Findings From the 2025 Cholestatic Liver Disease Summit Research Roundtable Meetings

A Collaborative Initiative of the Alagille Syndrome Alliance (ALGSA), PFIC Network (PFIC), and Biliary Atresia Research and Education, Inc. (BARE)

Cher Bork[1], Melissa Kochanowsky[2], Jennifer Lau, MPH[3], Jordan Sarbaugh[3], Emily Ventura, RN, BSN[2], Roberta Smith[1]
[1]Alagille Syndrome Alliance/Washington, DC, USA/Email: alagille@alagille.org
[2]PFIC Network/city, state, USA/Email: info@pfic.org
[3]Biliary Atresia Research & Education, Inc./Montgomery, IL, USA/Email: contactus@bareinc.org

Abstract

The *2025 Cholestatic Liver Disease Summit Research Roundtable Meeting,* a collaborative initiative of the Alagille Syndrome Alliance (ALGSA), PFIC Network (PFIC), and Biliary Atresia Research and Education Inc. (BARE), brought together a global, multidisciplinary community of stakeholders to address urgent gaps in rare pediatric and adult cholestatic liver disease care. Through six facilitated sessions: Pruritus, Transplant, Transition of Care, Nutrition, Mental Health, and Comparative Effectiveness Research (CER), participants representing patients, caregivers, clinicians, scientists, and industry partners identified recurring challenges and opportunities across disease conditions.

Participant-reported discussions revealed shared priorities that transcend disease boundaries: improving mental-health integration, strengthening transition pathways, developing standardized nutritional and psychosocial guidelines, and embedding patient-reported outcomes into research and clinical practice. Stakeholders emphasized that scientific progress must be paired with equitable access, data that reflect lived experience, and systems that support the entire family unit.

This white paper synthesizes qualitative, participant-informed insights from the roundtables, offering a cross-cutting framework for collaboration between advocacy and academia. The findings highlight a collective imperative - to align innovation with empathy and evidence with experience, to ensure that children and adults living with Alagille Syndrome, PFIC, and Biliary Atresia receive coordinated, compassionate, and measurable improvements in care and quality of life.

1. Executive Summary

The *2025 Cholestatic Liver Disease Summit* convened multidisciplinary stakeholders from across the globe for a series of Research Roundtable Meetings focused on Alagille Syndrome (ALGS), Progressive Familial Intrahepatic Cholestasis (PFIC), and Biliary Atresia (BA). These sessions examined six core areas vital to the rare cholestatic liver disease community: Pruritus, Transplant, Transition of Care, Nutrition, Mental Health, and Comparative Effectiveness Research (CER).

Each roundtable provided a structured environment for caregivers, patients, clinicians, scientists, and industry partners to exchange insights, identify systemic barriers, and define actionable solutions. The conversations consistently highlighted that although medical innovation continues to advance, patient experience, mental health, and continuity of care remain underrepresented in research and practice.

Across all topics, participants highlighted the need to:

- Integrate lived experience and patient-reported outcomes into research design;
- Strengthen transition and mental health support across the care continuum;
- Develop standardized nutritional, psychosocial, and diagnostic guidelines;
- Expand cross-disease data sharing and registries for ALGS, PFIC, and BA;
- Ensure fair access to treatment, mental health care, and insurance coverage.

This White Paper summarizes the themes, findings, and recommendations that emerged from the roundtable meetings. It offers a qualitative analysis of stakeholder perspectives and provides a foundation for future collaborative initiatives that bridge clinical research and patient experience in rare cholestatic liver disease care and research.

2. Introduction

Cholestatic liver diseases, such as Alagille Syndrome (ALGS), Progressive Familial Intrahepatic Cholestasis (PFIC), and Biliary Atresia (BA), present complex clinical and psychosocial challenges for affected patients and their families. Historically viewed through the lens of pediatric hepatology, these conditions now extend into adolescence and adulthood as advances in care prolong survival. This evolution has created new demands on healthcare systems, including the need for lifelong management strategies that incorporate physical, mental, and social well-being.

Recognizing these challenges, the Alagille Syndrome Alliance (ALGSA), PFIC Network (PFIC), and Biliary Atresia Research and Education Inc. (BARE) collaborated to host the cross-disease Research Roundtable discussions at the *2025 Cholestatic Liver Disease Summit*. The goal was to establish a shared understanding of unmet needs across disease communities and to identify potential opportunities for research, education, and systems-level improvement.

Each roundtable session followed a structured, discussion-based model designed to:

- Capture insights from all stakeholder groups,
- Highlight barriers and emerging solutions,
- Identify actionable themes to inform research and policy, and

- Encourage collaboration across clinical and advocacy sectors.

