RARE AND ORPHAN DISEASE PROGRAMS
CRITICAL PATH INSTITUTE

RDCA-DAP®
Rare Disease Cures Accelerator
Data and Analytics Platform

Advancing Drug Development. Improving Lives. **Together.**

December 18, 2024

Rare and Orphan Disease Programs - Quarterly Newsletter

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

Introduction

As the holiday season approaches, we want to take a moment to express our heartfelt gratitude for being part of our community. Your support and engagement have made this year truly remarkable.

Let's celebrate the memorable moments and achievements of the past year, reflect on our journey, and look forward to the exciting opportunities ahead.

Thank you for being with us every step of the way. We can't wait to reconnect in the coming year.

Wishing you a joyful holiday season and a wonderful New Year!

RDCA-DAP Updates

The platform currently contains data for over 41 different rare diseases. More data will be accessible throughout 2024 and into 2025 as outreach efforts continue.

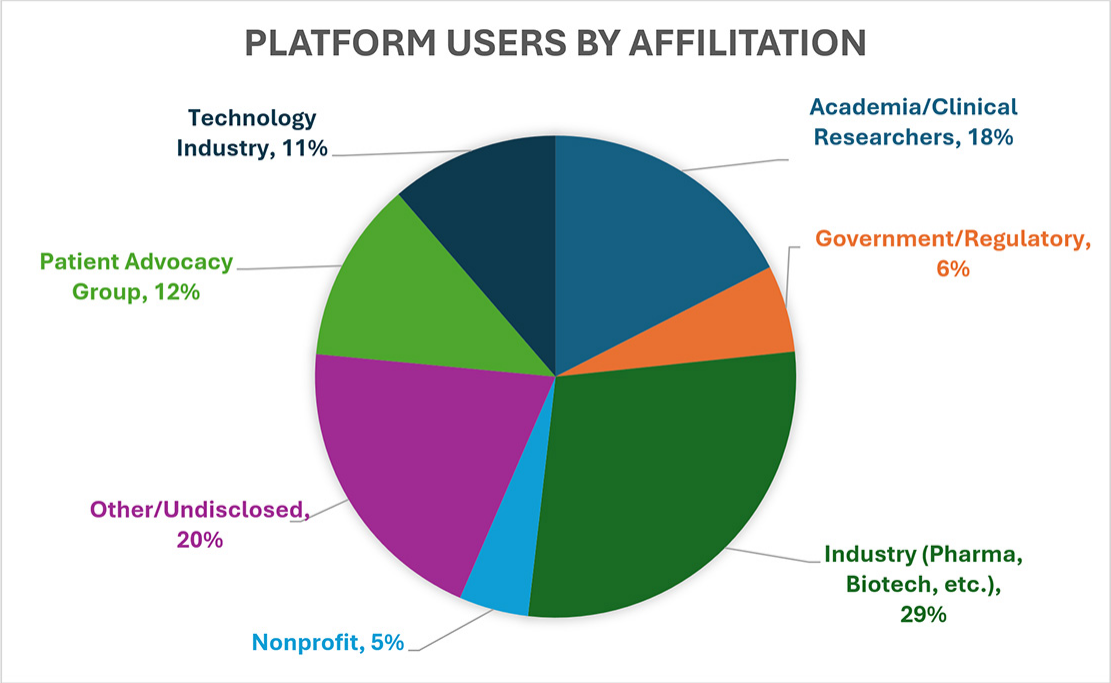
RDCA-DAP data includes:

Angelman Syndrome*	Mitochondrial Disease*
CACNA1*	Mucopolysaccharidoses*
Charcot-Marie-Tooth disease*	Myhre Syndrome
Choroideremia	Myotubular or centronuclear myopathy*
COL6-related dystrophies*	Necrotizing Enterocolitis*
Congenital Hyperinsulinism*	Niemann-Pick Disease
Cystic Fibrosis	Ocular Melanoma*
Desmoid Tumor*	Pemphigus & Pemphigoid*
Duchenne Muscular Dystrophy*	Phenylketonuria*
Facioscapulohumeral Muscular Dystrophy*	Polycystic Kidney Disease*
Fibrous Dysplasia Disorders*	Prader-Willi Syndrome*
Friedreich's Ataxia*	Progressive Supranuclear Palsy*
GNE Myopathy*	Rare Epilepsies*
hnRNPH2-related Disorders*	Ryanodine Receptor-1 Related Myopathies*
Kidney Transplant	SCN2A-related Disorders
KIF1A Associated Neurological Disorder (KAND)*	Spinal Muscle Atrophy with Respiratory Distress*
Kleefstra Syndrome*	Spinocerebellar ataxias type 1, 2, 3, 6 ,7, 8 & 10
Leigh Syndrome and other mitochondrial disorders*	Sturge-Weber Syndrome*
Lennox-Gastaut Syndrome*	Tuberous Sclerosis*
Limb-Girdle Muscular Dystrophy	VCP Disease

*Indicates disease with datasets that are currently discoverable on the platform

Engagement to date:

- 630 approved platform requests
- 42 approved workspaces for external users/research



Spotlight: Please Join us in Welcoming Noelle Burkhart to the team.

