced food intake, new skin bumps, or triglyceride results that are much higher than usual.

Seek urgent medical assessment for persistent upper abdominal pain, especially when it spreads to the back or occurs with vomiting.

Seek emergency care for severe or rapidly worsening abdominal pain, repeated vomiting with inability to keep down fluids, fainting, confusion, breathing difficulty, or signs of severe dehydration. These symptoms can have several causes and require in-person evaluation.

Sources

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