This White Paper documents those findings, synthesizing key insights and recommendations for the broader rare cholestatic liver disease community. It reflects a unified belief: that improving outcomes for patients with cholestatic liver disease requires partnership between patients, caregivers, clinicians, researchers, and industry, working together to define what truly matters in care and research.

3. Methods

The Research Roundtable Meetings were conducted as part of the *2025 Cholestatic Liver Disease Summit*, a multi-day conference held June 26–28, 2025, in Aurora, Colorado. The Summit convened a global, multidisciplinary cohort of stakeholders, including patients, caregivers, clinicians, researchers, and pharmaceutical/biotechnology industry representatives. The objective of the roundtable sessions was to capture stakeholder perspectives to guide future research priorities, clinical practice, and patient-centered care strategies for Alagille Syndrome (ALGS), Progressive Familial Intrahepatic Cholestasis (PFIC), and Biliary Atresia (BA).

A facilitated, qualitative data collection approach was employed using structured small-group discussions. A total of 28 roundtable groups were organized across six predefined thematic domains: Pruritus, Transplant, Transition of Care, Nutrition, Mental Health, and Comparative Effectiveness Research (CER). Groups were assembled using a purposive, mixed-stakeholder model to ensure representation across patient, caregiver, clinical, and research perspectives, with an average of approximately eight participants per table.

To allow participants to contribute to multiple areas of interest, a rotational discussion model was implemented. Each participant selected two topic areas prior to the session and participated in two sequential 60-90 minute discussion periods. Participants initially engaged at their first-choice table and, at the midpoint of the session, transitioned to a second table aligned with their additional area of interest. This approach enabled broader stakeholder input across multiple domains while maintaining focused, topic-specific discussions.

Each table was provided with a standardized set of topic-specific prompts and paper-based data collection worksheets (Appendix A). Participants were instructed to engage in open dialogue around key challenges, potential solutions, and research needs while documenting individual and group responses directly on the worksheets. A trained facilitator supported discussions to promote balanced participation and ensure consistency in data capture across groups.

All completed worksheets and supplementary notes were collected immediately following the session. Data were aggregated and subjected to qualitative content analysis using an inductive thematic synthesis approach. Coding and theme identification were conducted collaboratively by investigators from the Alagille Syndrome Alliance (ALGSA), PFIC Network (PFIC), and Biliary Atresia Research and Education, Inc. (BARE). Recurring patterns, salient insights, and representative quotations were identified and synthesized into overarching themes and domain-specific findings.

Given the qualitative and exploratory nature of the study design, findings are not intended to be generalizable but rather to provide a rich, stakeholder-informed understanding of current challenges and opportunities. The results are intended to inform hypothesis generation, guide future research design, and support the development of patient-centered care models across cholestatic liver diseases.

4. Findings by Discussion Domain

The following findings reflect themes and insights directly reported by roundtable participants across stakeholder groups. They represent a synthesis of participant perspectives and are not intended as formal organizational positions or population-level conclusions.

A. Pruritus

Pruritus remains one of the most burdensome and least quantifiable symptoms in cholestatic liver disease. Patients and caregivers described severe sleep disruption and emotional distress but noted that clinicians often underestimate the impact due to limited measurement tools. Stakeholders agreed that sleep patterns provide the most reliable proxy for itch severity and that wearable technologies (such as watches or apps) could enable objective tracking.

Some patients reported that IBAT inhibitors offered meaningful relief for some patients but also brought side effects and complex insurance barriers. Patients and caregivers expressed willingness to co-develop digital itch-tracking tools, including surveys and wearables, and advocated for clinician education to recognize non-obvious symptoms.

Implication: There is a strong community appetite for patient-led innovation. Co-created digital tools could redefine pruritus assessment, bridging subjective experience with measurable data.

B. Transplant

The transplant discussion revealed immense variability in patient journeys—from infants to adults—alongside shared concerns about medication side effects, transition readiness, and post-transplant isolation. Tacrolimus dominated the conversation; while effective, it carries unpredictable side effects and long-term kidney risks. Patients and caregivers called for improved education, more transparent communication, and research into alternative immunosuppressants, noting a need for more consistent, accessible information for both clinicians and families regarding long-term risks, side effects, and management strategies.

Participants also emphasized early transition of care planning, noting that adolescence is a critical period for medication adherence and psychosocial adjustment. Support programs for caregivers and peer connections were viewed as essential for long-term success.

Implication: Improved education, shared learning networks, and caregiver inclusion can reduce post-transplant complications and foster lifelong engagement in care.

C. Transition of Care

Transitioning from pediatric to adult care was described as one of the most anxiety-inducing experiences for patients and caregivers. Stakeholders identified inconsistent communication, inadequate preparation, and limited adult provider knowledge as major gaps. Participants advocated for standardized national transition protocols and Centers of Excellence dedicated to bridging pediatric and adult hepatology.

