FAMILIAL CHYLOMICRONEMIA SYNDROME

SPOTLIGHT ON FCS

Leveraging the Power of Individual Journeys Toward Collective Action

Provided by

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FORWARD AND 2026 UPDATE

Since the *Spotlight on FCS* paper was first published in recognition of Rare Disease Day 2025, patients, caregivers, and FCS advocates have leveraged its content to elevate awareness and understanding about what it is like to live with FCS. We continue to learn about ways in which the FCS community struggles with lack of awareness and understanding of FCS, even among healthcare providers. This paper helps to de-mystify this rare condition and support more timely diagnosis and care.

This paper has been disseminated widely by the FCS community. In October 2025, we had the opportunity to present content from the paper at the NORD Rare Diseases and Orphan Products Summit (see appendix for poster), which brought together more than 800 stakeholders from across the rare disease ecosystem. In June 2026, we had the opportunity to present content from the paper at the EURORDIS European Conference for Rare Disorders (ECRD), which brought together more than 500 stakeholders from across the rare disease ecosystem. We are proud of the overwhelmingly positive response to this paper and are delighted to re-issue it with several updates to reflect progress in the field. We are also working to translate this paper into additional languages for broader dissemination and wider impact. Among the updates now reflected in the paper are ways in which the treatment landscape continues to evolve. As we acknowledge this milestone, we continue our partnership with the FCS advocacy community in building and strengthening connections to raise patient and caregiver voices.

Collectively, we are all focused on improving the lives of people living with FCS and are committed to bringing forward better outcomes for people managing FCS. It is our sincere wish that this paper will continue to be used to empower individuals living with FCS to find answers, receive the care they need, and know that they are not alone.

Testimonials from the FCS and Rare Disease Community

> *As an FCS patient, the 'Spotlight on FCS' paper has become one of the most valuable tools in my FCS Toolbox. When you live with a rare disease, being able to advocate for yourself is absolutely vital. Self-advocacy is paramount when it comes to navigating doctor's appointments, hospitalizations, unexpected trips to the emergency room and even when navigating daily life with people who don't know what FCS is and what it means to live with this disease. Education goes hand in hand with Advocacy. I can't effectively advocate for myself if I can't educate people about FCS. Being able to hand that paper to my friends, family members and healthcare providers has given me a voice when I'm in pain, seeking help, or even just planning a get-together with extended family or friends. It covers every aspect of the patient journey and validates our experience to people who have little to no understanding of FCS. That, to me, is priceless.*
>
> — Julie, Living with FCS

Testimonials from the FCS and Rare Disease Community *(continued)*

The 'Spotlight on FCS' white paper has become a huge resource for the FCS community. Having this paper available for knowledge and education about FCS is invaluable. Where there is knowledge, there is power. Power over the disease... to be able to live your best life and make informed decisions about your care. FCS impacts the whole family. A person's family shares in the fear and anxiety of the unknown. So, having the paper as a reference can bring hope to a family facing uncharted terrain. The family members, and primarily the spouse of the person with FCS, tend to 'pick up the slack' when their loved one is suffering with a 'low day.' Knowing that other caregivers are experiencing similar feelings of guilt, uncertainty, and isolation can be encouraging on our own 'low days.' Having the 'Spotlight on FCS' paper as a guide for managing this disease, and having a community at large, can help us all live our lives to the fullest... making memories with those we love.

— Lynne, Caregiver for Someone with FCS

The 'Spotlight on FCS' paper brings forward the real-life experiences of individuals living with this rare condition, highlighting their challenges from diagnosis through all aspects of daily life. It is informative in the accessible way it describes FCS and powerful in its approach to elevating the voices of FCS patients and their families. As a psychologist who specializes in supporting patients, family members, and rare disease communities, I appreciate the way this report tackles these important topics head on. As a dad who cared for a child with a rare disease, I know how valuable this kind of information and community building can be in bringing hope to families.

— Dr. Al Freedman, Psychologist, U.S.

FCS is a rare but devastating disorder making it challenging to manage and hard to understand. This paper provides the community with a clear and powerful picture of what it is like to live with FCS. For the patient, they endure very high blood triglyceride levels leading to recurrent episodes of pancreatitis, chronic abdominal pain, difficulty with concentrating, and frequent hospitalizations despite trying to follow a very restrictive low-fat diet. Up until recently, none of the traditional triglyceride-lowering therapies were effective for these patients. Since this is a rare disorder, the patients are often not accurately diagnosed causing frustration both by the patient and their physician. It is very exciting to have new medications that are very effective in lowering triglycerides and reducing pancreatitis episodes in patients with FCS.

— Dr. Alan Brown, Clinical Lipidology Specialist, U.S.

LETTER TO THE COMMUNITY

As a company focused on serving patients by developing investigational and commercial medicines to silence genes that cause a variety of diseases, Arrowhead Pharmaceuticals has prioritized development and commercialization of potential treatments for individuals with very high triglycerides, including those with the rare condition known as Familial Chylomicronemia Syndrome (FCS). Given the rarity of this disease, there are significant gaps in understanding the true impact of FCS on the community.

To fill some of these gaps, our team was privileged to host several Lived Experience Convenings in Fall 2024 with individuals from the U.S. and Europe, delving into the challenges facing those affected by FCS, learning how they want to be supported, and understanding what brings them hope. We are appreciative of the patients and their caregivers for opening up to us about their lived experiences, challenges, and hopes for the future.

A resounding learning from our conversations is the critical need and strong desire to increase awareness and understanding of FCS, especially among healthcare providers. This is vital to support timely diagnosis and improved management of care. People living with FCS are eager to share personal experiences to help others with this condition and build deep connections across the FCS community.

While our Arrowhead colleagues sought to gain insights through these Lived Experience sessions to inform our own programs and our approach to supporting people impacted by FCS, we were gratified that through our meeting, these 14 participants were able to express their "why" for engaging in these efforts, including hopes to:

- ⇒ *"Educate and inform as many people as possible about FCS."*
- ⇒ *"Show that you can live a full life and be 75 with FCS."*
- ⇒ *"Provide further awareness or help 'brothers and sisters' with FCS."*
- ⇒ *"Raise awareness to allow for earlier diagnosis."*
- ⇒ *"Reach out to people to educate on lifestyles, what FCS is, and how to deal and cope with it, learning how to not make it a burden in life."*
- ⇒ *"Do anything to help educate people and be an advocate for those who don't have one."*

With this Lived Experience white paper, we aim to honour the hopes and goals of these individuals and families, as well as the entire FCS community. Together we will work to raise awareness for FCS, foster a strong sense of community, convey to healthcare providers and others what life looks like for those living with this ultra-rare condition, and bring a "face" to a condition that is often overlooked or ignored.

We are deeply grateful to those who shared their perspectives so that others might have an improved experience as we collectively contribute to a brighter future for those living with FCS.

Alexandra Weiss Rosser
Director, Patient Advocacy
Arrowhead Pharmaceuticals

BACKGROUND

About Familial Chylomicronemia Syndrome[1-5]

FCS is an underdiagnosed, ultra-rare, serious condition that affects the way your body processes fat from food. It's so rare that it only affects about 1 to 10 people per million worldwide. People with FCS have a genetic issue that causes their blood to have extremely high levels of triglycerides. This happens because their body doesn't produce enough of an enzyme, called lipoprotein lipase, that's needed to break down triglycerides properly into chylomicrons. Chylomicrons are small particles involved in transporting dietary fats such as triglycerides, cholesterol and fat-soluble vitamins from the intestines to muscles and fat tissue for energy storage after a meal.

Because the chylomicrons aren't broken down, they buildup in the pancreas, leading to inflammatory damage. The pancreas releases pancreatic lipase to act on the accumulated chylomicrons, and, as they break down, they release free fatty acids that can be toxic and can cause further inflammation and damage to the pancreatic cells.

Since chylomicrons are the primary dietary triglyceride carrier, individuals affected by FCS have persistent extremely high triglycerides as well. Fasting triglycerides can occur at levels > 10 mmol/L (880 mg/dL), and in many cases, far higher. People with FCS can have triglyceride levels 10 to 100 times higher than normal.

These individuals have little to no response to conventional triglyceride lowering therapies and are unable to process consumed fats. Strict dietary restrictions and lifestyle changes are essential for FCS management, but they create significant stress. Even with careful management, the risk of pancreatitis remains high, affecting both mental and physical well-being. These symptoms diminish quality of life and can impair an individual's ability to work, thus expanding the burden of disease. Due to its rarity and a general lack of physician knowledge, diagnosis is usually a long, winding clinical process, even though there is an available genetic test to confirm the condition as well as criteria to provide a clinical diagnosis.

