Overview of NORD’s Registry Program

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RDCA-DAP Launch Meeting September 17, 2019
Overview

- Brief Introduction to NORD
- IAMRARE™ Registry Program: History, Development, and Growth
- Partnerships and Collaborative Research Models
- Real-World Case Studies from NORD’s Registry Community
- Value and Impact of RDCA-DAP in Context
Introduction to NORD
NORD, an independent nonprofit, is leading the fight to improve the lives of rare disease patients and families.

We do this by supporting patients and organizations, accelerating research, providing education, disseminating information and driving public policy.
Meetings attended on Capitol Hill in 2018

Member Organizations

Phone calls answered annually from patients and caregivers

Website visits annually in 2018 to NORD’s online rare disease reports

Attendees at NORD’s Rare Summit in 2018
Intersecting Programmatic Areas

- Access Support
- Drive Innovation
- Gain Knowledge
- Create Change
- Join Our Rare Family

rarediseases.org
IAMRARE™ Registry Program: History, Development, and Growth
FIRST REGISTRY LAUNCHES ON NORD PLATFORM

10,000 SURVEY SUBMISSIONS

LAUNCH OF FIRST REGISTRIES SUPPORTED BY NORD-FDA COOPERATIVE AGREEMENT

NORD-FDA AWARDEE SELECTIONS

NORD-FDA COOPERATIVE AGREEMENT IS ANNOUNCED

REGISTRY COMMUNITY MEETINGS

NEW MODELS OF ENGAGEMENT

5TH ANNIVERSARY OF PROGRAM

RDCA-DAP LAUNCH MEETING

SUB-STUDY FEATURE

NORD-FDA WORKSHOP
IAMRARE™ Program

- IAMRARE™ Platform
- Training, User Guides
- Study Resources
  - Core Survey Library
  - Custom Survey Support
  - Templates (e.g. consent, marketing)
  - Centralized IRB Partnership
- IAMRARE™ Community
  - Portal, Meetings, Webinars, Newsletters, Videos, Peer-to-peer Discovery
Design & Build
Training
Optimize & Deploy
Ongoing Support
IAMRARE™ Program

Are you an individual or do you know an individual diagnosed with a rare disorder?

You can help us find out more about rare diseases

Why it's important that you participate →

Make an impact on rare disease research in 3 easy steps!

1. Create an Account
2. Add Participants
3. Take Surveys

For patients, parents, guardians, caregivers and legal representatives

See how the Power of Patients is helping to inform our understanding of rare diseases

IAMRARE™ Registry Program →

powered by NORD

rarediseases.org
IAMRARE™ Program
IAMRARE™ Program
Since 2014 Launch:

40+ Registry Partners

10,000+ Participants

80,000+ Survey Submissions
Collaborative Research Models
NORD Partnership Models

Industry & Academia

NORD

Community Organization
Real-World Case Studies from NORD’s Registry Community
Case Studies

**SYNGAP1 heterozygosity disrupts sensory processing by reducing touch-related activity within somatosensory cortex circuits**

Chadwick D. Milford, Emin D. Ozkan, Massimiliano Aceti, Sabyasachi Maity, Nerea Llamosas, Monica Weldon, Elisa Mizrahi, Thomas Vaissiere, Michael A. Gaffield, Jason M. Christie, J. Lloyd Holder Jr., Courtney A. Miller and Gavin Rumbaugh

The Registry Offers a Platform for Patients to Share Their Stories via Surveys
The Foundation for Prader-Willi Research (FPWR) and Zafgen are pleased to announce that enrollment is now open for PATH for PWS, a natural history study intended to better understand serious medical events in Prader-Willi syndrome (PWS) and evaluate how PWS-related behaviors change over time. The data from this study is intended to inform the development and clinical trial design of potential new treatments for PWS. Those interested in participating can find more information about the study and how to enroll at www.PATHforPWS.com.

