



Advancing Drug Development. Improving Lives. Together.

September 30, 2026

C-Path 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 ROPD programs, as well as the Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP®). None of these advancements are possible without the participation of our members, collaborators, and data contributors. Thank you.

Earlier this month, C-Path successfully concluded its annual C-Path Global Impact Conference (CGIC) in Washington, D.C., bringing together leaders from industry, regulatory agencies, academia, patient communities, and nonprofit organizations to advance drug development and regulatory science.

CGIC 2026 highlighted the power of collaboration to address some of the most pressing challenges in drug development, with discussions spanning responsible data sharing, patient-informed drug development, biomarkers, emerging technologies, and innovative approaches for rare and individualized therapies. Throughout the conference, participants emphasized the importance of making high-quality data more reusable, incorporating patient perspectives throughout the drug development process, and advancing scalable, regulator-aligned solutions through public-private partnerships.



While CGIC 2026 has concluded, the collaborative work continues. C-Path will build on the ideas and connections generated during the conference to translate pre-competitive frameworks and partnerships into actionable progress across its programs and initiatives. On-demand session recordings and post-conference materials will be available in the coming weeks on the C-Path YouTube Channel [here](#).

Stay up to date on CGIC and other C-Path initiatives by subscribing to C-Path news [here](#).

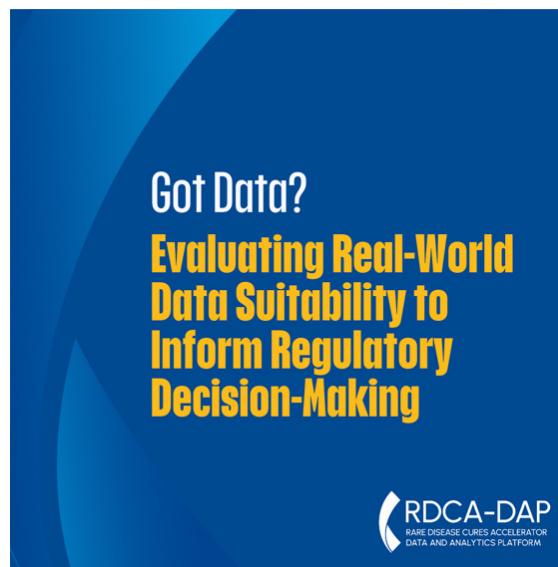
SPOTLIGHT:

'Got Data? Evaluating real-world data suitability to inform regulatory decision-making' RDCA-DAP Workshop

On Monday Sept 14, the RDCA-DAP and Data Collaboration Center teams hosted the first in-person workshop on real-world data (RWD) as a pre-CGIC workshop in Washington, DC. RWD is increasingly used to inform regulatory decision-making, particularly in rare diseases. However, variability in data sources and inconsistent data quality often limit the utility of RWD for regulatory decision-making.

A central challenge is determining whether a given RWD source is fit-for-purpose before substantial analytical investment. This workshop addressed this challenge by providing an interactive, use-case driven exercise focusing on the evaluation of RWD rather than downstream analyses. The session opened with a presentation from Dr. Wei Hua, Director of Division of Epidemiology with Center for Drug Evaluation and Research at FDA. Dr. Wei set the stage by providing a regulatory perspective on the design of studies using RWD, highlighting key considerations for generating robust and reproducible real-world evidence (RWE). She introduced insights from the "HARmonized Protocol Template to Enhance Reproducibility" (HARPER), a template that provides a set of core recommendations for clear and reproducible RWE study protocols.

The core of the workshop guided attendees through assessments of RWD across four core dimensions: (1) relevance (availability of required data elements), (2) observability (continuity of care and longitudinal follow-up), (3) structural comparability (ability to define populations and comparators), and (4) reliability (data capture, consistency, missingness, and provenance), with explicit consideration of the underlying data generating process and its implications. Participants used a custom app to evaluate information and aggregated results for several fictitious rare diseases datasets from different RWD sources (including electronic health records and patient registries). A team of C-Path RWD experts guided the participants through this evaluation and helped them determine the fitness for use of the datasets against drug development use cases (historical controls, biomarker development, etc.), highlighting the importance of a collaborative approach in these assessments.



"Fit for purpose starts with purpose" was a recurring theme as participants explored how the same real-world data source can be well suited to one research question—and fall short for another. Building on the energy and insights from this first workshop, RDCA-DAP is already looking ahead to the next opportunity to bring stakeholders together: a second RWD workshop to be offered in conjunction with CGIC 2027 in Washington, DC.