Key Symptoms of FCS

Acute pancreatitis
which can be life-threatening

Severe abdominal pain
Often caused by acute pancreatitis

Eruptive xanthomas
Small, yellowish skin bumps due to high triglycerides

Lipemia retinalis
A rare eye condition where blood vessels appear milky

Cognitive & emotional effects
Anxiety, depression, and brain fog are common due to chronic stress

Organ complications
Enlarged liver and spleen (hepatosplenomegaly) and increased diabetes risk

We lead a life of bomb diffusers. Any wrong move . . . that could be the end. That's exactly how I feel . . . Is it around the corner? Is it underneath my feet?

— *Person living with FCS*

BACKGROUND (continued)

Complications of FCS[2,6-8]

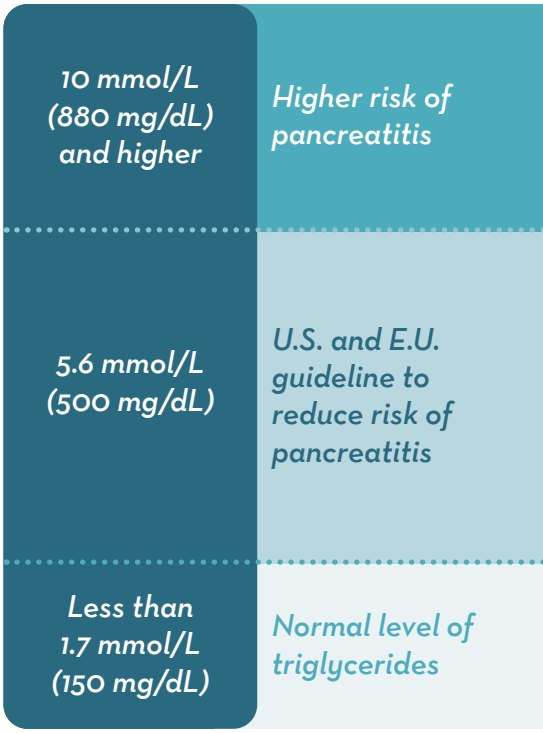
The most serious complication of FCS is pancreatitis, or inflammation of the pancreas, caused by the extremely high triglyceride levels. The majority of patients with FCS experience severe and extremely painful acute pancreatitis. Of those, many will experience recurrent attacks.

The high serum triglyceride levels not only lead to acute pancreatitis, but often to recurrent acute pancreatitis in greater than 50% of patients, which can lead to chronic pancreatitis.

Pancreatitis comes with its own complications of exocrine deficiency underproducing enzymes leading to digestive issues, steatorrhea or fat excretion in stools, limited nutrient absorption and endocrine deficiency such as diabetes and other complications. Pancreatitis may even be fatal.

Cases of cardiovascular disease (CVD) have also been reported in a number of individuals with FCS. As FCS progresses, it can lead to long-term organ damage or insulin-dependent diabetes.

Triglycerides Levels in People with FCS[12-19]



What is Acute Pancreatitis?[9-11]

Acute pancreatitis is a sudden inflammation of the pancreas, characterized by severe abdominal pain, nausea, vomiting, fever, and a rapid pulse. The pancreas, located behind the stomach, plays a crucial role in digestion and blood sugar regulation.

In patients with FCS, pancreatitis is due to elevated triglyceride levels, called hypertriglyceride-induced acute pancreatitis. Other common causes of acute pancreatitis include: gallstones (which can block the bile duct, causing pancreatic enzymes to back up into the pancreas, leading to inflammation), alcohol use (which can damage the pancreas over time), certain medications, infections, or trauma.

Hypertriglyceridemia-Induced Pancreatitis[12-19]

Hypertriglyceridemia-induced pancreatitis may occur when triglyceride levels exceed 5.6 mmol/L (500 mg/dL), with a higher risk when levels are above 10 mmol/L (880 mg/dL). This event is more common in individuals with obesity, diabetes, or a history of high triglyceride levels, and can be particularly severe, requiring prompt medical attention to manage triglyceride levels and prevent further episodes. The risk of severe acute pancreatitis increases with increasing triglyceride levels. Acute pancreatitis caused by high triglycerides tends to be more severe than other causes.

Overarching themes relating to the FCS lived experience

- ⇛ Diagnostic timelines must be shortened as too many people currently suffer through extended, frustrating diagnostic journeys related to FCS
- ⇛ Most FCS management requires meticulous attention to a restricted diet designed to prevent pancreatitis and this imposes significant physical, emotional, social, and financial impacts on families
- ⇛ There is a need for education and advocacy about FCS to address gaps in supporting patients and understanding FCS by health care providers
- ⇛ Families seek easy-to-find resources for accurate, objective FCS information
- ⇛ Self-advocacy is crucial
- ⇛ Connection and support within the FCS community are vital
- ⇛ People living with FCS seek to live a "normal" life without the constant pressures of FCS

Over time and through continued injury of the pancreas, acute necrotic fluid collections and pancreatic necrosis, or death of cells or tissues, can develop and lead to persistent organ failure, multiple organ failure, and persistent systemic inflammatory response syndrome.

Understanding the True Impact of FCS

As research and development efforts to produce effective therapies for FCS advance, it is increasingly important to raise awareness and promote understanding of the lived experience of FCS patients and their families to ensure that people are receiving the most timely and appropriate care. The patient and caregiver insights throughout this paper have all been collected from ongoing discussions with the FCS community.

Despite dealing with an ultra-rare, socially isolating condition, the FCS community has coalesced, especially in recent years with the launch of several regional and global patient advocacy organizations dedicated to FCS advocacy. The growth of this community provided us with a platform and unique opportunity to bring together individuals with FCS and their caregivers to generate insights and a first-person narrative of this difficult disease. The following white paper reflects those learnings and chronicles the FCS lived experience in 2024, across various demographics and geographies. Developed from insights gained through engagement with 14 individuals from the FCS community in the U.S. and Europe, this white paper is presented to the global FCS community by Arrowhead Pharmaceuticals.

Designed to amplify the voices of people impacted by FCS, and to drive awareness of FCS among clinicians and the broader public, this white paper is aimed at spurring action by all those invested in improving the lives of people with FCS. We hope to help pave the way for a better future for the FCS community, by empowering advocates to raise awareness of the challenges for people living with FCS, strengthen connectivity within the community, and advocate for systemic changes to improve care across geographies.

INSIGHTS COLLECTION METHODS

U.S. FCS Lived Experience Insights Group

In September and October 2024, a group of five FCS patients was convened virtually for two sessions. The first meeting was a listening session where each of the participants could share, in their own words, their experience living with FCS and what their diagnostic odyssey has been.

The second meeting was an interactive session where participants responded to prompts and exercises pertaining to their lived experience managing FCS, resource needs, gaps and areas of opportunity, along with hopes for the future. Prior to the meeting, participants were sent a pre-meeting activity to capture their insights related to their FCS experiences.

The Lived Experience Insights Group participants self-identified and volunteered after seeing the outreach opportunity through the FCS Foundation, the National Pancreas Foundation, and through the FCS closed Facebook group, and included 5 individuals – two men and three women who were either genetic or clinically diagnosed with FCS. At least three have participated in clinical trials relating to their condition. One person was diagnosed as an infant, and all others later in life.

Global FCS Patient & Caregiver Advisory Board

In November 2024, in conjunction with the FH Europe Foundation's annual conference in Vienna, Austria, a Patient and Caregiver Advisory Board meeting was held, bringing nine patients/caregivers together for a three-hour in-person session.

These participants were invited to complete a pre-meeting series of questions (using the same set of questions the U.S. group received). Participants self-identified and volunteered after hearing about the opportunity through the FH Europe Foundation Patient Ambassador Program.

The group included four adult patients and one caregiver from the U.K., a caregiver of a pediatric patient from Estonia, one adult patient from Austria, one adult patient from the U.S., and one adult patient from the Netherlands.



FCS ADVOCACY LANDSCAPE

In recent years the FCS patient advocacy community has expanded, with multiple organizations focusing on supporting this ultra-rare patient population and their loved ones. We highlight here several of these organizations and appreciate their ongoing collaboration as we collectively work to elevate awareness of FCS and improve outcomes for people impacted by this condition in the U.S.