Patients and caregivers emphasized the importance of mental health and self-advocacy training beginning in early adolescence. Incorporating digital tools like AI note-takers or transition apps was viewed as a practical next step.

Implication: Successful transition requires structure, communication, and social-emotional readiness. Establishing consistent frameworks will prevent loss to follow-up and improve continuity of care.

D. Nutrition

The nutrition discussion exposed deep inequities in access to vitamins, supplements, and coverage for medical nutrition products. Patients and caregivers face inconsistent guidance from providers and limited reimbursement for essential nutrients like DEKA formulations and MCT oil. Clinicians acknowledged the absence of standardized nutritional guidelines for cholestatic liver disease and called for dedicated research to address absorption, bile acid metabolism, and growth outcomes.

Participants recommended developing disease-specific nutrition protocols, expanding patient education through cooking classes and hospital programs, and embedding dietitians within liver care teams.

Implication: Nutrition must be elevated from supportive care to a formal medical priority. Evidence-based guidelines and equitable access could dramatically improve growth, energy, and quality of life outcomes.

E. Mental Health

Mental health emerged as a universal and urgent issue across all disease states. Patients and caregivers described the lasting impact of medical trauma, isolation, and chronic stress related to their disease. Many caregivers reported depression or anxiety yet had never been referred to mental health services. Stakeholders agreed that mental health should be integrated into medical care beginning at diagnosis, with routine screenings, trauma-informed practices, and resources for siblings and parents.

Clinicians acknowledged a gap in training and the need for systemic change to normalize mental health discussions in liver clinics. Advocacy organizations were recognized as key partners in connecting families to support and education.

Implication: Treating emotional wellness as a core clinical outcome—not an optional service—can improve adherence, strengthen families, and enhance overall outcomes.

F. Comparative Effectiveness Research (CER)

The CER roundtable emphasized that effectiveness must be measured in terms of real-world evidence, not only lab results. Families urged researchers to include quality of life, mental health, and nutrition as core metrics in comparative studies. Participants called for unified patient registries across ALGS, PFIC, and BA to capture shared data and cross-disease learnings.

Misdiagnosis and delayed detection were recurring frustrations, underscoring the need for standardized screening and global collaboration. Families expressed strong interest in co-designing studies and contributing longitudinal data through registries and advocacy networks.

Implication: CER should evolve into a holistic, family-informed framework that measures what matters most—living well, not just living longer.

5. Cross-Cutting Themes

Across every roundtable discussion, a consistent message emerged: scientific progress alone cannot improve lives unless it is informed by lived experience. Families, clinicians, and researchers identified recurring barriers that cut across disease type and topic—revealing shared opportunities for collaboration and innovation.

A. Mental Health as a Foundation for Care

Emotional well-being was repeatedly identified as a prerequisite for successful treatment adherence, transition, and recovery. Mental health challenges—rooted in medical trauma, chronic uncertainty, and isolation—affect entire family systems. Integrating mental health services into standard liver care, beginning at diagnosis, was seen as essential rather than optional.

B. The Family as a Unit of Care

Participants agreed that rare liver disease affects far more than the patient. Siblings, parents, and even grandparents experience emotional, financial, and logistical strain. Families called for "whole-family care" models that include counseling, education, and peer support.

C. Continuity Across the Lifespan

As more individuals with ALGS, PFIC, and BA survive into adulthood, the lack of structured transition pathways has become a critical weakness. Early, gradual preparation for transition—paired with coordinated handoffs between pediatric and adult hepatology—was viewed as one of the most achievable and high-impact improvements.

D. Fairness and Access

Insurance denials, inconsistent access to IBATs and vitamins, and the absence of adult specialists in many regions leave patients vulnerable. Expanding assistance programs, telehealth options, and global drug availability were central priorities for every group.

E. Data that Reflects Daily Life

Families are already collecting meaningful data through videos, sleep tracking, and symptom logs. Participants urged researchers to formally integrate these patient-generated data into clinical studies and CER frameworks. Doing so would bridge the gap between quantitative and qualitative evidence and help define success in terms that matter to families.

F. Shared Learning Across Diseases

Although each condition presents unique challenges, participants recognized remarkable overlap in symptoms, treatment barriers, and social-emotional impacts. A unified approach—combining insights from ALGS, PFIC, and BA—can accelerate progress across the entire cholestatic liver disease spectrum.

6. Participant-Identified Priorities and Feasibility

The findings in this section reflect priorities and potential solutions identified by roundtable participants, along with their perspectives on feasibility and timing. These insights represent a stakeholder-informed foundation for ongoing cross-disease dialogue among ALGSA, PFIC Network, and BARE. They are not intended as formal recommendations, but as a starting point for future collaboration and roadmap development.

