RDCA-DAP®

RARE DISEASE CURES ACCELERATOR

DATA AND ANALYTICS PLATFORM

October 3, 2023

RDCA-DAP® Quarterly Newsletter

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

Introduction

The RDCA-DAP team has had an impactful summer, integrating new disease types into the Platform, welcoming a scientific director, continuing task force launch efforts, and concluding with participation in the inaugural Rare and Orphan Disease Conference presented by Critical Path Institute.

RARE AND ORPHAN DISEASE PROGRAMS

The Rare and Orphan Disease Conference held September 11 - 13, 2023, brought together over 200 leading experts, pharmaceutical industry leaders, regulatory agencies, and patient advocates to explore the latest advancements in rare disease research and drug development. The conference featured a packed agenda of informative sessions, showcases, and networking opportunities. RDCA-DAP was a highlight throughout the conference as a pivotal connecting component between all rare and orphan activities within C-Path, with sessions demonstrating the power of "FAIR" data and ontology applications in rare epilepsies and Friedreich's ataxia (FA), task force exploration in rare mitochondrial disorders and case study examples supporting patient advocacy groups' research objectives.

The conference's main sessions can be accessed on C-Path's YouTube channel here, C-Path 2023 Rare & Orphan Disease Conference. Read the post-event press release, here.

Data Developments

The platform continues to expand since going live September 2021, containing data for over 32 different rare diseases. More data will be accessible throughout 2023 and into 2024 as outreach efforts continue.

RDCA-DAP currently contains data for the following diseases:

Angelman Syndrome*	Mitochondrial Disease
CACNA1*	Mucopolysaccharidoses (MPS)

Cerebellar Ataxia	Myotubular or centronuclear myopathy*
COL6-related dystrophies*	Necrotizing Enterocolitis*
Congenital Hyperinsulinism*	Ocular Melanoma
Desmoid Tumor*	Pemphigus & Pemphigoid*
Duchenne Muscular Dystrophy*	Phenylketonuria (PKU)*
Facioscapulohumeral muscular dystrophy (FSHD)*	Polycystic Kidney Disease
Fibrous Dysplasia disorders*	Prader-Willi Syndrome*
Friedreich's Ataxia*	Progressive Supranuclear Palsy*
GNE Myopathy*	Rare Epilepsies*
hnRNP related disorders*	Ryanodine Receptor-1 (RYR-1) Related Myopathies*
Kidney Transplant	Spinal Muscle Atrophy with Respiratory Distress*
KIF1A Associated Neurological Disorder*	Spinocerebellar ataxias type 1, 2, 3 & 6
Leigh Syndrome and other mitochondrial disorders*	Sturge-Weber Syndrome
Lennox-Gastaut Syndrome*	Tuberous Sclerosis*

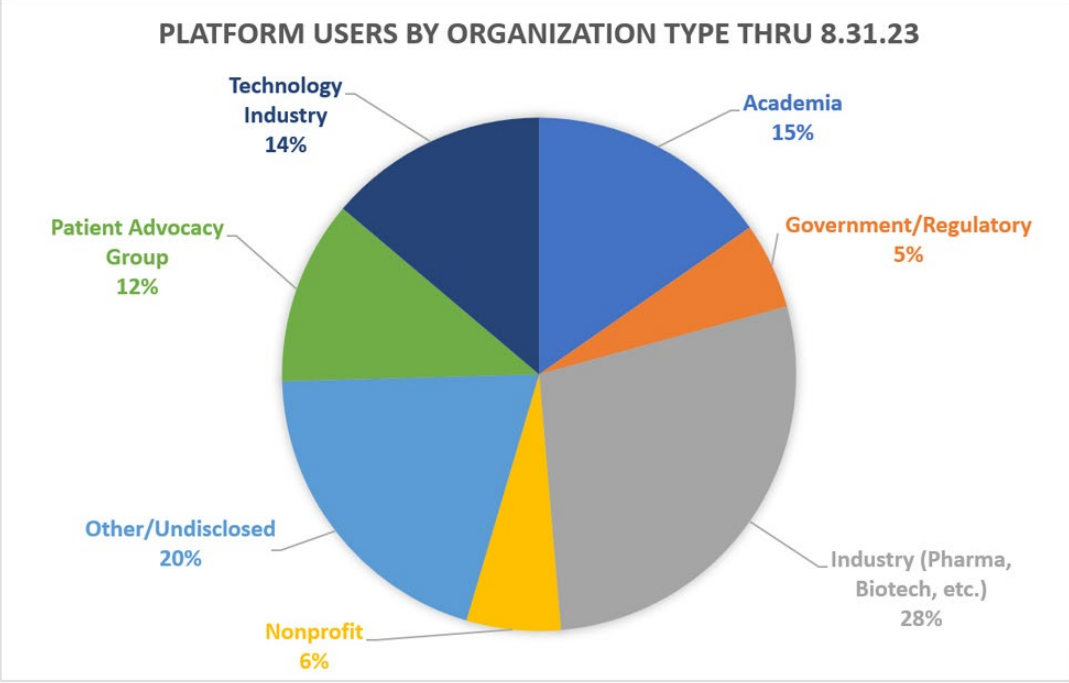
*Indicates disease with datasets that are currently discoverable on the platform

Interested in contributing data to RDCA-DAP? Fill out this form to be contacted by a team member.

