



PFIC Network

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Progressive Familial Intrahepatic Cholestasis (PFIC)

is an umbrella term referring to a group of rare liver disorders caused by mutations in genes controlling bile composition, bile acid secretion, or transport. PFIC typically presents in the first year of life with jaundice and debilitating pruritus, and leads to liver failure requiring liver transplant over time, though some presentations are diagnosed later in life. Two medical treatments for pruritus have emerged within the past few years with close to one-third of the patient community reporting symptom improvement, but liver transplant remains the only curative option.



The PFIC Network Leadership Team — including staff, board and Scientific Medical Advisory Board members — at the 2025 Cholestatic Liver Disease (CLD) Summit in Aurora, Colorado, which the PFIC Network cohosted in partnership with the Alagille Syndrome Alliance, Biliary Atresia Research Education and Children's Hospital Colorado.

During the Grant Period

Impact Spotlight

During the grant period, the PFIC Network focused on building meaningful community engagement in research through family-scientific convenings, patient-driven data efforts, and Project IMPACT (Identifying research targets by Merging Patient And Clinician Treatment information). During this period, the organization hosted three major convenings, achieving a 30% year-over-year increase in attendance. These gatherings included four roundtable sessions that surfaced critical gaps in clinical management and treatment development, underscoring the urgent need to reduce uncertainty in treatment pathways for families affected by PFIC.

In response, the PFIC Network launched Project IMPACT — the first initiative to systematically map all

available treatments for all PFIC subtypes alongside their clinical and patient-reported outcomes. This work resulted in the Treatment Experience App (TEA), an interactive web platform that empowers patients, caregivers, and clinicians to explore real-world treatment experiences and outcomes.

Through additional surveys and focus groups, the PFIC Network engaged over 150 patients, parents, and clinicians to define research priorities and outline how the community can serve as equal partners in future studies. The culmination of this work is the [PFIC Roadmap](#), built on community-defined priorities and practical guidance for patient engagement in research, published in summer 2025.

Key research and research infrastructure achievements

- Hosted three patient-researcher convenings, including

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the first-ever PFIC scientific meeting in partnership with Cincinnati Children's Hospital.

- Re-launched the PFIC Network Patient Registry on the REDCap platform, with 140 patients enrolled by the close of the grant and 158 enrolled as of July 1, 2025.
- Catalyzed cross-sector collaboration by joining initiatives such as FasterCures LeadersLink, the Forum for Collaborative Research's Pediatric Cholestatic Liver Disease Steering Committee and PFIC Working Group; outcomes include a forthcoming white paper on patient perspectives in clinical trial risk-taking.
- Supported research dissemination by attending eight major scientific conferences, including the American Association for the Study of Liver Diseases (AASLD), the North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition (NASPGHAN), and the European Society for Paediatric Gastroenterology, Hepatology, and Nutrition (ESPGHAN). Notably, a poster abstract on the PFIC Network Patient Registry was presented at the 2024 NASPGHAN meeting, and since 2022, a booth has been hosted annually at ESPGHAN to promote disease awareness and provider education globally.
- Implemented a two-year PCORI Engagement award project titled IMPACT (Identifying research targets by Merging Patient And Clinician Treatment information), to build capacity for patient-centered comparative clinical effectiveness research (CER) in PFIC. The project brought together patients, families, clinicians, and researchers to build a shared understanding of patient-centered CER, define PFIC-specific CER targets, and develop strategies for equitable and sustainable patient-researcher partnerships.



The PFIC Network patient, family, clinicians and scientific community at the 2024 Family & Scientific Conference, co-hosted with Cincinnati Children's Hospital Medical Center.

Key publications

- Journal of Hepatology (2024): a paper on the Clinical Practice Guidelines of several genetic diseases of cholestasis, including a detailed approach to a certain subset of these diseases taking into account new treatments; acknowledges PFIC Network Executive Director Emily Ventura as a member of the Delphi Panel of this Clinical Practice Guideline
- Journal of Pediatric Gastroenterology and Nutrition Reports (2024): a poster abstract presented at the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN) Annual Meeting, giving an overview of data from the patient registry. A full paper is currently under review; poster co-authored by PFIC Network Executive Director Emily Ventura

“We believe in a world where every person with PFIC has a diagnosis, understands their diagnosis and has a treatment pathway that is accessible to them. Passion, motivation and determination will drive us forward. We will continue to dream big and take pride in our work, while reminding ourselves that small gains make great strides for our collective future.”



Emily Ventura, BSN, RN
Executive Director, PFIC Network

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Key operational and community achievements

- Hired a full-time Associate Director of Programs and contracted a statistician/science advisor, strengthening program capacity and contributing to a successful PCORI Engagement Award in 2023.
- Expanded fundraising capacity by hiring an experienced Development Director, which has led to an over 300% increase in first quarter funding following the CZI Rare As One grant period.
- Strengthened scientific leadership by appointing a new board member with research expertise, and formalizing the Scientific and Medical Advisory Board with a charter and inaugural Chair.

After the Grant

Since November 2024, the PFIC Network has sharpened its focus on transforming the PFIC research landscape by embedding patients and families as early and consistent partners. This year, we...

- Co-organized the 2025 Cholestatic Liver Disease Summit, a joint multi-disease event focusing on PFIC, Alagille Syndrome, Biliary Atresia, and Primary Sclerosing Cholangitis. The meeting was attended by nearly 400 patients, parents, clinicians, researchers, regulators, and industry representatives focused on rare cholestatic liver diseases.
- Co-authored a paper focused on looking beyond genetic testing results to extend the spectrum of PFIC, to highlight the various different diseases under the genetic diagnosis of PFIC



PFIC Network Executive Director, Emily Ventura, shares her Story of Impact during the CZI Science in Society 2025 Meeting.

Over the next three years, the organization will advance this vision through three key initiatives:

- Expanding enrollment and retention in the PFIC Patient Registry while leveraging tools like the organization's recently completed registry paper to increase researcher interest and promote integration of patient-reported data into research studies.
- Building on the foundation of Project IMPACT by developing a prioritized research agenda based on identified comparative effectiveness research (CER) targets and forming a multi-stakeholder patient-centered research consortium that will provide the infrastructure to collaboratively design and conduct CER projects reflecting PFIC patient priorities.



PFIC Network Executive Director Emily Ventura (center back), Program Director Melissa Kochanowsky (right), and PFIC patient Cedar Ventura (center front) visit Dr. Chunyue Yin's (left) zebrafish lab at Cincinnati Children's Hospital.

- Expanding the annual convening to foster international and cross-disease collaboration. Planned strategies include co-hosting with other rare liver disease patient organizations, holding events outside the U.S., and incorporating more research-focused working sessions.

Collectively, these efforts aim to ensure that PFIC research advances are not only scientifically sound but truly aligned with the needs of patients and families.