RDCA-DAP UPDATES



On Wednesday, September 16, Dr. Alexandre Bétourné, Executive Director of RDCA-DAP and Dr. Mingyuan Wang, Quantitative Medicine Scientist at C-Path, hosted a breakout session at CGIC entitled "Leveraging Rare Disease Data to Inform Drug Development Tools for Rare Neurodegenerative Diseases."

Dr. Wang presented an overview of RDCA-DAP latest results for two rare neurodegenerative diseases, including the development of Clinical Trial Simulation tool for Friedreich's ataxia (FA), and evaluations of different variations of the progressive supranuclear palsy (PSP) Rating scales' ability

to predict patient survival and track rate of disease progression. The session continued with a panel discussion that brought together rare neurodegenerative experts: Dr. Mohammed Hamdani Vice President, statistics and quantitative sciences at Larimar Therapeutics, Dr. Mark Gordon, senior medical director in clinical development - neuroscience at Arvinas and Zoe Cooper Capella, clinical research coordinator at MGH neurology.

The group discussed current unmet needs in FA, PSP and multiple system atrophy, highlighting the complexity of administering the available clinical scales to patients, their limitations, and the critical need for better endpoints to track disease progression in rare neurodegenerative disorders. Dr. Hamdani also insisted on the usefulness of MIDD and Clinical Trial Simulation Tools such as the FA tool in development by RDCA-DAP.

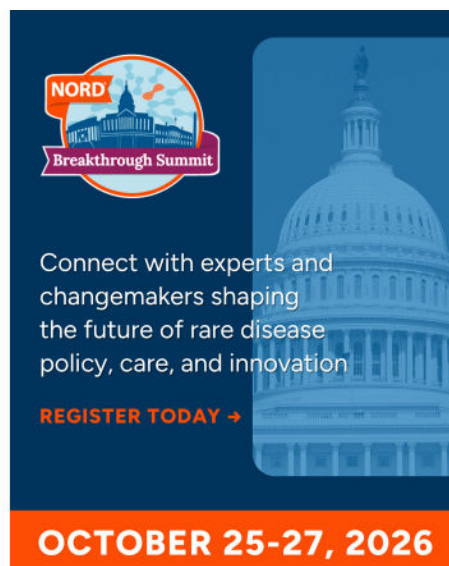
Through the session and a lively Q&A with the audiences, the critical importance of data sharing was renewed. In particular, the discussion pointed to the lack of data sharing for two highly relevant biomarkers for FA (frataxin) and for PSP (NfL), and positioned RDCA-DAP as an ideal platform to host an integrated database that would make such data available to industry, to the ultimate benefit of rare neurodegenerative disease patients.

NORD CORNER

Last Chance to Register for the NORD Breakthrough Summit!

At a transformational time for rare disease research, treatment development, and policy, the [NORD Rare Disease and Orphan Products Breakthrough Summit](#) will bring together the nation's leading patient advocates, clinical researchers, biopharma and medtech executives, investors, and government health officials to explore the opportunities and challenges shaping what's next.

Explore platform-based solutions for scaling treatment development beyond one disease at a time, the latest state and federal policy debates, novel uses for artificial intelligence (AI), and the future of access and affordability for today's breakthrough therapies. Featuring many of the nation's top health officials, experts from the NORD® Rare Disease Centers of Excellence Network, and leaders of hundreds of rare disease patient advocacy groups.

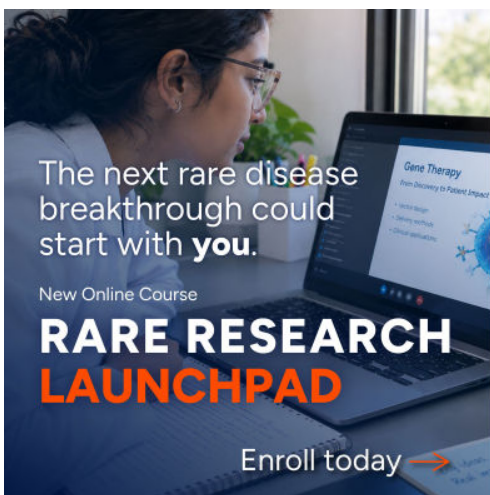


[Explore the agenda](#) and commit to joining us by registering for the NORD Breakthrough Summit today.

