Advancing Drug Development. Improving Lives. Together.

October 7, 2025

RARE/ORPHAN AND PEDIATRIC DISEASE PROGRAMS

QUARTERLY NEWSLETTER

Welcome to the Rare/Orphan and Pediatric Disease Programs quarterly newsletter. In between major announcements, webinars and meetings, this communication serves to update you on the latest developments within C-Path's rare/orphan and pediatrics disease programs and the RDCA-DAP platform. None of C-Path's advancements are possible without the participation of our members, collaborators and data contributors. Thank you.

C-PATH GLOBAL IMPACT CONFERENCE

In September, C-Path successfully concluded its annual Global Impact Conference (CGIC) in Washington, D.C. This landmark event brought together industry leaders, regulatory agencies, academic experts, those with lived experience, and nonprofits, all dedicated to advancing drug development and regulatory science for rare diseases. Together, participants explored the theme, "Every Stage, Every Step: Transforming Lives Through Optimizing Drug Development" with a specific focus on pediatric, rare and orphan diseases, modernized randomized controlled trials, and the future of regulatory science. Overall, the outcomes from this event highlighted C-Path's ongoing commitment to lead initiatives that accelerate drug development processes and enhance outcomes globally, setting a strong foundation for future CGIC events.

As C-Path looks to the future, plans are already underway for the next Global Impact Conference, scheduled for the week of September 14, 2026.

Full session recordings will be published on C-Path's YouTube channel. To be notified when additional materials are available, be sure to subscribe to C-Path news at c-path.org/subscribe.

SPOTLIGHT ON INC DAY AND THE CPA-1 WORKSHOP

International Neonatal Consortium: Shaping the Future of Neonatal Drug Development

On September 8, INC convened its annual INC Day, bringing together clinicians, researchers, regulators, industry partners, and patient advocates to address some of the most pressing challenges in neonatal and pediatric drug development. The meeting emphasized cross-sector collaboration, global harmonization, and patient-centered innovation as essential drivers of progress.

Highlights from the Sessions:

- **Lab Values:** Showcased INC's new real-world data–driven modeling tool to address gaps in neonatal lab reference ranges, with commitments to expand data sources, publish a methodological framework, and engage regulators early.
- **Safety Reporting (NAESS 2.0):** Tackled fragmented adverse event frameworks by outlining plans for a next-generation neonatal severity scale, integration with EHRs, international harmonization, and structured training.
- **Neonatal Seizure Therapies:** Explored challenges such as early patient identification and EEG , with next steps focused on consensus building, expanding diagnostics, and piloting innovative trial designs.
- **Brain Injury Collaborative (NBIC):** Marked the official launch of the NBIC, prioritizing MRI/EEG biomarkers and committing to standardized data elements, a biomarker implementation guide, and stakeholders' multicenter pilot projects.
- **Future of Pediatric & Neonatal Development:** Reflected on regulatory openness and the importance of global harmonization, proposing multi-agency roundtables, biomarker advancement, and digital innovation pilots.

Impact:

By aligning scientific priorities with regulatory needs and patient perspectives, INC Day advanced concrete actions that will improve trial feasibility, accelerate biomarker qualification, and enhance safety reporting. These initiatives are paving the way for safer, more effective therapies for the most vulnerable patients—premature and critically ill newborns.

Shaping the Future of AATD: Patient Perspectives to Drive the Next Decade of Innovation in Alpha-1 Antitrypsin Deficiency

On September 8 — twenty years after the groundbreaking and original FDA PFDD meeting — over 100 patients, caregivers, patient advocacy groups, clinicians, scientists and industry professionals came together to hear about the disease, life burdens, and challenges for patients of all AATD genotypes, while brainstorming the future of therapeutic and clinical trial advances for alpha-1 antitrypsin deficiency.

The day-long workshop, **designed by C-Path's CPA-1 consortium in collaboration with the Alpha-1 Foundation**, convened in Washington, DC and virtually to update, fill gaps, and connect the dots between community and regulatory agency understanding of current patient diversity in AATD disease burden, tolerance of therapeutic innovations, and clinical trial risk. Workshop planning coincided with the release of an **ongoing patient/caregiver survey (CPA-1 Patient Experience Survey Update for iNnovation, or CAPESUN)**.

Highlights of the day included fireside chats moderated by CPA-1 member patient advocacy leaders on key topics, including challenges in diagnosis; health issues not currently identified as specifically AATD-related, such as gallstones, osteoporosis, vitamin deficiencies and connective tissue disorders; and goals of current therapies including preserving health and easier infusion options. In these panels, those with lived experience were partnered with clinicians to provide in-depth discussions. Not to waste a moment, particularly engaging was a lunchtime session "Clinician Conversation: One Big Idea," focused on how to improve clinical trials in AATD for accelerated regulatory decision-making.