THE FOUNDATION FOR TRIGLYCERIDE DISORDERS

Foundation for Triglyceride Disorders

www.tgdisorders.com

The Foundation for Triglyceride Disorders (formerly the FCS Foundation) is a U.S.-based, patient-led organization dedicated to education, support, and advocacy for individuals affected by triglyceride-related conditions. Founded in 2016 by two disease advocates directly impacted by Familial Chylomicronemia Syndrome (FCS), the organization has since expanded its focus to improving understanding, care, and quality of life for people with triglyceride disorders, including FCS, multifactorial chylomicronemia syndrome (MCS), severe hypertriglyceridemia (sHTG), and triglyceride-induced pancreatitis. In 2026, the organization rebranded from the FCS Foundation to its current name, reflecting this expanded mission of serving the wider triglyceride disorder community through education, awareness, and patient advocacy. The Foundation hosts an annual Patient & Caregiver Meeting, along with regular virtual connection events.

action FCS — Familial Chylomicronaemia Syndrome

Action FCS

www.actionfcs.org

Founded in 2015 by a patient, Action FCS is a U.K. charity to raise awareness and educate about FCS, running an annual FCS Awareness Day on the first Friday of November, and running education events for the community both alone and with other organizations. They have resources to help manage the condition and a supportive community for people affected by FCS. The organization advocates for excellent care and has represented patients in medicines regulatory processes, and works with their medical advisory group and other specialists to improve care.

CORD — Canadian Organization for Rare Disorders

Canadian Organization for Rare Disorders

www.raredisorders.ca

The Canadian Organization for Rare Disorders (CORD) is Canada's national network and charity advocating for individuals and families affected by rare disorders. It serves as a unified voice for the rare disease community, working to advance policy, research, and patient support so that Canada's healthcare system better meets their needs. Rather than providing clinical services or diagnoses, CORD focuses on advocacy, education, and research support — connecting patient groups, clinicians, researchers, and policymakers around shared priorities. CORD has particular familiarity with rare lipid disorders, including Familial Chylomicronemia Syndrome (FCS), reflecting its broader work supporting communities affected by ultra-rare metabolic and genetic conditions.

> **The FCS Facebook peer-to-peer group has been invaluable. It has others who have FCS, they provide education, support, share recipes and good ideas, you don't feel as 'weird' as they are in the same position and have those same isolating feelings.**
>
> *— Person living with FCS*

FCS ADVOCACY LANDSCAPE (continued)

FH Europe Foundation
www.fheurope.org

The Foundation's (FHEF) mission is to enhance awareness, understanding, and access to diagnosis and treatment for inherited lipid conditions—including FCS among others—across Europe and beyond. By doing so, the Foundation ensures that those affected receive optimal care and support, leading to longer, healthier lives. In addition to its European efforts, FHEF is progressively extending its reach to global communities through a series of international initiatives like the FCS WhatsApp Group and fostering global collaboration through the Patient Ambassador Program, international patient support groups, and advocacy efforts like the recent call for a World Health Assembly (WHA) Resolution on Rare Diseases. By acting as a convener for the international community, FHEF drives meaningful progress in addressing familial hyperlipidemia on a global scale.

The National Pancreas Foundation
www.pancreasfoundation.org

Based in the U.S., the mission of the National Pancreas Foundation (NPF) is to provide hope for those suffering from pancreatitis and pancreatic cancer through funding cutting edge research, advocating for new and better therapies, and providing support and education for patients, caregivers, and health care professionals. The NPF helps those living with FCS navigate the challenges of this often misunderstood disorder by providing patient and provider resources on FCS and pancreatitis, raising awareness, and building connection through events. By amplifying the voices of those affected by FCS, the NPF helps ensure the needs of this community are recognized and addressed, ultimately improving quality of life and outcomes for patients.

Mission:Cure
www.mission-cure.org

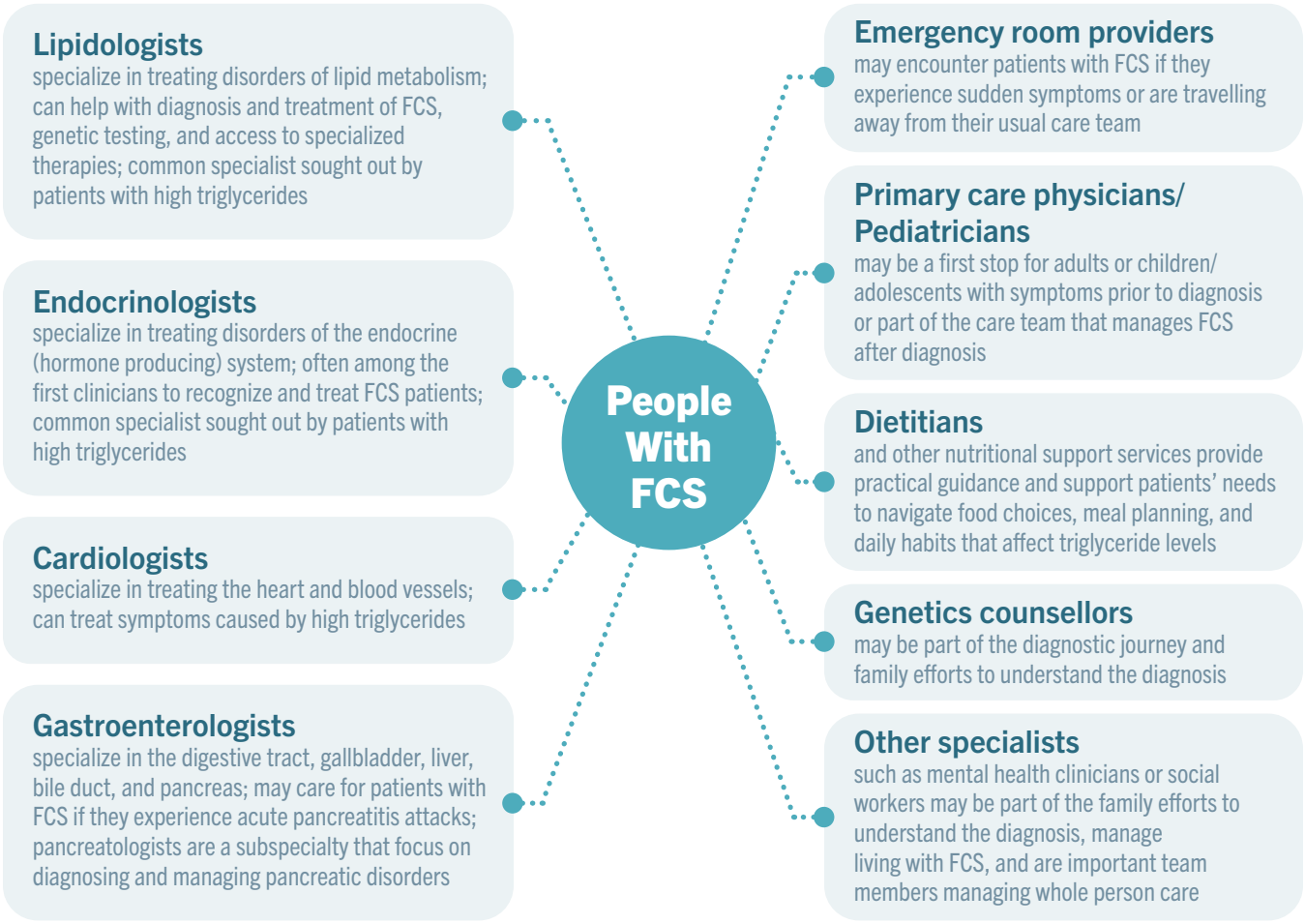
Mission:Cure is a pioneering non-profit based in the U.S. dedicated to transforming the lives of those affected by pancreatitis through a strategic, patient-centred approach aimed at finding effective treatments and ultimately a cure. Mission:Cure will continue to expand its support for the FCS, MCS, and severe hypertriglyceridemia communities by delivering educational resources through webinars, web pages, blog posts, and stories to help patients navigate their disease and management while fostering hope and empowerment. Through strategic collaborations and relentless advocacy, Mission:Cure drives meaningful change, ensuring pancreatitis patients receive the care, support, and hope they deserve.

THERE ARE ALSO TWO ONLINE, CLOSED AND CURATED FACEBOOK COMMUNITIES FOR THOSE WITH FCS

U.S.: FCS Facebook Support Group U.K.: FCS Community

CLINICAL CARE FOR FCS

Given the complexities of FCS and its manifestations, there is a wide range of providers and specialists that may play a role in diagnosing and caring for people impacted by this condition. Some of those specialists that come into contact with individuals managing FCS include:



This broad array of practitioners – combined with the rarity of FCS – creates a significant challenge for the FCS community in ensuring sufficient awareness of this disease and its symptoms among those providers who may encounter an FCS patient.

Current and Emerging Therapies

Expert clinical guidelines acknowledge that traditional triglyceride lowering therapies such as statins, fibrates, and omega-3 fatty acids do not typically work for this rare disease population.[16-19] Until recently, severe dietary restriction and alcohol abstention have been the most effective form of therapy, which is difficult to maintain. Research and development of new therapeutic approaches for FCS have been underway for a number of years, and the treatment landscape is rapidly evolving.

For up-to-date information on changes in the treatment landscape, consult your healthcare provider and consider utilizing online resources provided by patient organizations (see *FCS Advocacy Landscape* section for organizational information).