Please join us in welcoming Noelle Burkhart to C-Path as Project Coordinator, RDCA-DAP. Noelle joined us on December 2, 2024.

Noelle is a local Tucsonan and a graduate of Pima Community College with an AA in Liberal Studies. She has a background of working in various aspects of the biotech industry including pathogen testing and molecular diagnostic research. Her motivation to make a positive impact and passion for supporting others has driven her enthusiasm for healthcare.

NORD Corner

NORD®
National Organization for Rare Disorders

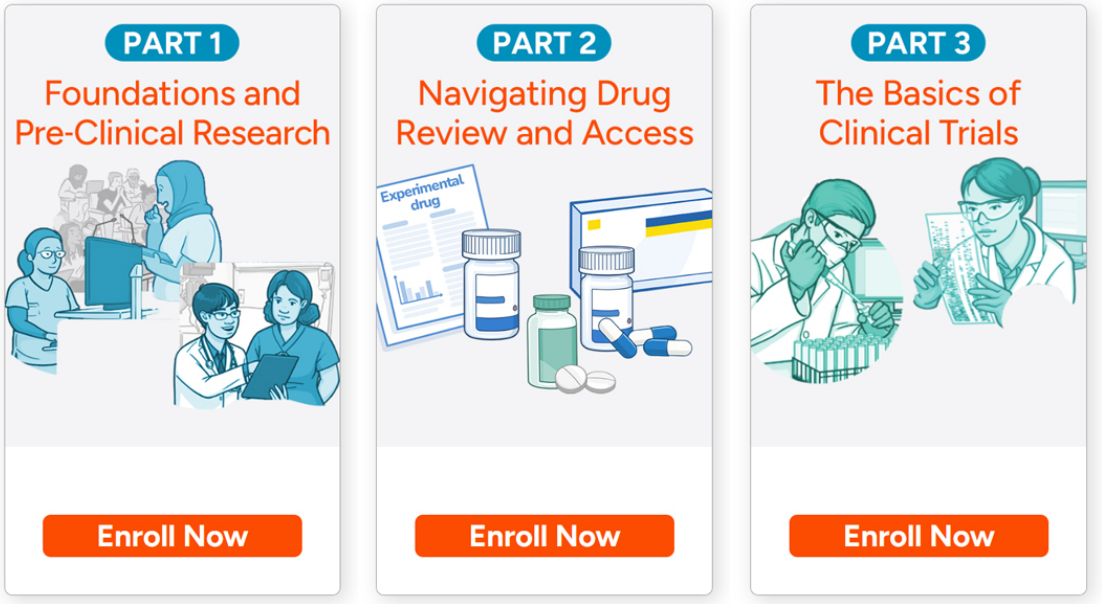
Help Develop New Therapies for Your Rare Disease

Living with a rare disease without an available treatment can be daunting, but you have the power to help with the discovery and development of new treatments. The National Organization for Rare Disorders (NORD), in collaboration with the FDA and C-Path, has developed the educational course, "Rare Disease Drug Development: What Patients and

Advocates Need to Know," to empower patients and caregivers with the knowledge needed to actively participate in each step of the research and FDA drug review process.

Want to share the news about this free education course with your patient community? Scroll to the bottom of the page for a downloadable toolkit that makes sharing this news quick and easy.

Click here for more information.



