

ABOUT FCS

Familial chylomicronaemia syndrome

Introduction

You (or someone close to you) have been diagnosed with familial chylomicronemia syndrome, or FCS for short. FCS is a rare genetic disorder characterised by elevated triglyceride levels, often resulting in recurrent pancreatitis and other complications.¹ Navigating life with FCS can be challenging, but this brochure provides important information about FCS and how to manage the disease.

What is Familial chylomicronaemia syndrome?

Familial chylomicronaemia syndrome (FCS) is a genetic condition that affects only a small number of people worldwide.

People with FCS are unable to fully break down certain fats, known as triglycerides, from their diet. Instead, the triglycerides accumulate in the blood, where they are transported via chylomicrons.¹ These chylomicrons can become lodged in small blood vessels causing inflammation and damage to the surrounding organs.²

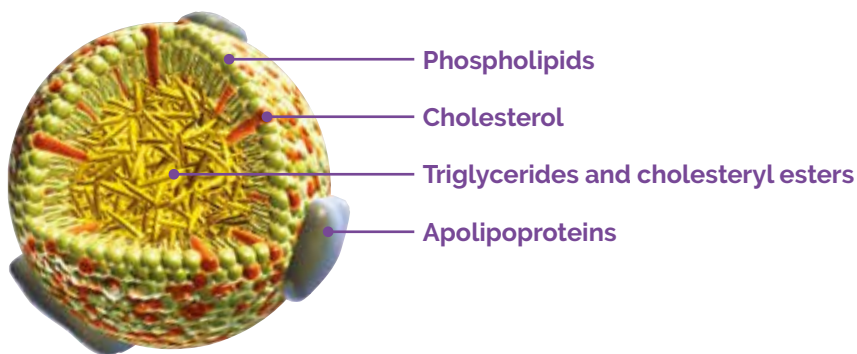
People with FCS are unable to digest fats

The food we eat daily contains fat in the form of triglycerides, which are absorbed from the gut and transported to the bloodstream. In the blood, compounds called chylomicrons carry the triglycerides to muscles, fatty tissue and other organs.²

In healthy individuals, chylomicrons are usually only present briefly in the bloodstream directly after a meal. They are then rapidly cleared by an enzyme* called lipoprotein lipase (LPL), which breaks down the triglycerides into fatty acids and glycerol.^{2,3} In people with FCS the LPL enzyme is missing or not working properly, causing extremely high triglyceride levels.¹

Chylomicrons are composed mostly of TGs

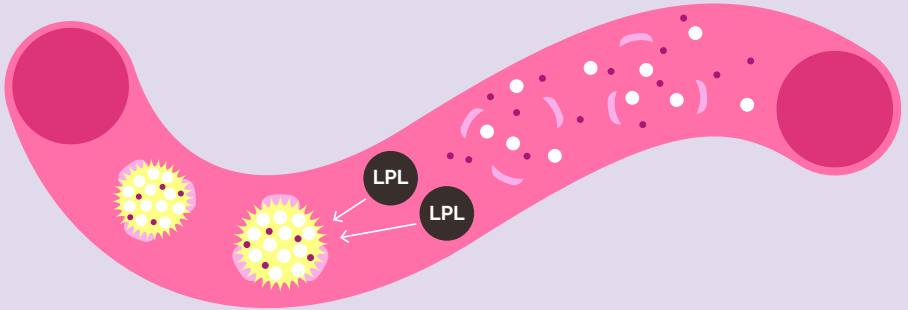
The structure of the chylomicron enables fats to travel through the bloodstream during digestion.^{2,3}



* Enzymes are compounds that are produced naturally by the body and regulate biochemical reactions to help speed up metabolism.

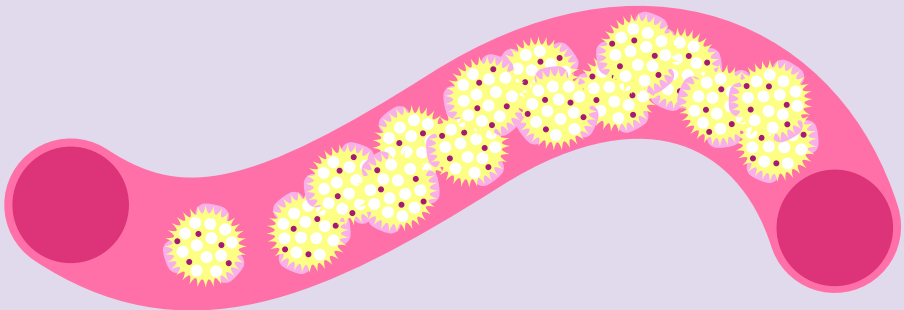
Normal triglyceride metabolism

Lipoprotein lipase (LPL) is an enzyme that helps digest fats. It breaks down triglycerides and bulky chylomicrons so our bodies can use the by-products for energy.^{3,4}



LPL dysfunction

People who have FCS are unable to process triglycerides because LPL is missing or broken.¹ As a result, fat accumulates in their blood in the form of chylomicrons. This accumulation is called chylomicronaemia.