FCS DIAGNOSTIC ODYSSEY

> *When I started having pancreatitis attacks I would come home and lay in bed and would Google 'Reoccurring pancreatitis attacks. Why are my triglycerides so high? Why is my blood milky?'*
>
> — Person living with FCS

The journey to an FCS diagnosis can vary among patients. How the condition manifests itself also can vary depending on the individual. FCS can be diagnosed genetically or clinically. While there are patients who are diagnosed during infancy, people with FCS are diagnosed at all ages. Some are identified at birth due to inability to process triglycerides from their mother's breast milk or formula and failure to thrive, while others do not have symptoms until after puberty or far later and may not be diagnosed until they are well into adulthood. Many patients are left undiagnosed until later in life, only after symptoms continue to mount for many years ahead of a diagnosis. This helps to explain the differing journeys seen by those with FCS. Among the 14 individuals who provided insights to inform this white paper, some did not receive a diagnosis until they were in their mid-50s, while others were diagnosed much earlier.

Arriving at a diagnosis can take years and requires tireless persistence on the part of patients and their caregivers in the face of frequent misdiagnoses. For most of the participants who were not diagnosed during infancy, their journey to diagnosis included extensive independent research and presenting findings to their healthcare team for further discussion. There can be a long and difficult journey to understand why someone is experiencing acute and serious symptoms that often send them to a hospital emergency room, where typically the clinicians are unaware of FCS and focus on more common causes of these symptoms.

For adults with acute pancreatitis, healthcare providers generally start by asking about excess alcohol use or other lifestyle choices; some have even been accused of being alcoholics. These questions creates a sense among people impacted by FCS that their symptoms are "their fault." Delayed diagnosis or misdiagnosis also leads to unnecessary, painful, and costly procedures or treatments for other conditions and can create havoc in the lives of individuals and their families.

> *I kept hearing 'you must be doing something' from doctors, assuming too much alcohol.*
>
> — Person living with FCS

One participant in the U.S. described having 12 years of critical medical complications and hospitalizations (being told he had a 1 in 20 chance of surviving), multiple surgeries, diabetes, and living with the fear that he can't survive another pancreatitis attack. Another participant from the U.S. detailed enduring two decades of medical issues and major surgeries, including spending 7-10 days per month over a two-year period in the hospital because of pancreatitis attacks

FCS DIAGNOSTIC ODYSSEY (continued)

(while having an infant at home), undergoing weight loss surgery, subsequent revision, radical hysterectomy, and plasmapheresis three times per week. She recalled that she and her husband once tracked one specific year's annual medical expenses at $2 million USD. Unfortunately, stories like this were common among those who shared their experiences. One of the male contributors shared that he spent time regularly in the hospital (with three young children and a wife at home) due to pancreatitis attacks over a decade ahead of his eventual diagnosis.

> *I spent a lot of years wondering what was wrong with me. When I finally got a diagnosis, I felt a relief just to know what it was even though it wasn't a very hopeful diagnosis. Being able to put a label on it helped me know how to advocate for myself.*
>
> — Person living with FCS

IN THEIR OWN WORDS:

Participants' responses to when they were the most affected:

"During life threatening and chronic pancreatitis, when no one could explain why this was continuing to occur..., the mental fatigue of missing a week of work every month or so..., the social and emotional isolation of being hospitalized so frequently."

"For 12 years I was in and out of hospital every 2-3 months (7-10 days every time). I see 17 specialists on a regular basis at this point because of all the problems the acute pancreatitis caused."

"[I had] to put education and career goals on hold and structure life around treatments and surgeries."

"When my youngest child was in hospital for 5 weeks, not knowing if he would live or die, waiting for a diagnosis."

"I would have liked an explanation of what chylomicronemia meant so I understood what was happening inside my body. It took more than a year for anyone to explain that to me."

SUPPORTING NEWLY DIAGNOSED INDIVIDUALS AND THEIR FAMILIES

> *It was such a relief to know the devil I was dancing with.*
>
> — Person living with FCS

Although receiving a diagnosis of FCS was scary, upsetting, and challenging, individuals and families say they often felt a sense of relief when the relevant diagnosis was finally made.

Among those sharing their experiences, there was a strong sense of validation in having a diagnosis and learning that there are others who have similar experiences. Having a name for their condition and a reason for their symptoms offers the opportunity for them to regain some control over their experiences through diet, exercise, and medication.

Additionally, the experience of going through a protracted diagnostic odyssey motivated these individuals impacted by FCS to improve the process for others who follow in their footsteps. A key focus is the importance of nurturing the global FCS community so people struggling with FCS symptoms feel less alone and find support from others with similar experiences. All participants expressed how valuable the existing patient communities are in navigating a diagnosis of FCS, learning how best to manage a new way of living.

For earlier diagnosis, the FCS community has been advocating for access to genetic testing and for the inclusion of genetic testing for FCS within newborn screening programs, where possible. In the U.K., the Newborn Genomes Programme includes the 5 most common genetic mutations that cause FCS. There is also a call for raising awareness for more standardized triglyceride testing, especially when patients present with acute pancreatitis.

There is hope that policy changes will provide opportunities for families to begin effectively managing FCS from a young age. However, as newborn screening requirements are new and only relevant in certain regions and few include FCS or related screening, there is still limited data on the impact newborn screening will have on the overall FCS experience.

> *[This community needs] ways to educate others and share to avoid future generations suffering as I have with finding the right diagnosis and right treatment with my care team.*
>
> — Person living with FCS

Additionally, there is a strong desire and commitment among people living with FCS to enhance provider awareness of the condition and provide more timely support and relevant resources to those who are newly diagnosed. In many instances, newly diagnosed patients were left responsible for self-education and independent research.

SUPPORTING NEWLY DIAGNOSED (continued) INDIVIDUALS AND THEIR FAMILIES

ADVICE FROM PATIENTS FOR THE NEWLY DIAGNOSED

- Learn as much as you can about FCS, join the FCS patient groups to share and learn from each other's experiences. Find another patient who is willing to help guide you through the process, almost like a mentor or sponsor.
- Ask for help. Find your support system. A support system includes family or friends who can help you with managing your care plan, getting to appointments, getting to the hospital if you need it. You are not alone.
- Become proactive about your own health. Newly diagnosed patients should form a medical team that will collaborate on their care. Ask many questions about possible treatments and if you are eligible.
- Educate yourself about dietary restrictions and read food labels. Ask others in the community for recipe ideas and practical tips on switching from a traditional diet to an extremely low-fat diet. It starts with good new nutrition and substitutions!
- There is a ton of information out there and it isn't all true, so do the research; your case may be specific to other conditions you have.
- Have your doctor write you a letter explaining your diagnosis and a protocol for emergency care to take with you to the hospital when you have pancreatitis attacks.
- Give yourself some grace. You are going to have days where you struggle and it is ok to take a break. Grieve the diagnosis and get back on the horse to ride again.
- This disease does not define you. You can still have a life. You can still have dreams and goals. Make plans. You will learn to be flexible and make adjustments, but don't stop dreaming.
- Be open with people, you aren't a burden, once they know why you eat before you visit or bring your own food, people love to try cooking in support of you!
- Seek emotional support around peer pressure to conform; it is ok to be different. Do not see the diagnosis of "FCS" as a threat but as a hope. With this knowledge, the patient can do something about it together with the doctor. Without knowledge, you are groping in the dark.

It's a big deal

Get support from others who understand.

LIVING WITH FCS

Navigating life with FCS presents a tremendous burden on the wellbeing of the patient and their families. In addition to significant physical symptoms and health impacts, having FCS also presents a long list of modifications to living a normal and full life. In many instances, managing their lives with FCS means taking them away from being present with their families or missing important milestones or occasions. Constant worry relating to food and social activity also creates mental health burdens for people living with FCS and their families.

> *[Moments that have been affected most were] having to put education and career goals on hold. Having to structure my life around treatments and surgeries.*
>
> — Person living with FCS

The physical symptoms associated with FCS frequently include abdominal pain associated with extremely high triglycerides. During a pancreatitis attack, the pain can be excruciating. Treatment for pancreatitis can require lengthy, costly hospital stays and time away from family obligations and work. Some patients have high cholesterol in addition to high triglycerides. Likewise, individuals living with FCS are concerned about other physical impacts from their FCS, including onset of diabetes, liver disease, hormone fluctuations, fear of impacts on fertility and reproduction, and potential need for surgeries.

People with FCS tend to feel isolated, anxious, and uncertain of their future managing their condition and living their everyday lives. Those who had symptoms during adolescence and in their 20s, expressed what a challenge any form of social interaction was for them. Travel is difficult, as is caring for family members, and being present at work. Many believe stress levels and hormonal factors, specifically among women, are directly correlated with high triglyceride levels. These concerns and worries are shared by family members and caregivers, who also suffer the mental and physical health consequences of a life impacted by FCS. Guilt is a normal, near constant feeling, as individuals with FCS and their caregivers worry about the impact that managing FCS has on one another.