Announcements and Consortium News

October 16 - Sanford Health Rare Disease Data Registry and C-Path's RDCA-DAP Enter into a Master Data Contribution Agreement to Aggregate Rare Disease Data

September 30 - C-Path to Lead Comprehensive Task Force Focused on Accelerating Drug Development for Limb-Girdle Muscular Dystrophies

September 19 - C-Path's PKD Outcomes Consortium Receives BAA Award for Project to Advance Drug Development Tools for Autosomal Dominant Tubulointerstitial Kidney Disease

September 12 - C-Path's Inaugural Global Impact Conference Charts the Future of Drug Development

September 5 - Cystic Fibrosis Therapeutics Development Network Contributes New Data Sets to C-Path's RDCA-DAP, Advancing CF Research and Treatment Strategies

August 22 - C-Path Awarded $1.5 Million Grant from Ara Parseghian Medical Research Fund at Notre Dame to Enhance Collaboration in Niemann-Pick Disease Research and Drug Development

August 20 - C-Path Receives Data Transfer from The Champ Foundation Registry

August 20 - C-Path's RDCA-DAP Announces Strategic Partnership with Vivli to Enhance Clinical Trials Data Sharing for Rare Diseases

August 6 - C-Path Welcomes New Advisory Members to Alpha-1 Antitrypsin Deficiency Consortium

July 23 - C-Path's Lysosomal Disease Consortium Welcomes Chiesi Group as its Newest Member

***For previous announcements from 2024 please visit our website.**

Webinar Series 2024

All 2024 ROD Webinars are available now!

December 12: The Power of Data Sharing and Collaboration Across LGMDs in a Pre-Competitive, Neutral Environment

This webinar featured **Dr. Volker Straub**, a renowned neuromuscular genetics expert and Director of the John Walton Muscular Dystrophy Research Centre at Newcastle University. Dr. Straub provided an overview of Limb-Girdle muscular dystrophies (LGMDs) and highlighted critical unmet needs in the field. He was joined by an exceptional panel of experts: **Dr. Doug Sproule** (ML Bio Solutions), **Dr. Jennifer Levy** (Cure Calpain 3), **Kathryn Bryant Knudson** (Speak Foundation), **Dr. Sophie Olivier** (Atamyo), and **Cécile Ollivier** (C-Path).

November 14: Advanced In Vitro Modelling of Neuromuscular Diseases and Therapeutics

Led by **Dr. Francesco Saverio Tedesco** of the MAGIC project, this webinar explored innovative, multidisciplinary approaches in disease modeling, drug testing, and tissue engineering using advanced in vitro systems. Dr. Tedesco shared cutting-edge examples, including the development of advanced muscle models for studying muscular dystrophies and designing targeted therapies for neuromuscular diseases. Attendees gained insight into how multi-stakeholder, patient-centered collaborations are driving progress in this rapidly evolving field.

October 29: New Findings on Inequities in Rare Diseases – How they impact Research and Clinical Trial Processes

This webinar focused on a groundbreaking national survey, *Inequities in the Rare Disease Community: The Voices of Diverse Patients and Caregivers*. The research, involving over 2,800 participants, illuminated barriers faced by underrepresented patients with rare diseases. Key findings revealed significant emotional and logistical burdens, including 50% of respondents feeling overwhelmed by life, care, or caregiving responsibilities. These insights emphasize how inequities influence both healthcare access and research protocols.

July 18: The Influential Role of Patient Advocacy Groups in Registry Data Efforts

This session, hosted by the Mitochondrial and Inherited Metabolic Disease Task Force, highlighted how patient advocacy groups are pivotal in driving registry data efforts for mitochondrial and inherited metabolic diseases. The webinar emphasized the importance of centering patient experiences and outcomes in research and regulatory decision-making.

June 20: Hidden in Plain Sight: Autosomal Dominant Tubulointerstitial Kidney Disease (ADTKD)

This presentation covered the genetics, biology, and mechanisms of ADTKD, along with a description of the Wake Forest registry and the unmet drug development needs for this rare condition.

May 23: The Use and Development of Digital Health Technologies (DHTs) with Patient Advocacy Groups

Led by **Dr. Laurent Servais** of Oxford University, this webinar provided an overview of **Actimyo** and learnings from its use across rare disorders with varying levels of complexity and readiness. Leaders from patient advocacy groups shared their experiences with Actimyo's application in assessing individuals with neuromuscular disorders (e.g., Duchenne muscular dystrophy) and non-NMDs (e.g., Angelman syndrome and CTNNB1).

March 14: Improving Data Collection for Rare Epilepsies – Case Example from the TSC Natural History Database

This webinar discussed the need for better standardization in data collection for rare epilepsies. It highlighted case examples from the TSC Natural History Database to illustrate how improved practices can enhance research and care.

February 15: Understanding Disease Progression Models – What Are They, Why Are They Useful, and How Are They Applied?

This high-level overview explored how disease progression modeling combines statistics, disease knowledge, and data to inform predictions about disease course in populations and subpopulations. The session broke down the basics of these models and their applications in model-informed drug development.

***You can now view all 2023 RDCA-DAP Webinars on demand here.**

For more information about Rare and Orphan Diseases, click here.

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FDA Acknowledgment