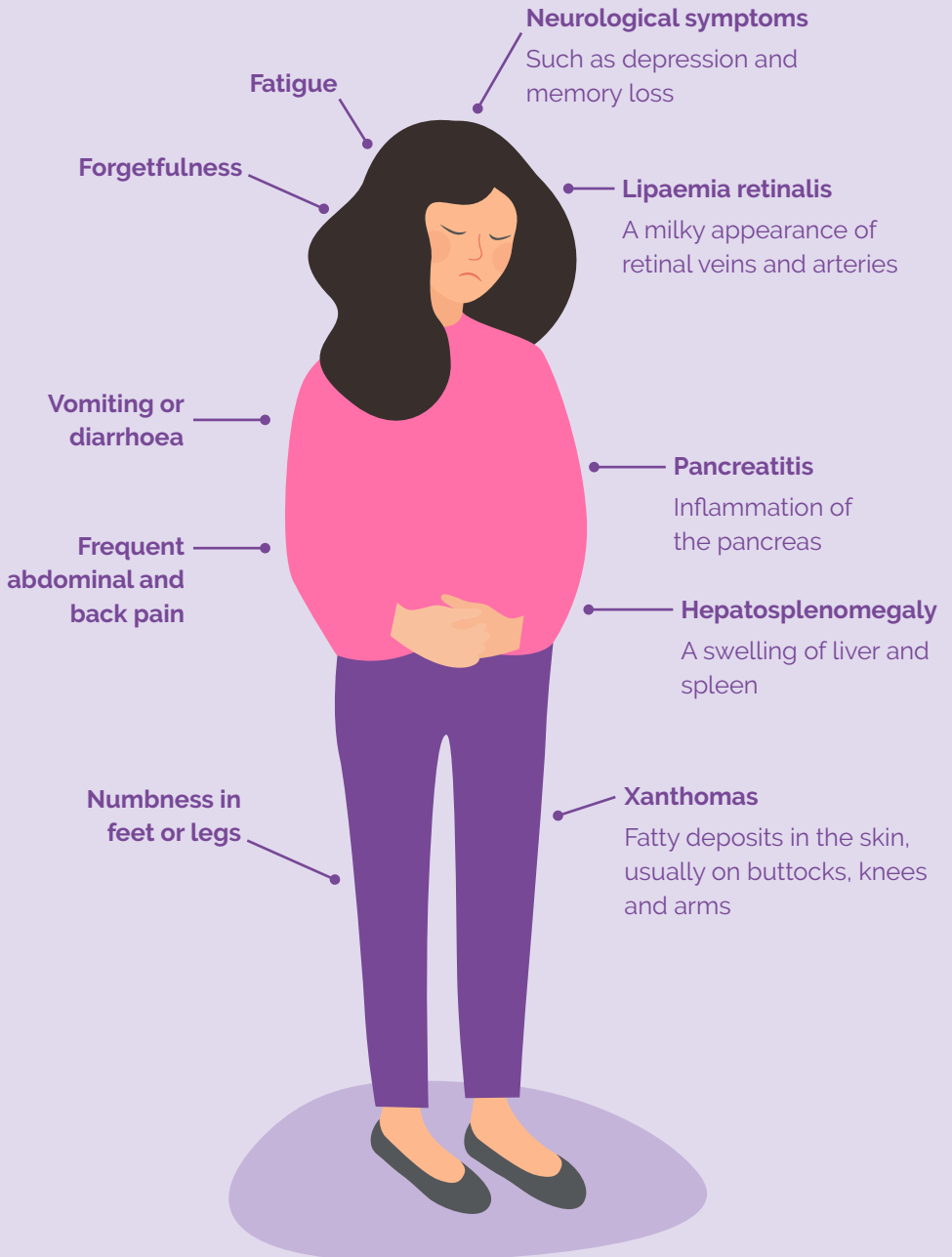
FCS signs and symptoms

The age at which patients first show symptoms, and the types of symptoms they experience may vary. Some people may notice warning signs of FCS when they are young. Others may not recognise the symptoms until adulthood.

The first physical symptom of FCS may be severe abdominal pain. Excess fat that builds up in various parts of the body may cause deposits of fat under the skin called eruptive xanthomas or "fat spots". Other common events are pancreatitis and cognitive symptoms such as brain fog.^{3,5}



A distinctive sign of FCS is the blood appearing fatty or 'milky' after a blood test.⁶ This is due to the high levels of fat in the bloodstream.



