

FAMILIAL CHYLOMICRONEMIA SYNDROME

WHAT YOU NEED TO KNOW

Familial chylomicronemia syndrome (FCS) is a serious disease that prevents the body from breaking down fats.

Eating even a little fat can make someone with FCS ill, and the condition causes chronic symptoms and can lead to potentially fatal pancreatitis. FCS is a genetic disorder passed down from parents. Because it is rare, many healthcare providers have never heard of FCS or may not know how to diagnose it.

Lipoprotein lipase is a digestive enzyme that helps the body break down structures called chylomicrons. People who have FCS have a problem with lipoprotein lipase: it is either missing or broken. Chylomicrons carry triglycerides (a type of fat) to where they are needed in the body for energy. A buildup of these particles causes an increase in triglycerides levels.

Patients with FCS usually have extremely high levels of triglycerides. Normal triglyceride levels fall below 150 mg/dL.

SYMPTOMS OF FCS







- Severe pain in the abdomen, often including back pain
- Acute or chronic pancreatitis
- Xanthomas, fatty deposits in the skin

PATIENTS WITH FCS MAY ALSO EXPERIENCE

- Vomiting or diarrhea
- Blood that, when drawn, appears “milky”
- Extremely high levels of triglycerides
- Numbness in feet or legs
- Memory loss or “foggy-headedness”
- A sense of isolation
- Feeling sad or depressed

PHYSICAL SYMPTOMS:

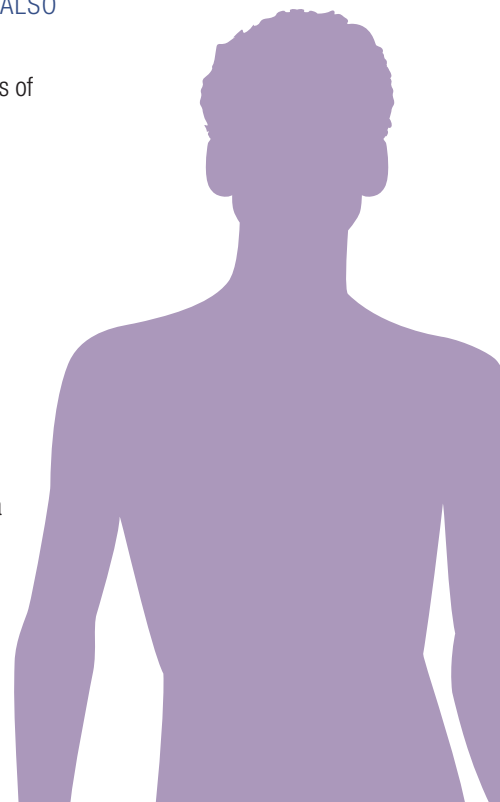
The 5 most commonly reported symptoms in a survey of FCS patients were:

-  **LIPEMIA RETINALIS** a condition in which the retinal veins of the eyes appear “milky”
-  **MEMORY LOSS** or ‘foggy-headedness’
-  **DEPRESSION** or a sense of isolation
-  **SEVERE PAIN** in the abdomen often including back pain
-  Acute or chronic **PANCREATITIS**
-  **XANTHOMAS**, fatty deposits in the skin

PATIENTS WITH FCS ALSO

EXPERIENCE:

- Extremely high levels of Triglycerides
- Blood when drawn, appears “milky”
- Fatigue (27%)
- Indigestion (23%)
- Bloating (35%)
- Generalized abdominal pain (33%)
- Vomiting or diarrhea
- Numbness in feet or legs
- Asthenia – Weakness or lack of energy (33%)



Visit [endocrine.org](https://www.endocrine.org) for more information.

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DIAGNOSIS

Diagnosis is critical to receiving the appropriate care. But because the age of onset and symptoms can vary, diagnosis may be difficult. There is no quick and simple test that tells you if you have FCS. Frequent attacks of pancreatitis, along with extremely high triglyceride levels, may suggest that FCS is the cause.

FCS can be clinically diagnosed. Physicians may look for:

- A history of abdominal pain or acute pancreatitis, which can be recurrent
- Severe, treatment-refractory hypertriglyceridemia
- An absence of secondary causes such as excess alcohol intake, uncontrolled diabetes, medical conditions, or medications known to cause hypertriglyceridemia