> *[I need] connection with other patients as they travel the same journey with FCS, there is so much power and support in the stories of other's journeys.*
>
> — Person living with FCS

LIVING WITH FCS (continued)

Because FCS families worry about their diet all the time, it is difficult for them to engage in normal, regular social interaction, which is often built around food. People impacted by FCS often limit their involvement in social gatherings or travel with friends and family. Not only is it challenging to evaluate the food served in these settings, but it is also upsetting to have to explain themselves repeatedly to others who may not understand the severity of FCS.

Some patients diagnosed with FCS say they have experienced stigma and felt misunderstood by the healthcare system. They worry that spikes in their triglycerides or pancreatitis attacks may be blamed on something they did or did not do, like failing to follow recommendations for diet, exercise, and medicine adherence. People impacted by this condition tend to seek ways to advance knowledge and validate effective strategies for their healthcare providers to more successfully manage FCS.

PERSPECTIVES FROM THOSE LIVING WITH FCS

"People should know that [having FCS] is hard, and it's restrictive. There is no time limit to this strange diet and 'no, I can't just join in and have one doughnut or join in with an office lunch,' and that it's isolating when it isn't recognized."

"[We live with] the mental fatigue of missing a week of work every month or so and the social and emotional isolation of being hospitalized so frequently."

"[I am most concerned about] Lowering my triglycerides enough to be able to live a normal life [like] be a reliable employee and to enjoy holidays with my family without the fear of hospitalization."

"I feel affected by it every day. [It is like] walking the tight rope all the time."

"[I face] difficulties with travelling away from home and an inability to explore 'foreign' foods. I carry food with me wherever I go and self-cater when away."

"That constant fear of looming pancreatitis... no matter where I am, what I'm doing, that fear is always there—that the 'pancreatitis monster' is looming behind me."

"Being in so much pain that I couldn't be as present as I wanted to be to watch my child grow up. [Sometimes] I am completely debilitated by pancreatitis and [can't] take care of my children."

LIVING WITH FCS (continued)

Disease Management[16-19]

Managing life with FCS requires meticulous attention to a restricted diet designed to prevent pancreatitis which imposes significant physical, emotional, social, and financial impacts on patients and families.

> *When diagnosed, [I saw] 3 choices: either let disease define me, destroy me, or strengthen me.*
>
> — Person living with FCS

Regionally, there are more similarities than there are differences in the FCS experience, although differences in care do exist due to countries' varied health systems whether multi-disciplinary care/centres of excellence are present, easy access to varied food types, and ability to engage with FCS-specific support resources. In general, a large share of the burden for managing FCS is currently shouldered by patients and their caregivers.

With a focus on preventing pancreatitis attacks (which can be incredibly painful, often require days-long hospital stays, and are disruptive of normal work and family life), people living with FCS spend an inordinate amount of time and energy planning and restricting their daily diet. The continuing fear of a pancreatitis attack often severely impacts individuals' social engagement, ability to travel freely, and overall mental health. During a symptom flare-up, management often includes hospitalization, plasmapheresis, liquid-only diets, and the use of insulin.

Measurement of triglyceride levels differs across various countries. While people living with FCS generally acknowledge the need to regularly measure and be aware of their own triglyceride levels, many believe their bodies have their own unique baseline for triglyceride levels and there may be no rhyme or reason for when pancreatitis attacks occur. Despite considerable efforts to adhere to a highly restrictive low-fat diet, many individuals are unable to consistently maintain triglyceride levels below 5.6 mmol/L (500 mg/dL), a threshold recommended by international expert consensus statements and clinical guidelines to help reduce the risk of acute pancreatitis.

While hopeful about a future with multiple effective therapy options, participants in the 2024 lived experience sessions were generally skeptical about the treatments that were available as of November 2024. In addition to listening to their clinicians, patients seek to learn what others in the community think of treatment options, especially understanding potential side effects and whether there will be enough benefit to their daily lives to make using new therapies "worth it."

> *I am hopeful for new treatments on the horizon...*
>
> — Person living with FCS

LIVING WITH FCS (continued)

> *[I hope that] we could have a social life that didn't require pre-planning...*
>
> — Person living with FCS

People with FCS and their families are acutely interested in recipes and food options that can be "safe" for them, while still providing sufficient nutrition, variation, and opportunities for enjoyment. Finding appropriate ingredients for use in FCS-friendly recipes (e.g., alternatives to fat) can be a challenge, especially in certain parts of the world, where options are limited and costly. Going out to events or restaurants, for work or pleasure, can feel like being in a mine field, where skepticism is necessary, even where food has been pre-arranged to be 'FCS friendly.'

There is tremendous burden placed on caregivers to support their loved ones living with FCS, from helping to plan and prepare meals, to managing physical and psychosocial impacts of the disease, to serving as a key social outlet in the face of isolation associated with FCS, to advocating with doctors, insurers, chefs, and others whose support is needed and often not being given.

Understanding FCS

Even though there are more providers today that know about FCS than 5-10 years ago, gaps in provider understanding of FCS must also be addressed. Despite the expertise of lipid specialists, there remains a lack of sufficient physician knowledge and awareness in the broader healthcare system, especially given the rarity of FCS. This reality has created the strong feeling among patients and caregivers that they must be their own disease experts. As a result, those who contributed in the meetings felt they were typically more knowledgeable about their condition than their treating physician team due, in part, to their long journey to diagnosis, having to self-advocate to arrive at diagnosis, and the rarity of their condition.

To support these efforts and help patients and caregivers find the right resources at the right time during their FCS journey, there is a clear and continuing need for an easy to find resources for people within the FCS community to find key information about living with FCS. Currently there are several often-frequented online sources of excellent information, including patient organizations aiming to provide adequate resources, although it is not always easy for families in the throes of seeking a diagnosis, dealing with an acute pancreatitis attack, or managing their care to find or navigate.

> *We look for a way to embrace life.*
>
> — Person living with FCS

Additionally, patients and caregivers emphasize the importance of the language used to discuss FCS. There is a desire to see the narrative around FCS evolve from a series of negatives and prohibitions toward something more positive. So much of the dialogue is focused on what people with FCS "can't" do; they want to know what they "can" do. Additionally, people with FCS who have pancreatitis often feel like this represents a "failure" on their part to comply with their diet or exercise goals to manage their disease.

PERSPECTIVES FROM THOSE LIVING WITH FCS

"I've managed and driven my own care more than any doctor I've ever seen... I call my mom, and I know what I need because I've been doing it for 15 years."

"[It is like] walking the tight rope all the time."

"I live with FCS, but it is not my life. I have goals and dreams and a family that depends on me. I take my responsibilities seriously. Sometimes I'm tired. I'm often in pain. But I'm doing my best. To my family, I'm sorry for what this has put you through. I see your pain too. To my doctors, please listen to me."

"My family is in need of proper support as well as I am."

"[To be successful in managing my FCS] I stick to my diet goals, keep looking for support options that are out there, educate the public and healthcare world, find a medication that helps without debilitating side effects."

"I want real recipes that people will actually eat."

"It would be good to have support in managing mental health and not be made to feel that you're just lab results on a piece of paper."

"[I wish people knew] Just how difficult it is to live with the condition day to day. Maintaining an incredibly restricted diet, keeping a positive outlook and avoiding pancreatitis."

"People should understand that FCS doesn't disappear just with the blink of an eye. Diet is not a choice; it is matter of life. We feel alone."

"I carry food with me wherever I go and self-cater when away."

"I am concerned that as [my daughter] gets bigger, she will lose "her[self]" and her joyful spirit. Managing FCS may break her mentally and physically. It already has affected my health in multiple levels, but it will be a lot harder for her in the future. [I am concerned that] she will be exhausted during studies, hormonal changes, etc. That she will not have a chance to have her own family."