Adapted from: Davidson M, et al. J Clin Lipidol. 2018 Jul-Aug;12(4):898-907.e2.

What makes the doctor suspect FCS?

Signs and factors that can make the doctor think that a patient may have FCS include the following:⁶

- Very high triglyceride levels, above 885 mg/dl or 10 mmol/l
- Very high triglyceride levels despite medication and sticking to a low-fat diet
- A family member with high triglyceride levels or a congenital fat metabolism disorder
- One or more attacks of pancreatitis that required emergency treatment and for which no cause was found
- A congenital lipid, triglyceride, or fat metabolism disorder.

What is the risk of FCS for family members?

FCS is a hereditary disease.⁶ Family members may also be affected by FCS. Any family member with high triglyceride levels should also consult a specialist (such as a lipidologist or endocrinologist).

How FCS is diagnosed

FCS is diagnosed by measuring the triglyceride level in a blood sample, usually after fasting.

The triglyceride level is a measure of the amount of chylomicrons in the blood. Healthy people have a triglyceride level of less than 150 mg/dl or 1.7 mmol/l in the blood.²

If the triglyceride level in the blood is too high, this does not necessarily mean that the patient has FCS. Other causes of high triglyceride levels can include diabetes, liver or kidney disease, excess alcohol consumption, or certain medications. All other causes of high triglyceride levels must be ruled out before FCS can be diagnosed.²

If repeated blood tests continue to show very high triglyceride levels (above 885 mg/dl or 10 mmol/l) and all other causes have been ruled out, the FCS diagnosis is confirmed by genetic testing.⁶

Pancreatitis is a serious complication of FCS

The most serious complication of FCS is pancreatitis, which may happen when triglycerides are very high.²

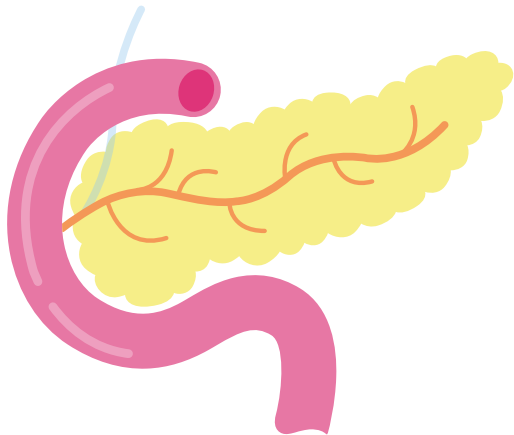
The pancreas is a small pear-shaped organ located in the abdomen behind the stomach. The pancreas is a part of the digestive system.

The pancreas is responsible for:⁷

- producing hormones (including insulin, which regulates blood sugar levels)
- producing digestive juices containing enzymes which break down food in the gut. Therefore, the pancreas is important for digestion.

Any of these signs may indicate a pancreatic event⁷

- Severe upper abdomen pain that may be felt in the back
- Tender abdomen or pain that is worse after eating
- Nausea and vomiting
- Fever



High triglyceride levels can lead to pancreatitis (inflammation of the pancreas). The higher the triglyceride levels, the higher the risk.

Pancreatitis is a condition where the pancreas becomes inflamed. It can be extremely painful and may worsen quickly. Each attack can become life-threatening. It may take several days or weeks for a pancreatitis attack to resolve. The treatment consists of painkillers and avoiding food until the symptoms subside. If symptoms persist, a doctor or hospital should be consulted.

Recurrent episodes of less severe pancreatitis – known as chronic pancreatitis – can, in the longer term, affect the function of the pancreas. This means that the pancreas may eventually stop producing the proteins (enzymes) that enables food digestion, which can cause digestive problems.

Repeated acute pancreatitis attacks can lead to chronic pancreatitis, which in turn can lead to:¹

- diabetes
- digestive problems with malnutrition
- recurring or persistent pain

To prevent pancreatitis attacks⁸, triglyceride levels in the blood must be kept as close to normal as possible. The best way to achieve this is to follow a strict diet and check triglyceride levels regularly, to ensure they remain stable. The diet must be followed carefully without errors or lapses.

In addition to adhering to a strict diet, daily medications can help minimise the risk of pancreatitis.

Living with FCS

A very restricted intake of fat and abstinence from alcohol are critical first steps of management.²

The FCS diet is limited to 15–20 grams of fat per day.² That is equivalent to approximately one tablespoon of olive oil or half an avocado. It also means restrictions on simple and refined carbohydrates and foods with added sugars, and no alcohol.

Different people may be able to tolerate different levels of fat intake. It usually takes time to fully understand what an FCS diagnosis means for you, your diet and how much fat you can tolerate.

Learning to manage FCS

Make a habit of reading the list of ingredients and nutritional values of foods in the supermarket. Keep a food diary to monitor your eating habits.