Platform Use

Engagement to date:

- 405 approved platform requests
- 24 approved workspaces for external users/research



Spotlight: Heidi Grabenstatter Scientific Director, RDCA-DAP

In August, C-Path welcomed Heidi Grabenstatter, Ph.D., M.S. to the RDCA-DAP team as its Scientific Director. In her first six months at C-Path, Heidi will work primarily on advancing data aggregation and solutions development in rare diseases new to RDCA-DAP, as well as driving work in rare dystrophies in conjunction with the Duchenne Regulatory Science Consortium. Connect with Heidi here.

She has previously served as the Science Director of the International Foundation for CDKL5 Research (IFCR) managing the research portfolio for the organization. Heidi worked closely with IFCR's Board of Directors, MSAB, Centers of Excellence, and International CDKL5 Clinical Research Network (ICCRN) to ensure all projects were scientifically vetted and met patients' needs. Her research background is in molecular and physiological mechanisms of epileptogenesis and rationale therapy development. Her postdoctoral appointments were in adult and pediatric neurology departments at the University of Wisconsin School of Medicine and Children's Hospital Colorado, respectively. She later led an independent lab at University of Colorado, Boulder. As an epilepsy patient herself, Heidi promotes patient-driven research and caregiver engagement in the development of new tools that impact treatment development. Her commitment to rare disease patient advocacy is strengthened by her personal understanding of the extreme challenges that people with epilepsy face and experience as a caregiver to her mother and sister, both of whom passed from rare cancer diagnoses. The combination of her passion, her academic experience, and her patient perspective make Dr. Grabenstatter uniquely positioned to act as a conduit between patients and caregivers with unmet needs, industry, and regulatory communities to foster innovative research solutions.

NORD Corner

You are invited to attend the NORD Rare Diseases and Orphan Products Breakthrough Summit taking place on October 16-17 in Washington, D.C. This annual gathering brings together hundreds of rare disease stakeholders to share the latest updates on drug development, research, patient engagement, public policy, market accessibility of orphan products, and more.

We are pleased to announce that the U.S. Food and Drug Administration (FDA) Commissioner, Robert Califf, M.D., will be joining us at 8 a.m. ET on Tuesday, October 17, at the 2023 NORD Breakthrough Summit. Dr. Califf was confirmed as the 25th Commissioner of Food and Drugs, and is a nationally recognized expert in cardiovascular medicine, health outcomes research, health care quality, and clinical research. He is a leader in the growing field of translational research, which is key to ensuring that advances in science translate into medical care. Don't miss the opportunity to hear from Dr. Califf and many other experts! Register for the NORD Breakthrough Summit here.

NORD is also pleased to announce a request for applications for the implementation of two new patient registries on the IAMRARE platform, with funding made available through

RDCA-DAP. Full details and application available at: https://rarediseases.org/iamrare-2024-rfp.

Announcements

October 3, 2023: C-Path Rare and Orphan Disease Conference Highlights Unprecedented Collaborations and Breakthroughs in Rare Disease Research

August 15, 2023: NORDs' IAMRARE Registry Contributes Two New Datasets to C-Path's RDCA-DAP, Boosting Rare Disease Research and Therapeutic Advancement

July 19, 2023: The Foundation for Angelman Syndrome Therapeutics to Contribute Data to C-Path's RDCA-DAP, Accelerating Neurological Disease Research and Therapeutic Development

June 7, 2023: Newcastle University's John Walton Muscular Dystrophy Research Centre Contributes Data to C-Path's RDCA-DAP

June 2, 2023: New collaboration between Monarch Initiative and Critical Path Institute: Working towards improving drug discovery for rare diseases

May 31, 2023: CACNA1A Foundation to Contribute Data to C-Path's RDCA-DAP, Accelerating Neurological Disease Research and Therapeutic Development

February 28, 2023: Drug Repurposing Provides Big Impact for Patients

February 17, 2023: C-Path Stands with Bruce Willis, Neurodegenerative Disease Community After FTD Diagnosis

January 25, 2023: C-Path Receives Letter of Support from EMA on Duchenne Muscular Dystrophy Clinical Trial Simulation Platform

Upcoming and Past Events

Webinar Series 2023

Final RDCA-DAP Webinar of 2023 will be held November 16, noon ET: TBD

On Demand

August 10: Teaching Patients and Caregivers: The Role of Education in Research and Data Sharing

This webinar focused on the role of foundational education in research concepts, such as data literacy and the research process, in creating a culture of research participation and

data sharing in the rare disease space. Intended for patient advocacy groups and researchers who work with rare disease patients, the webinar provided knowledge and resources to help patients and caregivers understand and engage in the research process.

May 18: GUIDs and De-Identification Tools for Rare Diseases

This webinar discussed the need for rare disease communities to adopt common GUIDs and why it helps maximize the use of patient data integrated in RDCA-DAP. An overview of the de-identification solutions available to foundations and registries was also included.

April 27: Clinical Outcome Assessments - Does one size fit all?

This webinar discussed the complexity of identifying appropriate clinical outcome assessments for rare disease clinical trials and provided an overview of the Rare Disease Clinical Outcome Assessment Consortium.

March 15: Critical Path for Rare Neurological Diseases (CP-RND): An Introduction to the Patient Community

An overview of CP-RND with feedback and questions from participants.

View all RDCA-DAP 2022 Webinars here.

For questions or additional information about participating in RDCA-DAP, please email rdcadap@c-path.org. For more information about the initiative, visit: c-path.org/programs/rdca-dap.

Critical Path Institute is supported by the Food and Drug Administration (FDA) of the Department of Health and Human Services (HHS) and is 55% funded by the FDA/HHS, totaling $17,612,250, and 45% funded by non-government source(s), totaling $14,203,111. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement by, FDA/HHS or the U.S. Government.

RDCA-DAP® | CRITICAL PATH INSTITUTE NORD®

#RDCADAP