UNMET NEEDS

Stemming from the lived experience engagements with these individuals and their candid discussions of challenges and opportunities they perceive, a series of critical, priority unmet needs for the FCS community has emerged. These include:

Diagnosis

- A more streamlined and straightforward FCS diagnostic process
- More timely, affordable, and broad-based access to genetic testing to confirm FCS diagnosis
- There should be universal newborn screening that includes genetic testing for FCS
- For patients hospitalized with pancreatitis whose triglyceride levels are high and who state that they are not alcohol or drug induced, other possible causes should be explored as standard

Disease Management

- More effective therapies for FCS to reduce triglycerides and reduce the risk of pancreatitis, to help people live more "normal" lives
- Routine access to regular triglyceride testing
- People in all geographies need access to appropriate food ingredients, and recipes that can augment the food options for people with FCS

Awareness and Information

- A wide array of healthcare providers could benefit from enhanced awareness and understanding of FCS
- Our society needs to better understand FCS and its impact on families' lives
- There should be an easily accessible central repository of information about FCS

Community

- There are opportunities to enhance community engagement among people with FCS, their caregivers, and their healthcare providers
- The FCS community should engage with the wider lipidemia and rare disease communities

> *I would not see the diagnosis of 'FCS' as a threat but as a hope. I would point out my own experience of having to live with severe symptoms for 38 years and not knowing why. With this knowledge, the patient can do something about it together with the doctor. Without knowledge, you are groping in the dark.*
>
> — Person living with FCS

STORIES

Julie's Story

JULIE, MOTHER AND WIFE **Living with FCS in Minnesota, U.S.**



Julie started to notice something was off when she had blood labs drawn as part of health screenings for travelling abroad in early 2000, ahead of her 18th birthday. Her triglycerides came back over 55.6 mmol/L (1,000 mg/dL), although no follow up was taken and there was no connection made to FCS by her healthcare team at the time. In hindsight, her symptoms began during puberty.

Julie's first episode of acute pancreatitis occurred when she was 20 years old. Her doctor believed that the pancreatitis was caused by elevated triglycerides due to birth control she was prescribed to regulate her menstrual cycle to alleviate symptoms she had from PCOS (polycystic ovary syndrome). The birth control caused her triglycerides to spike to 111.1 mmol/L (2,000 mg/dL) and she was admitted to the hospital for over a week. Julie's memory was that her first pancreatitis attack was one of her worst. She was unconscious for much of her hospital stay. Upon discharge from the hospital, she was prescribed medicine, discontinued use of hormonal birth control, and was not referred to any form of specialist for follow up. Life began to get back to normal.

Shortly after Julie and her husband had their son at age 28, her pancreatitis attacks returned. Every month, like clockwork, she had an attack and was hospitalized. The first month her triglycerides were 111.1 mmol/L (2,000 mg/dL). The next month they were 166.7 mmol/L (3,000 mg/dL). Every month her triglycerides went up by 55 points until they reached over 555.6 mmol/L (10,000 mg/dL). In the first two years of her son's life, she spent seven to ten days every month in the hospital with a pancreatitis attack, while her son and husband were home without her and in fear for her health and safety.

After two years of this constant crisis, Julie began to explore surgical interventions to try to get to the root cause of her high triglycerides and recurrent pancreatitis. Over the course of about seven years, she saw numerous doctors determined to identify what was causing her sickness. She had a number of procedures and surgeries to attempt to fix what was causing her health problems, with no alleviation or solutions, rather many serious setbacks and complications making her even more sick, including a radical hysterectomy at 34, preventing her from continuing to build a larger family.

Julie's Story (continued)

It wasn't until she was, by fluke, visiting a new hospital for a plasmapheresis procedure that a practitioner told her that she had chylomicrons in her blood after seeing her blood draw. She had never heard of chylomicrons before, leading her to search the internet to learn about chylomicrons. This brought her to a website with information on FCS. She took the information she found online to her lipidologist who screened her for chylomicrons. This led to Julie's diagnosis in Fall 2017, three months after learning about chylomicrons.

Julie's diagnosis provided her relief because she finally had a name to call it. She knew that if it had a name, she was not alone and that there were other people who could relate. Within a month, she was able to meet other people living with FCS for the first time at an in-person meeting hosted by the FCS Foundation (a U.S. patient organization). Over her 25-year journey, she has seen countless doctors at a handful of hospitals. Julie and her family made the decision to relocate to be closer to an academic hospital for care. She currently manages her FCS with the support of her endocrinologist. Her diagnosis and long-term navigation of the healthcare system brought her to change her career into healthcare to help others who are also navigating care.

Providing a voice for FCS and sharing her story is very important to Julie because she wants to ensure others don't have to experience what she had to in order to reach a diagnosis. Her advice for a newly diagnosed patient is:

> Give yourself some grace. You are going to have days where you struggle, and it is okay to rest. Ask for help. Find your support system. A support system includes family or friends who can help you with managing your care plan, getting to appointments, getting to the hospital if you need it. Also, an extended support system, like online communities where you can ask for advice and meet people who get it. You are not alone. Do not be discouraged. This disease does not define you. You can still have a life. You can still have dreams and goals. Make plans. You will learn to be flexible and make adjustments, but don't stop dreaming.

Scott's Story

SCOTT, HUSBAND AND FATHER OF THREE SONS | **Living with FCS in North Carolina, U.S.**



As an adolescent, Scott was aware that he had very high cholesterol, although no further exploration or testing was done to get to the root cause. In college, he started having bad health concerns and noticed eruptive xanthomas on his skin and started having stomach pains with no known cause. In his 20s, his doctor decided to test his triglycerides, and his numbers were "through the roof," although no action was taken. He was directed to keep an eye on it. Scott had his first pancreatitis attack at 30 years old. Scott's triglyceride levels once reached 1,333.3 mmol/L (24,000 mg/dL) while hospitalized with pancreatitis. It was a long wait for a diagnosis for Scott and his family. Scott dealt with 23 years of symptoms and over a decade of persistent pancreatitis attacks ahead of his diagnosis.

Scott faced difficult stigmas with his regular pancreatitis hospitalizations because many doctors questioned the role he played in causing his pancreatitis. He was accused numerous times of being an alcoholic and drug addict searching for pain medicine. Scott found himself in the ICU for two weeks every eight weeks for almost a year at the height of his misdiagnosis while he tried his best to be a husband, father to three young boys, and provide for his family. Scott had to resign from his demanding job due to his health challenges and time in the hospital. He was desperate for answers and incapable of having a positive quality of life.

The middle of the night trips to the hospital where he would admit himself was difficult because his children would wake up to their dad gone. His children knew they wouldn't see their dad for two weeks whenever he was in the hospital. Scott felt completely isolated as though he was on an island by himself through all of this. During this time, Scott continued fighting for a diagnosis and his health. His motivation for fighting was his family.

Scott lends his diagnosis to his own determination, self-advocacy, and independent online research. Scott believes firmly in taking your care into your own hands and that you are your best advocate. Scott would search, "reoccurring pancreatitis attacks," "why are my triglycerides so high," and "why is my blood milky?" The online search finally brought him to the patient advocacy organization, Action FCS, where his relationship with fellow patient and founder, Jill Prawer, began.

Scott's Story (continued)

Scott armed himself with the information he learned and presented it to his endocrinologist to help aid in his ultimate diagnosis. He expressed that learning about FCS and being able to put a name to it was freeing. He could no longer get upset and mad at himself when a pancreatitis attack would happen because there was a reason for the attack. Scott felt relief and comfort to know there are others in the world with FCS. He is grateful to the FCS community for helping him establish a new outlook on life and for the support they provide. After attending his first patient meeting hosted by the FCS Foundation (a U.S. patient organization) in Spring 2024, he believed he could now live a full, long life (after meeting someone 75 years old living with FCS), something he had previously felt was impossible. After returning home from the conference, his resolve to be fully on board with the special diet of less than 10g of fat per day has made all the difference in his health.

Scott believes that supporting his mental health by seeing a therapist regularly is critical for his successful management of FCS. He additionally focuses his time to help others with FCS (or those wondering if they have FCS) so they don't have to experience the struggles that he went through.

Scott's advice for newly diagnosed patients:

> "I would encourage them to find another patient who is willing to help guide them through the process – almost like a mentor or sponsor. I would provide practical tips on switching from a traditional diet to an extremely low-fat diet. Form a medical team not just a single doctor that will collaborate on their care. I would also encourage the new patients to show themselves grace. There will be many opportunities to get very frustrated, take a break and grieve the diagnosis and get back on the horse to ride again."

AMPLIFYING VOICES & DRIVING CHANGE FOR FCS

There is much that can and should be done to improve the experience of people dealing with FCS. By elevating and amplifying their voices, we can collectively help to drive awareness of FCS among healthcare providers and spur action by all those invested in improving the lives of people with FCS. General public awareness is also important to help support the FCS community and their families in leading the most 'normal' lives possible.

It is equally important to share the voices of the community to help others who might be suffering alone. Our goal is to help pave the way for a more hopeful future, by empowering advocates to raise awareness of the challenges for people living with FCS, improve general provider knowledge, strengthen connectivity within the community, and advocate for systemic changes to improve care across geographies.

When we asked the patients and caregivers who participated in our meetings about their hope for the future, they talked about a wish for consistently low triglyceride levels, reduced risk of pancreatitis or hospitalization, improved organ health, and the opportunity to experience "normal" life through social gatherings, career, family, travel, and their overall wellbeing.

