



## Familial Chylomicronemia Syndrome (FCS)

Familial chylomicronemia syndrome (FCS) is a rare genetic lipid disorder characterized by the build-up of chylomicrons (chylomicronemia), a large lipoprotein particle that transports dietary fat and cholesterol in the body.<sup>1</sup> Because chylomicrons are rich in triglycerides, patients with FCS have extremely high levels of triglycerides, reaching 10 to 20 times the normal level. Because people with FCS cannot break down chylomicrons, their blood can appear milky in color (lipemic), and they live at risk of severe recurrent abdominal pain and potentially fatal pancreatitis, long-term complications from pancreatic damage, and symptoms that can interfere with daily living.<sup>2,3,4</sup>

### What Causes FCS?

Lipoprotein lipase (LPL) is an enzyme that works to help break down chylomicrons in the body. In people living with FCS, LPL does not function correctly. This causes a buildup of chylomicrons (particles that carry triglycerides) in the bloodstream.<sup>5,6,7</sup>

### Burden of FCS

People with FCS face significant morbidity and mortality risks including the threat of acute pancreatitis, which can be fatal, and pancreatic damage, which can cause long-term morbidities that can be disabling and affect many aspects of daily life.<sup>6</sup> In women, factors that increase estrogen levels, including use of oral contraceptives and pregnancy, can spike triglycerides, increasing the risk of pancreatitis, miscarriage and maternal fatality.<sup>8,9</sup>

### A survey of FCS patients showed that:

- 59% reported a history of recurrent abdominal pain without hospitalization; and
- 50% had a history of recurrent pancreatitis (ranging from 2 to 96 episodes).<sup>10</sup>

### In a separate study, of patients experiencing triglyceride-induced pancreatitis,

- 60% required care at an intensive care unit;
- 50% had pancreatic necrosis as identified via contrast CT-scan findings;
- 48% developed persistent organ failure; and
- there was an 8% mortality rate.<sup>11</sup>

Among all symptoms of FCS, abdominal pain and pancreatitis present the most significant risks to patients. Pancreatitis typically begins with extreme nausea, vomiting, sweats and pain in the upper abdomen that radiates to the back and ribcage.<sup>12</sup> In severe cases, patients describe the pain associated with pancreatitis as excruciating and intolerable – “like a knife repeatedly stabbing me on the side” or “like someone took a power drill to the side of your rib cage.”<sup>7,13,14,15</sup>

Many people living with FCS report experiencing episodes of fatigue, lack of energy (asthenia), impaired cognition and numbness/tingling.<sup>13,16,17</sup> Patients report that fatigue and pain caused by FCS can affect their ability to work, while dietary restrictions and fear of attacks of pancreatitis can restrict opportunities to participate in many social activities or travel.<sup>5</sup>

## Diagnosis

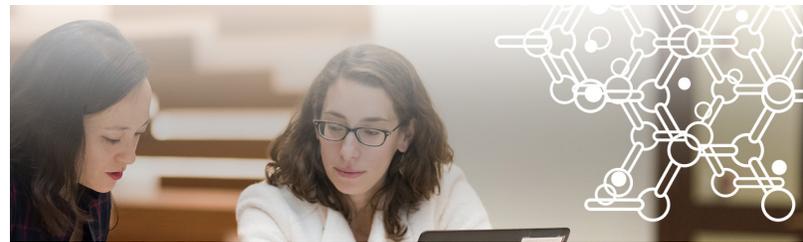
Receiving a specific diagnosis of FCS is crucial to receiving the right education, medical care, and support from others living with this disease. Because high triglycerides and pancreatitis have many causes, doctors may not recognize cases of FCS. Many patients report that they experienced symptoms including severe abdominal pain and pancreatitis for several months or even years before they received a FCS diagnosis.

Optimal management requires a care team of HCPs, and may include a lipid specialist for triglyceride management, gastroenterologist for support managing abdominal pain and gastrointestinal symptoms, a registered dietitian to help with dietary choices, and a primary care physician.<sup>8</sup> With a diagnosis, patients with FCS can meet and support each other by connecting through organizations such as the FCS Foundation.

## The Continuing Unmet Need in FCS<sup>18,19,20,21</sup>

There are currently no approved therapies for the treatment of FCS. Available therapies used to reduce triglyceride levels, including niacin and fibrates, work by targeting the LPL pathway. In FCS this pathway does not function properly, making these options ineffective.

Current standard of care in managing FCS often involves severe dietary restrictions to avoid increasing triglyceride levels. Many people living with FCS limit fat intake to only 10-12 grams per day (there are 14 grams of fat in a single tablespoon of olive oil) and still experience episodes of pancreatitis.



## Learn More

For more information about FCS diagnosis and management, visit [fcsfocus.com](https://www.fcsfocus.com).

The FCS Foundation is also on the web at [livingwithfcs.org](https://www.livingwithfcs.org), on Facebook at [facebook.com/fightFCS](https://www.facebook.com/fightFCS), and on Twitter at [@livingwithfcs](https://twitter.com/livingwithfcs).

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