Enrollment is now open for the four-year study using the Global PWS Registry, which is powered by the National Organization for Rare Disorders' (NORD) IAMRARE™ Registry Program. To be eligible for the study, participants must have a confirmed diagnosis of PWS, be at least 5 years of age, live in the United States, Canada or Australia, and be enrolled or willing to enroll in the Global PWS Registry. The primary caregiver of the enrolled person with PWS must have access to the internet to enter study data and consent to being contacted by registry staff.
Case Study

OMS Life Foundation

OMS – Opsoclonus-Myoclonus Syndrome

OMS is a rare, orphan disease affecting as few as 1 in 5,000,000 and primarily impacting children 15 years old. Suspected to result from an autoimmune process involving the nervous system, OMS may be manifested as deficiencies in speech, physical activities, learning, and other general life skills.

Bridge the Gap SYNGAP

SYNGAP1-related NSID

SYNGAP1 NSID is thought to result from limited functional levels of SynGAP protein, a protein critical in proper brain development and function. Predominantly affecting children, SYNGAP1 mutations lead to developmental delay, intellectual disability, and additional symptoms that are common with other causes. Here, we describe patient-reported quality of life as collected through the SYNAPGAP (MRDS) patient registry.

National PKU Alliance

PKU – Phenylketonuria

Phenylketonuria (PKU) is a rare, brain threatening, inherited metabolic disorder characterized by inability of the body to utilize the essential amino acid, phenylalanine (PHE).

When left untreated, PKU patients with excess PHE are at risk of severe neurological complications, including IQ loss, memory loss, concentration problems, mood disorders, and, in some cases, severe mental retardation.

Damage is irreversible, so early detection is crucial.

The Desmoid Tumor Research Foundation

Desmoid Tumor

As few as 5 - 6 per 1 million people are diagnosed with desmoid tumors annually, which may be an underestimate of the actual affected population due to difficulty in correctly diagnosing the disease. These locally invasive, noncancerous tumors can yield a wide range of symptoms including no symptoms at all. Conclusive diagnosis requires biopsy and a pathologist experienced in or knowledgeable of rare cancers. To improve awareness of desmoid tumors, DTRF, in partnership

IPPF International Pemphigus and Pemphigoid Foundation

Pemphigus and Pemphigoid

Pemphigus and pemphigoid (PP) are rare, autoimmune blistering diseases that affect a very small percentage of the population, thus real-world data is needed to better understand diagnosis and treatment patterns in these difficult to diagnose patients.

Platelet Disorder Support Disorder

ITP – Immune Thrombocytopenia

Patients with the autoimmune disease ITP suffer from bleeding events as a result of low platelet counts. These events may manifest as bruises, petechiae, blood blisters, bloody stools, blood in urine, or even bleeding in the brain. Treatments vary by severity of disease but include medications (immunosuppressives, XIG, platelet boosters) and splenectomy. Both the disease and the treatments impact quality of life for these patients, who commonly state concerns of anxiety and fatigue. In 2017, PDSA in collaboration with NORD launched the ITP Natural History
Value and Impact of RDCA-DAP in Context
Value and Impact of RDCA-DAP

- Transformative collaboration
- Leveraging capabilities and expertise
- Development of new tool(s) and data optimization to accelerate discovery and therapeutic product development
- Flexibility to design solutions to overcome well-known data challenges
- Effective use of resources
- Innovative technologies that drive efficiencies and reduce costs
- Standard-setting and evaluation of measure sets*
- Convening a ready-made collaborative global network to support clinical trials*
RDCA-DAP in Context

NORD Registry-Now
- Centralized disease-neutral platform
- Prospective natural history data collection
- Consolidation of stakeholder efforts
- Community-driven

NORD Registry-Next
- Evaluate COAs for use across conditions
- Systematic collection of data
- Set standards for recruitment, retention, and engagement
- Education and training

NORD Registry-Near Future
- Support natural history study designs that can serve as external control
- In partnership, demonstrate successful usability of natural history for controls
- Define global rare disease data standards
- Return of value to community
- Consolidated source for registry data integration for RDCA-DAP
• Our rare disease research partnerships reflect authentic engagement and sustained collaboration.

• NORD is the primary initiation point for patient organizations interested in participating in the RDCA-DAP.

• Our model can keep data proprietary and separate, but the community together.

• With our partners at C-Path and FDA we are designing solutions to bridge stakeholder needs and deliver impact.

• We hope you will join us as registry partners, data partners, and research project collaborators!
Questions?

Contact: research@rarediseases.org
Thank you.