Introducing NORD's Rare Research Launchpad

Students and early-career trainees may get little exposure to rare disease research or may not realize how their interests and skills could contribute to the field. NORD is helping create that entry point for students while focusing on intentional workforce development with [Rare Research Launchpad: A Career Exploration Series in Rare Disease Research](#) — a new, free, self-paced course and accompanying Research Exploration Toolkit.

Participants explore cystic fibrosis and Krabbe disease as two very different examples of rare disease research and hear directly from families living with them. They also learn how discoveries move from the lab toward patient care, and about the many paths that can lead to a career in rare disease research, from established researchers at the NORD® Rare Disease Centers of Excellence. The accompanying Research Exploration Toolkit turns that



exploration into action with practical guidance for building skills, finding research and training opportunities, and connecting with mentors.

Help spread the word about this resource to the prospective researchers in your network, whether they are undergrads, graduates, med students, or early-career professionals. The course page includes promotional materials you can use to easily share it with others. Explore the Rare Research Launchpad [here](#).

Rare Research Launchpad was developed with support from the Rosenau Family Research

Foundation.

Blog: Advancing Hope in Limb Girdle Muscular Dystrophies: Collaboration, Data, and Innovation During LGMDs Awareness Month

Each September, Limb Girdle Muscular Dystrophies (LGMDs) Awareness Month shines a light on a devastating neuromuscular disease that remains widely underrecognized despite its profound impact on patients, families, and caregivers. LGMDs are a group of rare, inherited genetic disorders characterized by progressive weakness of the shoulder and hip girdle muscles due to loss of muscle fibers. Over time, people living with LGMD often experience increased difficulties with mobility and daily function, while facing a significant lack of effective treatment options.

For the LGMD community, September serves as a vital call to action to accelerate research, improve clinical understanding, and advance the development of therapies that can meaningfully change patients' lives. At Critical Path Institute® (C-Path), that mission is central to our work, and we take pride in it every day.

In September 2024, C-Path launched the Limb Girdle Muscular Dystrophies Task Force through its Rare Disease Cures Accelerator–Data and Analytics Platform (RDCA-DAP®). The initiative was created to address the urgent need for better tools, stronger collaboration, and more efficient drug development pathways for LGMDs.

Read the full blog [here](#).

WEBINAR SERIES 2026 | ON DEMAND

April 9: [Hope on the Horizon for ADTKD Patients](#)

This webinar discusses autosomal dominant tubulointerstitial kidney disease (ADTKD), the second most common genetic kidney disease after PKD, affecting an estimated 75,000–100,000 people in the U.S. and up to two million worldwide. Discover how physician-scientists are advancing toward the first-ever treatments for UMOD and MUC1 — the gene variants responsible for most ADTKD cases — and why a clinical trial may be on the horizon. Learn how the Rare Kidney Disease Foundation is empowering patients, families,

and advocates through education, community engagement, and advocacy efforts aimed at ending the impact of ADTKD on future generations.

March 19: [Meeting Patients Where They Are: Validating Remote Digital Tools for Multi-Indication NMD Research](#)

In this session, Dr. Tina Duong explores the scientific, clinical, and regulatory considerations involved in validating video-based assessments and other digital tools across multiple NMD indications. Through both presentation and panel discussion, attendees gained valuable insights into how these approaches can enhance accessibility for patients regardless of location, reduce the burden of participation in clinical trials, generate reliable, regulatory grade data, ensure innovation remains grounded in patient-centered research, and more.

****You can view all 2025 ROPD Webinars on demand [here](#).***

ANNOUNCEMENTS AND ADDITIONAL RESOURCES

September 3: [Advancing Neuromuscular Drug Development Through Patient-Centered Collaboration and Data Integration](#).

August 24: [Building a Stronger Path to Therapies for Hereditary Spastic Paraplegia and Primary Lateral Sclerosis](#).

August 6: [C-Path's Duchenne Consortium Awarded a \\$1.9 Million DoD Grant to Advance Understanding of Cardiomyopathy in Duchenne Muscular Dystrophy](#).

July 21: [Across Healthcare Matrix Partners with C-Path to Expand Rare Disease Data Platform Through Direct Integration](#).

July 9: [Wendy Ruyle Learned That Groundbreaking Science Alone Is Not Enough](#).

June 5: [Public Private Partnerships for Rare Neurodegenerative Disease Webinar Recording Available](#).

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.

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