Short-Term (0–2 Years)

- Integrate mental health screenings and referral pathways into liver clinics.
- Launch pilot digital tools for tracking pruritus, sleep, and medication adherence.
- Publish interim nutritional guidance through patient advocacy group websites.
- Develop family resource packets for newly diagnosed patients.
- Increase awareness of existing financial assistance programs.

Medium-Term (2–5 Years)

- Establish joint pediatric–adult transition of care clinics and Centers of Excellence.
- Create cross-disease patient registries capturing mental health, nutrition, and quality-of-life data.
- Improve training for clinicians in trauma-informed care and communication.
- Conduct CER studies that compare integrated care models (medical + behavioral + nutritional).

Long-Term (5 + Years)

- Develop standardized national protocols for Transition of Care.
- Build precision-nutrition and mental-health modules into a digital management system.
- Enhance education, awareness, and advocacy efforts to promote newborn screening and early detection of cholestatic liver diseases.
- Embed “family voice” advisory panels into ongoing research consortia.

7. Conclusion

The 2025 Cholestatic Liver Disease Summit Research Roundtable Meetings brought together the collective voice of a community united by shared purpose and resilience. Notably, this marked the first time that the Alagille Syndrome, Progressive Familial Intrahepatic Cholestasis, and Biliary Atresia communities were convened together in a single setting to identify shared challenges and explore potential solutions across disease states. While each topic—pruritus, transplant, transition of care, nutrition, mental health, and comparative research—revealed its own distinct challenges, together they form a single narrative: that care must evolve alongside the people it serves.

Across all discussions, participants reinforced that the path forward is not defined solely by new therapies or technologies, but by the ability to listen, integrate, and act on what families experience every day. This White Paper reflects that commitment—documenting the realities of rare cholestatic liver disease, the ingenuity of caregivers, and the openness of clinicians and researchers striving for better outcomes.

The findings presented here, derived from participant-reported perspectives and thematic analysis, lay the groundwork for new collaborations, research priorities, and systemic improvements. By aligning science with lived experience—and by continuing to build connections across disease communities—the rare cholestatic liver disease field can drive measurable progress toward a future where every child, adult, and family receives equitable, holistic, and compassionate care.

8. Acknowledgments

The authors - Alagille Syndrome Alliance (ALGSA), PFIC Network (PFIC), and Biliary Atresia Research and Education, Inc. (BARE) - extend their deepest gratitude to all roundtable participants and supporters of the *2025 Cholestatic Liver Disease Summit*.

Thank you to the patients, caregivers, clinicians, researchers, and industry representatives who generously shared their expertise, perspectives, and lived experiences. Your honesty and collaboration have shaped a document that reflects not only the challenges of rare disease, but the strength of the community determined to overcome them.

Special thanks to each participant, facilitator, note taker, and advocacy leader for ensuring that every voice was heard and every insight captured. Together, we are redefining what progress means for those living with rare cholestatic liver diseases—measured not just in clinical milestones, but in the daily lives made better through shared understanding, innovation, and hope.

(Appendix A)

RESEARCH ROUNDTABLE WORKSHEET

Each participant should complete this worksheet during or immediately after your table discussion. Please complete one sheet for each of the sessions.

SESSION #: ____________ ***TABLE #:*** ___________

1. What was the primary topic of discussion at your table? *(Check all that apply)*
☐ Nutrition
☐ Mental Health & Wellbeing
☐ Transplant
☐ Patient-Centered & Comparative Effectiveness Research (CER)
☐ Transition of Care
☐ Pruritus/Itch
☐ Disease Specific

2. Which stakeholder group do you fall into? *(Check all that apply)*
☐ Patient
☐ Caregiver/Family Member - parent, sibling, grandparent *(please circle)*
☐ Clinician (e.g., hepatologist, GI, nurse)
☐ Scientist/Academic Researcher
☐ Industry Representative (e.g., pharma, biotech)
☐ Advocate/Nonprofit Leader
☐ Other: _________________________

3. What were the key points raised during the discussion? *(Open text)*

____

____

____

4. What ideas or possible solutions did your group come up with? *(Open text)*

____

____

____

5. Do you feel these ideas or solutions are achievable?

☐Yes, immediately ☐Yes, within 2 years ☐Yes, within 5 years ☐Yes, over 10 years
☐No

Please describe your answer: *(Short answer)*

____

6. What, if anything, surprised you during this discussion? *(Open text)*

____

____

7. What was your most valuable takeaway from this roundtable? *(Open text)*

____

____

8. What were your top two key learnings during this session? *(Open text)*

____

____

9 If you would you be interested in continuing the conversation with with us, please provide

Email: ____

10. Did you feel like your voice was heard at the table?
(Rate 1–5, where 1 = Not at all, 5 = Very well)

☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5
Please explain: *(Optional short answer)*

____

11. Do you have any other comments or reflections about your Research Roundtable experience? *(Open text)*