RDCA-DAP

RARE DISEASE CURES ACCELERATOR
DATA AND ANALYTICS PLATFORM

June 29, 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 first half of the year has flown by, with the RDCA-DAP team busy at work welcoming several new disease areas to the platform, developing platform enhancements and preparing for the 2023 Rare and Orphan Disease Conference this fall. In part, the conference will highlight the newly formed Rare and Orphan Disease Program within C-Path that launched earlier this year. The goal of this expansive program is to increase collaboration among C-Path's rare and orphan disease consortia and initiatives, breaking down any silos that may exist, optimizing available resources and forging clearer collaborations with our partners and stakeholders. Overall, C-Path's Rare and Orphan Disease Program will be more efficient in fulfilling the needs of, and communicating with, the rare disease community.

Get to know C-Path's Rare & Orphan Disease Programs:

- Critical Path for Rare Neurodegenerative Diseases
- Critical Path for Sickle Cell Disease
- Critical Path to Therapeutics for the Ataxias
- Duchenne Regulatory Science Consortium
- Huntington's Disease Regulatory Science Consortium
- Polycystic Kidney Disease Outcomes Consortium
- Transplant Therapeutics Consortium
- Rare Disease Cures Accelerator-Data and Analytics Platform

Data Developments

The platform continues to expand since going live September 2021, containing data for over 26 different rare diseases. More data will be accessible throughout 2023 as outreach efforts continue.

RDCA-DAP currently contains data for the following diseases:

Angelman Syndrome	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
Lennox-Gastaut Syndrome*	Sturge-Weber Syndrome
Mitochondrial Disease	Tuberous Sclerosis

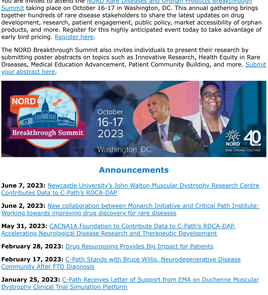
*Indicates disease with datasets that are currently discoverable on the platform

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

Platform Use

Since launching the platform, we've seen the following engagement:

- 361 approved platform requests
- 35 approved workspaces for external users/research



Spotlight: Duchenne Regulatory Science Consortium (D-RSC)

C-Path's D-RSC, created in collaboration with Parent Project Muscular Dystrophy, has generated numerous solutions to advance drug development in Duchenne muscular dystrophy and continues to work toward the expansion of available tools as more data is generated and shared.

D-RSC built an integrated database of patient-level clinical data from DMD studies, including clinical trials, registries and observational studies, which is partially available for analysis by the Duchenne community as permitted by the owners of each dataset. D-RSC published the first standard terminology to integrate data and has written the CDISC Duchenne Muscular Dystrophy Therapeutic Area User Guide, which is available to the community since 2017.

Using this reach database, D-RSC has completed the development of the first Clinical Trial Simulation tool for DMD and recently received a Letter of Support from the EMA while additional regulatory review processes are ongoing for multiple drug development tools.

D-RSC is currently working towards:

- Implementation of new data standard terminology for DMD and other neuromuscular disorders and augmentation of data interoperability in collaboration with multiple organization in the U.S. and Europe
- Regulatory endorsements of the developed disease progression models of five endpoints to integrate into a quantitative solution – a Clinical Trial Simulation (CTS) tool
- Qualification of glutamate dehydrogenase as a liver safety biomarker in trials involving patients affected by muscle disorders
- Regulatory acceptance of quantitative tools based on MRI biomarkers and other DMD outcomes
- Models of disease progression based on additional endpoints as data become available

D-RSC looks forward to expanding future activities to include supporting the regulatory acceptance of other relevant biomarkers and clinical outcome assessments, and enhancing the current tools, to bridge knowledge gaps and facilitate innovative trial design in DMD and other dystrophinopathies. To that end, D-RSC and RDCA-DAP are exploring opportunities to help other rare dystrophies communities and look forward to engaging interested stakeholders.

We welcome anyone to contact the D-RSC team if you're interested in joining our community or sharing data that will support high impact initiatives to advance drug development in Duchenne and other neuromuscular disorders.

NORD Corner

You are invited to attend the NORD Rare Diseases and Orphan Products Breakthrough Summit taking place on October 16-17 in Washington, DC. 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. Register for this highly anticipated event today to take advantage of early bird pricing. Register here.

The NORD Breakthrough Summit also invites individuals to present their research by submitting poster abstracts on topics such as Innovative Research, Health Equity in Rare Diseases, Medical Education Advancement, Patient Community Building, and more. Submit your abstract here.

Announcements

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

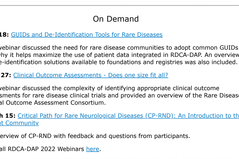
February 28, 2023: Drug Repurposing Provides Big Impact for Patients

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

2023 Rare and Orphan Disease Conference
presented by Critical Path Institute



ABOUT THE EVENT

Attendees of the 2.5-day conference, including patients, providers, researchers, clinicians, biopharmaceutical companies, regulatory reviewers and scientists, will leave with an understanding of:

- C-Path's Rare and Orphan Disease Programs' impact on drug development in rare diseases from the perspectives of patients, regulators and industry partners
- The Critical Path for Rare Neurodegenerative Disease (CP-RND) initiative's first year impact and highlights
- How RDCA-DAP has been enhanced in the last year, and how the platform informs rare disease drug development
- Transversal solutions to address bottlenecks in drug development

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Webinar Series 2023

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

This webinar will focus 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 will provide knowledge and resources to help patients and caregivers understand and engage in the research process.

On Demand

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.

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