The day was filled with important conversations around the logistical challenges of participating in trials, the willingness (or not) of patients to be in a placebo treatment group, and tolerance of the unknown for parents considering clinical trials for their children.

More learnings from the day and a survey will be shared throughout the coming months.

If you know of patients or caregivers living with alpha-1, please share this survey link.

RDCA-DAP UPDATES

The platform currently hosts data for over 43 disease areas, including the largest database for Friedreich's ataxia. For a full list of diseases and platform engagement to date, visit Additional RDCA-DAP Resources.

NORD CORNER

How Will a Changing Regulatory Landscape Impact Rare Disease? Learn More at the 2025 NORD Summit!

Gaining insight into evolving regulatory priorities is more important than ever — and the National Organization for Rare Disorders (NORD®) is bringing key players together for a dynamic conversation.

Join NORD in Washington, D.C., (or virtually) from **Oct. 19-21 for the 2025 NORD Rare Diseases & Orphan Products Breakthrough Summit** , high-impact gathering that puts you face-to-face with the decision makers who are shaping the next era of rare disease treatment and policy, including National Institutes of Health Director Jay Bhattacharya, M.D., Ph.D., and senior U.S. Food and Drug Administration officials and review staff. Register today!

WEBINAR SERIES 2025

Thursday, October 30, 12 PM ET | Register now for our upcoming webinar, 'Addressing the Lack of Standardized Laboratory Reference Ranges in Neonatal Trials: Pilot GUI Launch'

May 22: RDCA-DAP and NORD's IAMRARE Platform to Platform Federation: Lessons Learned

RDCA-DAP and NORD present an overview of their collaborative work enabling a direct connection of the RDCA-DAP to the NORD IAMRARE registry platform. Key highlights included lessons learned from the collaboration in data formatting and standardization and how these lessons helped enhance the RWD collected by IAMARE, as well as a robust discussion with concrete examples on how RWD is improved to make it more fit for use.

April 17: ACE Inhibitors as a Cardioprotective Treatment in Dystrophinopathies

Dr. Karim Wahbi discusses the use of ACE inhibitors as a cardioprotective treatment in dystrophinopathies, detailing their effectiveness in preventing heart failure and improving survival. The variety panelists, which included PPMD committee members Colin Werth and Pat Furlong, Parent Project Italy Founder Filippo Buccella, Lead Product Developer at Cumberland Pharmaceuticals Dr. Ines Macias-Perez, and Dr. Jonathan Soslow of Vanderbilt Pediatric Heart Institute, emphasized the importance of early treatment, ideally by the age of 10 years, and the call for better dissemination of these recommendations. The group highlighted the need for better understanding and care for Duchenne and Becker muscular dystrophy carriers, particularly mothers and sisters, and the importance of regular check-ins for carriers to address their overall well-being.

February 20: Vivli and RDCA-DAP: Data Platform Partnerships to promote Rare Disease

C-Path collaborator Julie Wood, Chief Operating Officer at Vivli, led this webinar, highlighting Vivli's mission as an independent nonprofit dedicated to global data sharing and analytics. She explored how Vivli and RDCA-DAP are working together to advance data sharing in rare diseases. Following her presentation, panelists from Vivli and C-Path engaged in a dynamic discussion on platform-to-platform collaboration and strategies to enhance data accessibility, concluding with a live Q&A. C-Path participants included Richard Liwski, Alexandre Bétourné, and Ramona Walls.

You can view all 2024 ROPD Webinars on demand here.

For more information about Rare/Orphan and Pediatric Disease Programs, visit: https://c-path.org/area-of-focus/rare-and-orphan-diseases/.

ANNOUNCEMENTS AND CONSORTIUM NEWS

September 25: International Ataxia Awareness Day: Advancing Hope Through Collaboration and Innovation

September 22: C-Path's Martinez Joins Help4HD Podcast in Special Guest Segment

September 18: The Critical Path Institute Podcast: Connecting the Dots in Pediatrics

September 16: EMPOWER & INSPIRE 4TH ANNUAL VIRTUAL LEIGH SYNDROME SYMPOSIUM

- Alexandre Bétourné presented a session titled, "Critical Path's Rare Mitochondrial Diseases Task Force" at the Empower & Inspire 4th Annual Virtual Leigh Syndrome Symposium on September 16, 2025. The presentation provided an overview of the Rare Mitochondrial Diseases Task Force mission, with an update on near term deliverables including the development of recommendations to enhance registry data collections and discussed long-term goals with cases examples of drug development tools that the group aims to generate.

September 7: World Duchenne Awareness Day: Celebrating 10 Years of D-RSC's Commitment to Serve the Duchenne Community

September 4: From Data to Hope: C-Path's Polycystic Kidney Disease Outcomes Consortium and the Power of Data Sharing

August 21: HDYO Joins C-Path's Huntington's Disease Consortium to Amplify Youth Voices in Drug Development

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Help support our mission. **MAKE A GIFT TODAY**

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FDA Acknowledgment