COMPLICATIONS

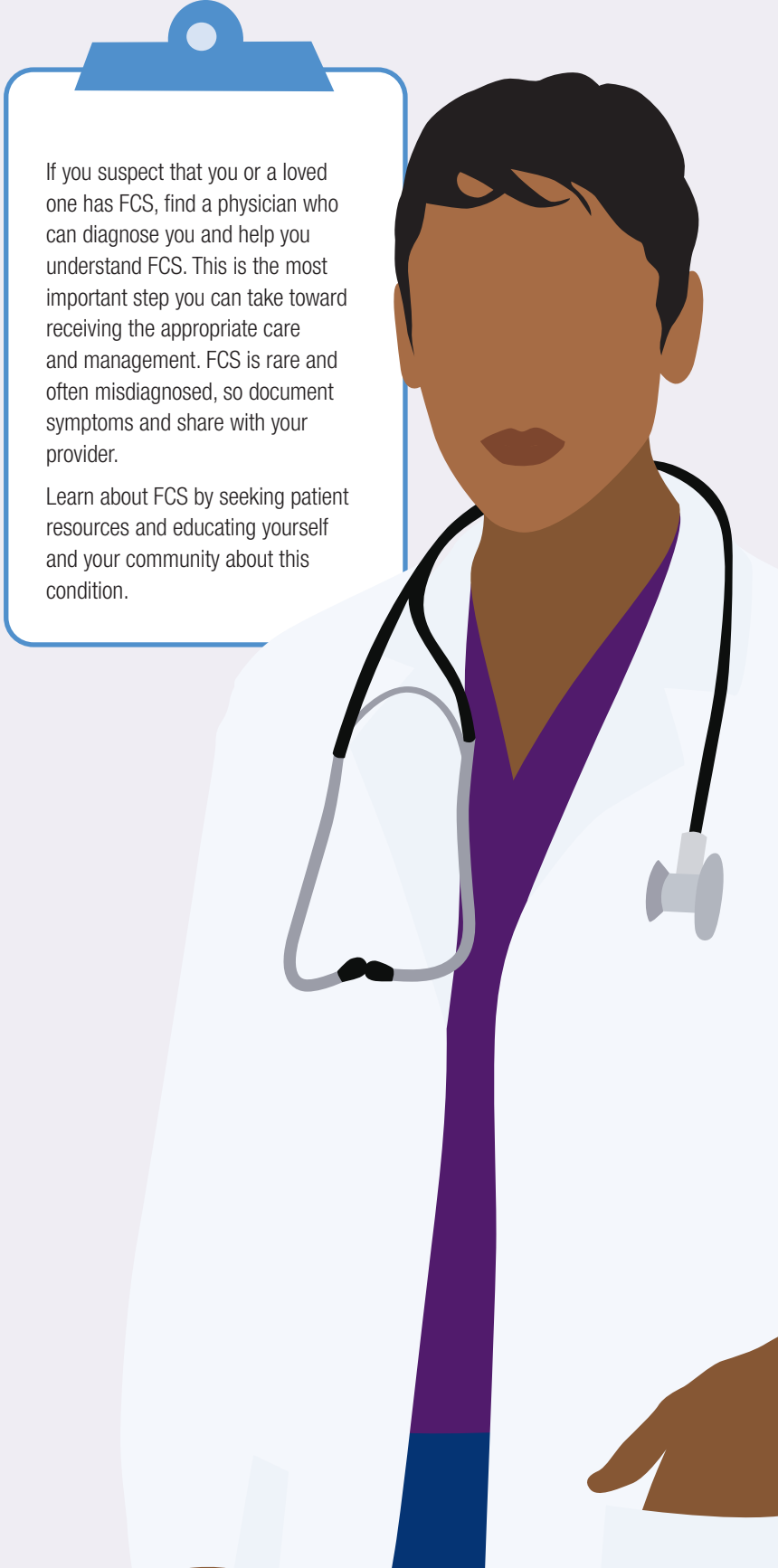
The most serious complication of FCS is pancreatitis, or inflammation of the pancreas, caused by extremely high triglyceride levels. The majority of patients with FCS experience pancreatitis. Of those, many will experience recurrent attacks. Pancreatitis can be extremely painful.

More importantly, it can lead to long-term organ damage, or insulin-dependent diabetes. Pancreatitis may even be fatal.

TREATMENT

Managing FCS begins with limiting daily fat intake to less than 10 to 15 grams of fat. Patients with FCS must avoid alcohol and limit simple carbs (such as those found in sodas and candy). Patients may also need to stop taking certain medications that can make symptoms worse. Do not stop any medication without first talking with a health care provider.

Visit FCSFocus.com for more information, support and other resources that have been created specifically for the FCS community in partnership with patients and caregivers.



If you suspect that you or a loved one has FCS, find a physician who can diagnose you and help you understand FCS. This is the most important step you can take toward receiving the appropriate care and management. FCS is rare and often misdiagnosed, so document symptoms and share with your provider.

Learn about FCS by seeking patient resources and educating yourself and your community about this condition.

Patients have questions. We have answers.

Endocrine Society is your trusted source for endocrine patient education.

Our free, online resources are available at endocrine.org/patient-engagement

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