How **YOU** Can Help Drive Change

Healthcare Providers

- Stay updated on FCS symptoms and diagnostic criteria
- Advocate for routine triglyceride testing in cases of unexplained pancreatitis

Policymakers & Industry

- Support newborn screening for FCS where feasible
- Advance access to genetic testing
- Expand funding for research on targeted therapies

Patients & Caregivers

- Share your story to spread awareness
- Consider a clinical trial if appropriate and available
- Join advocacy organizations to strengthen your support network and resources, build the community, and push for better healthcare policies

AMPLIFYING VOICES (continued) & DRIVING CHANGE FOR FCS

VOCALIZING HOPE

"I am hopeful for new treatments on the horizon that will allow us to be able to function more 'normally' in social gathering and most importantly stay healthy and pancreatitis free."

"I hope to use my paid time off (PTO) for vacation, not sick days."

"I hope to have the time to focus on important health needs other than my triglycerides."

"[I hope for a future where] my daughter who also has FCS can enjoy a birthday cake at a kids' birthday party with all the other 'normal' kids."

"I hope to have a social life that didn't require pre-planning and pre-packing of food, to travel with abandon and not worry about what to eat."

"I hope that my parents [could get] to travel instead of taking care of me."

"[I hope for] lowering my triglycerides enough to be able to live a normal life. For example, to be a reliable employee and to enjoy holidays with my family without the fear of hospitalization."

"I hope my daughter with FCS won't have to feel anxious about passing it on [to her children]."

"I hope for earlier diagnosis for patients with FCS symptoms."

"My hope for the future is early diagnosis for patients with symptoms of FCS. Financially affordable medications to reduce triglycerides and the probability of pancreatitis without side effects."

AMPLIFYING VOICES (continued) & DRIVING CHANGE FOR FCS

VISUALIZING HOPE

At the lived experience convenings, we asked the participants, "When you visualize hope for the future given the possibility of treatments on the horizon, what does it look like?" Here are some of their visual interpretations of "HOPE":







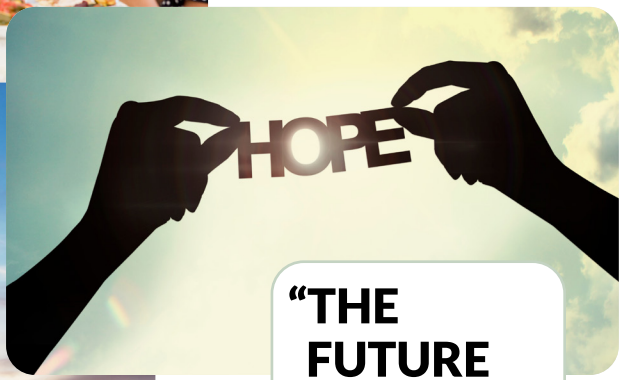












"THE FUTURE LOOKS NORMAL"

Being able to dine at a 3 Michelin star restaurant and eat the chef's menu

AMPLIFYING VOICES (continued) & DRIVING CHANGE FOR FCS

Opportunities for Progress

There are multiple near-term opportunities for progress and specific action steps for the community that emerged from the insights we gathered. Among those are:

Awareness & Education for FCS Families

- Creating and distributing information and support specific to the FCS experience and level of knowledge (newly diagnosed through to an experienced patient)
- Raising the profile of the FCS-specific community to share insights, advice, recipes and strategies specific to FCS
- Supporting self-advocacy, e.g., creating a hand-out for healthcare providers explaining FCS and a protocol for emergency care to take to the emergency room during pancreatitis attacks

Awareness & Education for Providers

- Developing and disseminating appropriate educational resources for healthcare providers
- Enabling patients/caregivers to meet with clinicians at conferences to help "put a face" on FCS and communicate the critical needs of patients and caregivers
- Developing patient/caregiver testimonials to help describe the impact of FCS to the broad range of healthcare providers that connect with patients

Improving FCS Diagnosis, Management & Care

- Advancing newborn screening for FCS to ensure timely confirmation of FCS diagnosis
- Developing a global FCS patient charter/care guidelines (to include routine support for mental health) that can be used as models by various regional organizations to raise the baseline standard of care for FCS
- Enhancing support for triglyceride testing
- Partnering with/expanding centers of excellence to provide multi-disciplinary care for FCS

Enhancing the Reach & Impact of the Global FCS Community

- Building online presence/boosting search engine optimization (SEO) for FCS searches (one-stop shop specifically for curated FCS information) to make it easier for families and healthcare providers to find valid information
- Building patient ambassador programs to expand the reach of the FCS community voice to key stakeholders
- Continuing to convene the global FCS community and expand support groups to bolster engagement and empowerment
- Connecting with organizations focused on similar conditions (e.g., pancreas organizations) to deepen the power of the FCS advocacy community

CONCLUSIONS

People living with FCS and their families demonstrate extraordinary courage, resilience, and vision. The insights shared by those we met with through this initiative have had a profound impact and place into clear focus the opportunity for all interested to take meaningful action on behalf of this community.

We look forward to partnering with our advocacy colleagues in sharing this information broadly and leveraging the power of the patients' and family members' lived experiences toward better quality of life and outcomes for everyone impacted by FCS.

ACKNOWLEDGEMENTS

We thank immensely the individuals who shared about their experiences with FCS to inform our learnings and inspire the creation of this paper to serve the broader community. We hope to honour and support their goals for raised awareness for FCS through the dissemination of this paper.

The Arrowhead team is grateful for the engagement and support of the FCS Foundation, Action FCS, and FH Europe Foundation in the creation of this paper and for assisting with the convenings described in this paper. We also thank the team at HAVAS Health and WSCollaborative for their guidance and expert facilitation of these sessions.

The content of this paper is intended for educational purposes only, and is not intended as medical advice. You should always seek the advice of your doctor or qualified healthcare provider if you have questions regarding a medical condition or treatment.

APPENDICES

Appendix A. Definitions

Appendix B. References

Appendix C. Lived Experience Convenings Pre-Meeting Activity

Appendix D. Global Patient & Caregiver Advisory Board Discussion Guide

Appendix E. Lived Experience Group Participants

Appendix F. NORD's Rare Diseases & Orphan Products Breakthrough Summit, October 19–21, 2025, Washington, DC

APPENDIX A

DEFINITIONS

Chylomicrons are lipoprotein particles involved in delivering dietary lipids such as triglycerides, cholesterol and fat soluble vitamins from the intestines to muscles and adipose tissue for energy storage.

Eruptive xanthomas are small bumps that appear on the skin, typically on the extensor surfaces (back of hands, knees, elbows), that are caused by high levels of triglycerides in the blood

Lipemia retinalis is an eye disease caused by high amounts of triglycerides in the blood or lipoprotein lipase deficiency (chylomicronemia), presenting with a creamy-white discoloration of the eye

Hepatosplenomegaly is the simultaneous enlargement of both the liver and the spleen

Exocrine deficiency is the impaired function of glands such as the pancreas that fails to secrete adequate digestive enzymes into the small intestine which leads to poor digestion and malabsorption of nutrients resulting in nutritional deficiencies

Steatorrhea is the presence of excess fat in feces

Hypertriglyceridemia is elevated triglycerides or high triglycerides

Acute necrotic fluid is a condition that can occur in the pancreas and can be caused by necrotizing pancreatitis, where part of the pancreas dies and causes fluid accumulation in a body cavity and can lead to complications including infection, inflammation, organ dysfunction.

Plasmapheresis is a procedure in which the liquid part of the blood, or plasma, is separated from the blood cells, and where the machine may clean the plasma and/or substitute it for healthy plasma. For FCS patients, this helps to reduce the chylomicrons in the blood and immediately lowers triglycerides in the blood for a short period of time.

MEASUREMENTS

mg/dL = milligrams per deciliter

mmol/L = millimoles per liter

Conversion for mg/dL and mmol/L: Take the mg/dL and divide by 88 to reach mmol/L. Conversely, you multiply mmol/L by 88 to reach mg/dL.

APPENDIX B

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APPENDIX C

LIVED EXPERIENCE CONVENINGS PRE-MEETING ACTIVITY

1. My biggest goal in FCS management is:

2. As I manage my FCS, I am most concerned about:

3. I believe my triglyceride levels should be:

4. To be successful in managing my FCS, I need to do:

5. If you were giving advice on physical and emotional needs to a newly diagnosed FCS patient or to yourself when you were first learning about FCS, what would you say? What would you provide in terms of support and education?

6. What do you find you need most in terms of education and support now, knowing your diagnosis?

7. What do you wish people (your doctor, family, partner, friends, coworkers, etc.) knew about FCS?

8. What are moments in your life you have felt most affected by FCS?

9. When you visualize hope for the future given the possibility of treatments on the horizon, what does it look like?

APPENDIX D

Global Patient & Caregiver Advisory Board Discussion Guide

Getting to Diagnosis

- What 2-3 words would you use to describe the process you/your loved one went through to be diagnosed with FCS?
- What barriers did you encounter along the way to the diagnosis? How could those barriers have been removed/reduced?
- What 2-3 words would you use to describe how you felt about this diagnosis?
- What information was most important to you (or do you wish you had received) upon diagnosis?
- What would you most like for a newly diagnosed person to know from the start?

