# FAMILIAL CHYLOMICRONEMIA SYNDROME (FCS)

## What is FCS?

Familial chylomicronemia syndrome (FCS) is a rare, genetic disease that prevents the body from breaking down fats and can result in potentially life-threatening acute pancreatitis (painful inflammation of the pancreas).[1]

This often causes people with FCS to have extremely high triglyceride levels, greater than 880 mg/dL.[2]

**Triglycerides** are a type of fat that the body uses as an important source of energy.[4] However, FCS leads to the build-up of large triglyceride-containing particles called **chylomicrons** in the blood, which can result in severe health complications.[1]

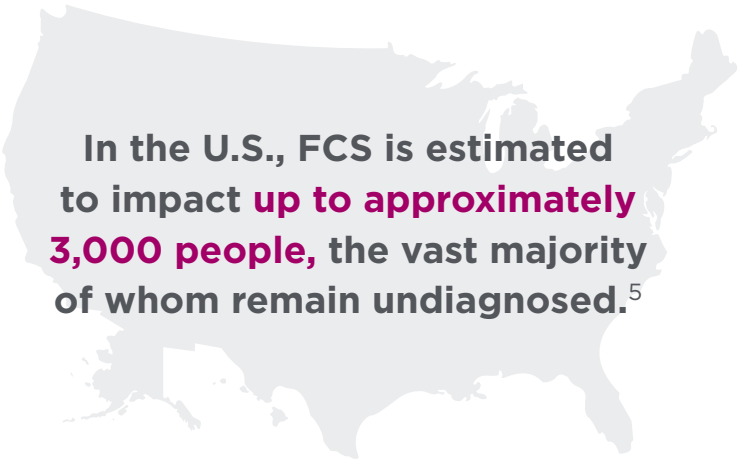
FCS is a form of severe hypertriglyceridemia (sHTG). sHTG is a condition in which a person's triglyceride levels can be dangerously high, 500 mg/dL or above.[3]

Although a healthy triglyceride level for adults is below 150 mg/dL, people living with FCS often have levels greater than or equal to 880 mg/dL.[2]

### Triglyceride Levels[2]

Healthy Adults <150 mg/dL

People Living with FCS ≥880 mg/dL



**In the U.S., FCS is estimated to impact up to approximately 3,000 people, the vast majority of whom remain undiagnosed.**[5]

## What causes FCS?

### FCS is caused by genetic changes (also known as disease-causing variants or mutations) in one of several genes.[2]

FCS is caused by mutations in several genes that impact the function of a protein called **lipoprotein lipase (LPL),** which is essential for removing triglycerides from the blood.[2]

About **80% of FCS cases** are due to genetic changes in the LPL gene itself.[6] Ongoing research continues to identify additional variants linked to FCS.

## What are the signs and symptoms?

In people with FCS, lack of LPL function means their body cannot break down fats, leading to the build up of chylomicrons.[1]

**Signs and symptoms include:[2,7]**

Severe, frequent stomach and back pain, which could lead to acute pancreatitis (AP)

Blood that appears fatty or milky white after it is drawn and stored

An enlarged liver or spleen, known as hepatosplenomegaly

Bloating and indigestion

Yellowish, waxy-like deposits of fatty material in the skin, called xanthomas

Depression, anxiety, fatigue, memory loss or brain fog

Fatty deposits in the retina of the eye, known as lipemia retinalis

## What is the disease burden?

FCS puts people at **high risk of AP**—painful inflammation of the pancreas—which often results in hospitalization and can be life-threatening.[7]

In severe cases, vital organs such as the heart, lungs, kidneys and pancreas can be damaged.[8] In addition to the physical burden, FCS can be a major source of psychological and financial stress, which can cause people to feel overwhelmed and experience a **significant impact on their quality of life.**[7]

For example:[7]

- FCS may **stop people from participating** in social activities or maintaining steady work.
- FCS can **cause anxiety** about relationships, **guilt** and **disappointment** about missing out on events or activities.
- It can also **create frustration and exhaustion** from managing the condition.

People with FCS can experience challenges in social settings centered around food and drink, which can add to emotional distress, social isolation or feelings of uncertainty or hopelessness.[7]

**Consuming even a small amount of fat can cause someone with FCS to experience physical symptoms.[2]**

## How is FCS diagnosed?

Because FCS is so rare, it can be **hard to recognize and identify.** This means many people with the condition are left without a complete clinical diagnosis or the appropriate help for long-term care.[2]

In many cases, FCS is **typically not diagnosed until an individual has been hospitalized** for AP, at which point their disease is considered more severe.

A diagnosis of FCS can be **suspected based on physical and laboratory signs,** including triglyceride levels greater than or equal to 880 mg/dL, that are unresponsive to standard triglyceride-lowering therapies without other causes.[2]

A proper diagnosis helps people with FCS get the care they need. A doctor can diagnose FCS by **clinical confirmation and/or genetic confirmation** using a validated clinical scoring tool.[2,9]

## How can people manage their FCS?

Due to the absence of LPL activity in people with FCS, medicines traditionally used to lower triglyceride levels are clinically ineffective because they rarely reduce triglycerides or decrease the risk of AP.[7]

People with FCS have historically relied on diet alone, **limiting their daily fat intake to less than 15 to 20 grams (or less than 10% to 15% of their daily calories).**[7] For context, this is equivalent to approximately one tablespoon of olive oil.[7]


**It is important to lower triglyceride levels,** which can help lower the risk of disease complications.

For more information about FCS, please visit **www.KnowYourTGs.com** or the FCS Foundation at **www.LivingWithFCS.org**.

### References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