A qualified nutritionist or dietitian can help with advice and recommendations on FCS-friendly foods. When cooking at home or eating out, always choose meals recommended by the nutritionist or dietitian.

One option for some people with FCS may be to replace their usual sources of dietary fat with so-called medium-chain triglycerides (MCT fats).²

FCS-friendly nutrition guidelines

Food group	What to eat	What to avoid
Lean protein 85 g serving	<ul style="list-style-type: none"> ✓ Non-oily white fish, such as cod, skate, sole, canned tuna (not in oil) or haddock ✓ Most shellfish, including prawn, squid and lobster ✓ Breast of most poultry, skinned and trimmed of all fat ✓ Egg whites ✓ Fat-free dairy products, such as milk, yoghurt and cottage cheese <p>Steam or boil proteins, or cook with Medium Chain Triglyceride (MCT) oil.</p>	<ul style="list-style-type: none"> ✗ Fish high in fat such as salmon, mackerel, sardines ✗ Egg yolk ✗ Fatty meat, such as beef, lamb and pork ✗ Processed meat, such as hot dogs and sausages ✗ Other saturated and unsaturated fats
Complex carbohydrates	<ul style="list-style-type: none"> ✓ Whole grain, such as wholemeal and brown rice ✓ Wholewheat pasta and bread 	<ul style="list-style-type: none"> ✗ Egg pasta and pasta dishes prepared with refined flour, fat and meat ✗ Most cereal, e.g. rice, wheat, millet, maize and corn
Simple carbohydrates Refined starches and added sugars should be restricted	<ul style="list-style-type: none"> ✓ Choose in moderation strawberries, blueberries, blackberries, raspberries, oranges and kiwi fruit 	<ul style="list-style-type: none"> ✗ Fruit juices, fruits, sweets and soft drinks ✗ Sugar, syrup and honey
Vegetables, nuts and seeds	<ul style="list-style-type: none"> ✓ Leafy greens, such as spinach and kale ✓ Colorful vegetables, such as red peppers, courgette and squash 	<ul style="list-style-type: none"> ✗ Edamame, soy nuts, seeds, avocado, olives, coconut, peanuts and tree nuts
Fat 10-20 g per day	<ul style="list-style-type: none"> ✓ Cook with MCT oil ✓ Meet requirements for essential fatty acids 	<ul style="list-style-type: none"> ✗ All oils and fats, including olive oil, sunflower oil, canola, soybean oil, butter, lard, margarine, seed and nut butters ✗ Saturated and unsaturated fats

FCS glossary

Apolipoproteins

Apolipoproteins are proteins that combine with fats in the bloodstream to help transport cholesterol and other lipids around the body. Apolipoproteins play a crucial role in the body's breakdown and regulation of fat.

Cholesterol

Cholesterol is a fat-like substance found in the body and in certain foods. Cholesterol is vital for building cells and producing hormones. However, excessive cholesterol can accumulate in the blood vessels and cause health problems such as heart disease.

Chylomicrons

Chylomicrons serve as transport for dietary fats, such as triglycerides. They are formed in the small intestine and pass through the lymphatic system and the bloodstream to muscles, fatty tissues and organs, where they are broken down and further processed.

Familial chylomicronaemia syndrome (FCS)

Familial chylomicronaemia syndrome (FCS) is a rare genetic disorder where the body is unable to effectively break down chylomicrons, leading to extremely high levels of triglycerides in the blood. FCS can result in recurrent episodes of severe abdominal pain and may increase the risk of pancreatitis.

Lipoprotein lipase (LPL)

Lipoprotein lipase (LPL) is an enzyme that breaks down dietary fats from chylomicrons to allow the cells in the body to use the released energy.

MCT (medium-chain triglycerides)

Medium-chain triglycerides (MCTs) are a type of dietary fat containing saturated fatty acids. MCTs are mainly found in tropical vegetable fats, such as coconut oil. Unlike conventional dietary fats, MCT fats can be metabolised by the body without forming chylomicrons.

Pancreatitis

Pancreatitis is an inflammation of the pancreas, an organ in the abdomen that produces digestive enzymes and hormones such as insulin. Symptoms of acute pancreatitis include severe upper abdominal pain, nausea, vomiting and fever, which can be life-threatening. Pancreatitis is treated by administering fluids and painkillers and avoiding food.

Triglycerides

Triglycerides are naturally occurring dietary fats that the body can use as energy and store as fatty tissue. High levels of triglycerides in the blood can be a health risk.

Xanthomas

Xanthomas are fatty deposits that build up under the skin, appearing as yellowish bumps or nodules. They can develop when there are abnormal levels of fats in the blood and are often associated with conditions such as hypertriglyceridaemia and FCS.

“A hero is an ordinary individual who finds the strength to persevere and endure in spite of overwhelming obstacles.”

Christopher Reeve [Superman]

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