Managing FCS

- How do you explain FCS to someone who doesn't know what it is?
- Define the pancreatitis safe zone for triglycerides.
- What is the most important thing to know about managing FCS?
- What type of healthcare provider/physician/clinician do you rely on for helping you manage your/your loved one's FCS?
- In what areas of your life do you feel there is a gap in support for FCS management and what would be helpful in those areas? What about for your loved ones?
- Where do you feel self-advocacy support is needed most? At the hospital, your doctor, work, friends, family, etc. And what do you feel is needed?

Goals for Treatment/Unmet Needs

- What treatments are part of your FCS experience?
- What are the benefits and challenges of those treatments?
- What do you wish a treatment for FCS could do for you/your loved one?

Information Gaps/Unmet Needs

- What information is most important to you as you navigate your life with FCS? Topics? Format?
- Where do you turn for information about FCS? How easy is it to find the information you need when you need it?
- What information is missing/hard to find? What do you most wish you had throughout your journey?
- When learning about FCS, do you find yourself reviewing or wanting to review materials more scientific in nature (e.g., clinical research activities), and do you find this difficult in any particular way?

Engaging with Community

- How did you find the FCS community you are engaged in today?
- What role does community play in your experience with FCS? In that of your loved ones?
- What additional opportunities for community do you wish you had?

Hopes for the Future

- What 2-3 words would you use to describe your hopes for the future in managing/treating FCS?
- How do you think your experience with FCS might have been different if you had been diagnosed in 2024?
- How would you hope it could be different for someone diagnosed in 2025 and beyond?

APPENDIX E

LIVED EXPERIENCE GROUP PARTICIPANTS

U.S. Lived Experience Insights Group:
5 patients – 2 men, 3 women

- Two participants are from the east coast, one from the midwest, one from the south and one from the west coast.
- One participant was diagnosed as an infant, the others later in life.
- Two were diagnosed by lipid specialists, two were diagnosed by endocrinologists, and one by at a hospital by a doctor newly out of medical school.
- Three have their conditions managed by endocrinologists, and two have their conditions managed by lipid specialists.
- Three participants experienced symptoms for many years (12-25 years) prior to diagnosis, and multiple participants have experienced episodes of acute pancreatitis.

Global Patient & Caregiver Advisory Board:
9 participants – 7 patients, 2 caregivers, 4 men, 5 women

- Three individuals diagnosed in childhood, all others later in life after years of symptoms, acute pancreatitis episodes/attacks, and/or misdiagnoses.
- Five individuals are from the U.K., one is a caregiver from Estonia, one is from the Netherlands, one is from Austria, and one individual is from the U.S.
- At least three individuals were not diagnosed until in their 40s.

Both groups contained participants who were diagnosed after bringing independent research to their physicians for consideration.

APPENDIX F

NORD's Rare Diseases & Orphan Products Breakthrough Summit, October 19–21, 2025, Washington, DC

The *Spotlight on FCS Voice of the Patient Paper* was selected as a top poster at the NORD Breakthrough Summit 2025 in the category of patient engagement and education, representing the forefront of innovation and advocacy in the rare disease space. The meeting brought together more than 800 participants from across the rare disease community, including researchers, policymakers, companies, and patient advocates. This recognition gave us the opportunity to present in a main session to raise awareness for FCS, highlighting the community's unmet needs, and ways to improve awareness, diagnosis, and care.

Spotlight on Familial Chylomicronemia Syndrome (FCS): Leveraging the Power of Individual Journeys Toward Collective Action – Voice of the Patient Paper

Alexandra Roeser, Wendy Selig

PURPOSE

Significant gaps remain in understanding the true impact of FCS on the lived experience community.

A lack of disease awareness among both healthcare providers and the public often leads to underdiagnosis and misdiagnosis

To help address gaps, several FCS lived experience convenings were held exploring challenges faced by individuals affected by FCS, helping to identify opportunities for progress.

One key finding was the strong need and desire to increase awareness and understanding of FCS, especially among providers – as this is vital for timely diagnosis and improved care management.

A ***Voice of the Patient*** paper was published to outline and highlight FCS patient and caregiver insights as a community resource and tool.



BACKGROUND ON FCS

Ultra-rare, serious genetic condition that affects how the body processes fat from food.

- FCS prevents one from properly processing dietary fats and causes blood to have extremely high levels of triglycerides.
- People with FCS have little to no response to conventional lipid lowering therapies.

Most serious complication is recurrent acute pancreatitis caused by having extremely high triglycerides.

Managing FCS requires significant lifestyle changes and adhering to strict dietary restrictions, which can be quite stressful.

FCS affects both mental and physical well-being.

METHODS

This qualitative study aimed to collect and analyze FCS lived experience insights through guided convenings in the U.S. and Europe in Fall 2024.



RESULTS

Overarching themes were collected relating to the FCS lived experience:

- Diagnostic timelines must be shortened as too many people currently suffer through extended, frustrating daily living journeys.
- Most FCS management requires meticulous attention to a restricted diet designed to prevent acute pancreatitis episodes. This imposes significant physical, emotional, social, and financial impacts.
- There is a need for education and advocacy about FCS in the healthcare community to address gaps in understanding and supporting patients.
- Families seek easy-to-find resources for accurate, objective FCS information.
- Self-advocacy is crucial.
- Connection and support within the FCS community are vital.
- Patients and caregivers seek to live a "normal" life without the constant pressures of FCS.

"Spotlight on FCS" Paper Demonstrates Direct, Positive Impact on the Patient Community

> *"As an FCS patient, the 'Spotlight on FCS' paper has become one of the most valuable tools in my FCS Toolbox. When you live with a rare disease, being able to advocate for yourself is absolutely vital... paramount when it comes to navigating doctor's appointments, hospitalizations, unexpected trips to the emergency room and even when navigating daily life with people who don't know what FCS is and what it means to live with this disease. Education goes hand in hand with Advocacy. I can't effectively advocate for myself if I can't educate people about FCS. Being able to hand that paper to my friends, family members and healthcare providers has given me a voice...*
>
> *It covers every aspect of the patient journey and validates our experience to people who have little to no understanding of FCS. That, to me, is priceless."*
>
> *– Person living with FCS*

CONCLUSIONS

Multiple near-term, multi-stakeholder opportunities for progress have emerged based on insights from patients and caregivers. These include specific, actionable strategies to enhance awareness, education, and advocacy utilizing technology or communications. The resulting *Voice of the Patient* paper serves as a foundation for collaborative efforts across advocacy, education, and clinical care.

OPPORTUNITIES FOR PROGRESS

Awareness & Education for Providers and Families

Providers

- Disseminate appropriate educational resources
- Enable conference engagement with patients/caregivers and HCPs to "put a face" on FCS and communicate needs
- Develop testimonials to describe the impact of FCS

Families

- Distribute tailored information of FCS experience to support level of knowledge
- Raise the profile of the FCS community to facilitate sharing of insights, advice, recipes and strategies
- Support self-advocacy

Improving Diagnosis, Management & Care

- Advance newborn screening to ensure timely diagnosis
- Develop a global FCS patient charter/care guidelines (including mental health) that can be used to raise the baseline standard of care for FCS
- Enhance support for triglyceride testing
- Partner with and expand centers of excellence to provide multi-disciplinary care

Enhancing the Reach & Impact of the Global FCS Community

- Build online presence/boosting search engine optimization (SEO) for FCS searches (one-stop shop for information)
- Build patient ambassador programs to expand the reach of the community voice
- Continue to convene the global FCS community and expand support groups
- Connect organizations focused on similar conditions to deepen the power of the advocacy community

> *[This community needs] ways to educate others and share to avoid future generations suffering as I have with finding the right diagnosis and right treatment with my care team.*
>
> *– Person living with FCS*

> *When diagnosed, [I saw] 3 choices: either let the disease define me, destroy me, or strengthen me.*
>
> *– Person living with FCS*

> *I would not see the diagnosis of 'FCS' as a threat but as a hope. I would point out my own experience of having to live with severe symptoms for 38 years and without a timely diagnosis. With this knowledge, the patient can do something about it together with the doctor. Without knowledge, you are groping in the dark.*
>
> *– Person living with FCS*

> *[I need] connection with other patients as they travel the same journey with FCS, there is so much power and support in the stories of other's journeys.*
>
> *– Person living with FCS*

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Poster presented at NORD Summit summarizing themes from the Spotlight on FCS paper