Advancing Drug Development. Improving Lives. Together.

July 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 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.

INTRODUCTION

As we welcome summer, the Rare/Orphan and Pediatric Disease Programs team has stayed active and engaged—joining conferences, speaking on expert panels, and connecting with the broader rare disease community. These interactions remain critical touchpoints for sharing knowledge, building partnerships, and reinforcing our commitment to improving outcomes for individuals and families affected by rare conditions.

Behind the scenes, our task force initiatives continue to make meaningful strides. These ongoing efforts are laying the groundwork for stronger collaborations and smarter, faster drug development strategies in specific disease areas.

We're also looking ahead to an exciting opportunity to connect in person. Join us for the 2025 **C-Path Global Impact Conference,** happening **September 9–11 in Washington, D.C**. The event will spotlight C-Path's expanding portfolio and showcase innovative work in rare diseases, neurology, pediatrics, and more.

We wish you a safe and enjoyable summer—and look forward to reconnecting with many of you this Fall.

SPOTLIGHT: ALS Awareness Month

As we reflect on ALS Awareness Month, it is a great opportunity to highlight the efforts C-Path has fostered under the collaborative network built through the ACT for ALS initiative.

Through the ACT for ALS initiative, C-Path and CP-RND have established partnerships spanning government organizations, industry, academic institutions, and non-profits to support drug development for ALS by bringing together collective expertise that is unmatched. Central to this effort is Patient Focused Drug Development, which ensures that people living with and impacted by ALS are integral to shaping research priorities and outcomes. This comprehensive network creates a foundation for addressing the complex challenges of ALS drug development through coordinated effort and shared resources.

Building on this patient-centered approach, one of the most significant achievements this year by CP-RND, has been addressing a critical gap in ALS research: ensuring clinical trials measure what actually matters to people living with the disease. Working closely with FDA guidance, the team conducted extensive research to compare what's meaningful to ALS patients with what is typically measured in clinical trials. The results revealed important gaps regarding measures to assess motor function, breathing, and speech—areas that profoundly impact daily life for people with ALS. Additional qualitative studies are being planned to address these gaps and better understand how individual measures can be improved. Parallel to this, efforts were initiated to understand the variation in collecting and interpreting data from various measures such as the ALSFRS-R, respiratory function assessments, and digital health technologies used in measuring gross motor function.

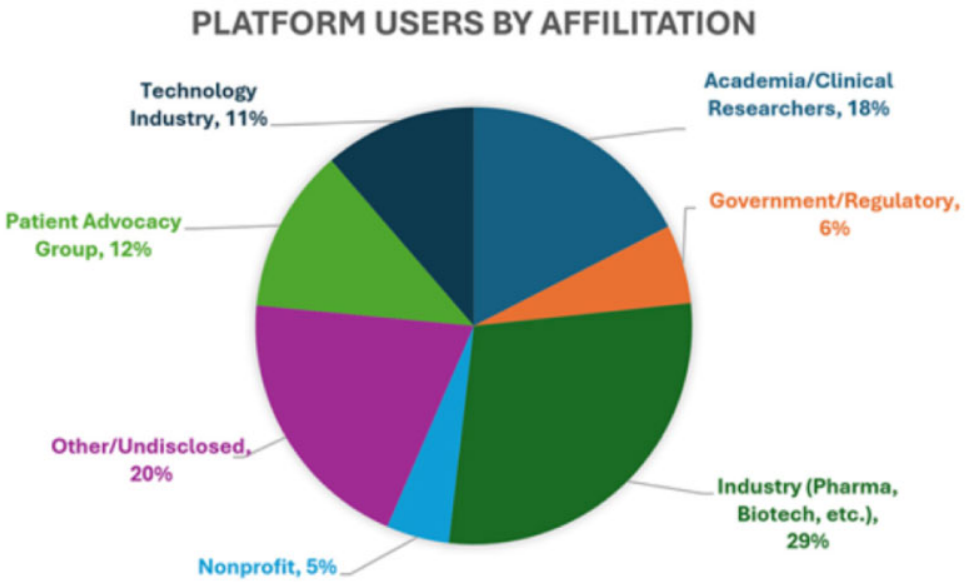
Complementing these measurement improvements, a major effort is underway to bring together high-priority ALS datasets into a centralized ALS Knowledge Portal. This year alone, C-Path successfully transferred and began curation of data from 10 different sources—doubling the initial goal. This makes valuable research information more accessible to the entire ALS community while providing the data foundation needed for standardization efforts. This collaborative approach represents more than just scientific progress—it's a testament to what's possible when the entire ALS community works together toward a common goal. The foundation being built today through these partnerships will support accelerated drug development for years to come, bringing hope to families affected by ALS and other rare neurodegenerative diseases.

RDCA-DAP UPDATES

The platform currently hosts data for over 43 disease areas, including the largest database for Friedriech’s ataxia. For a full list of diseases, visit Additional RDCA-DAP Resources.

Engagement to date:

- 760 approved platform requests
- 39 active workspaces for external users/research



NORD CORNER

Join the Rare Disease Community at the NORD Breakthrough Summit

Get ready for the rare disease community's premier event: the NORD Rare Diseases & Orphan Products Breakthrough Summit®. Join us Oct. 20-21, 2025, in Washington, D.C., as science, advocacy, and industry converge to shape the future of rare disease research, treatment, and policy. With more than 900 attendees, 90 expert speakers, 70 research poster presenters, and over 60 sponsors and exhibitors, this year's Summit will offer:

- A comprehensive agenda featuring the latest advancements in rare disease research and development
- Candid conversations with senior leaders from the NIH and FDA
- One-on-one networking with patients, caregivers, researchers, and industry professionals
- Access to all sessions, including our flagship CEO Perspectives Panel
- Entry to the Welcome Reception and Poster Hall
- Digital access to session recordings and materials

Visit https://nordsummit.org/ to learn more!

ANNOUNCEMENTS AND CONSORTIA NEWS

June 29: Critical Path Institute and Citizen Health Partner to Accelerate Drug Development for Neuromuscular Disorders

June 27: C-Path Welcomes Avidity Biosciences as Newest Member to Duchenne Consortium

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

June 20: C-Path Launches Global Collaborative to Advance Drug Development for Neonatal Brain Injury

June 2: Lysosomal Storage Disease Data Sharing Workshop, Webinar Series Session #4: Case Studies #3 – Data Sharing Collaboratives

May 15: From Awareness to Action: How C-Path's HD-RSC Is Transforming the Future of Huntington's Disease

May 8: Critical Path Institute Celebrates 20 Years of Advancing Drug Development through Innovation and Collaboration

May 6: C-Path Announces First Data Transfer from Takeda to Accelerate Alpha-1 Innovation

April 28: Lysosomal Storage Disease Data Sharing Webinar Series – Session #3: Retroactive Data Sharing

April 23: C-Path Recognizes HIE Awareness Month, Calls for Global Collaboration to Accelerate Neonatal Drug Development

April 14: C-Path Honors Memory, Legacy of Beloved CP-RND Member and ALS Patient Advocate Layne Oliff

March 27: C-Path Announces Paul E. Edmeier as New Chief Financial Officer

March 24: Lysosomal Storage Disease Data Sharing Workshop, Webinar Series Session #2: Case Studies #1 – Proactive Data Sharing

WEBINAR SERIES 2025

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.p

***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/

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

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FDA Acknowledgment