



NORD[®]
National Organization
for Rare Disorders

Overview of NORD's Registry Program

Vanessa Boulanger, Director of Research

RDCA-DAP Launch Meeting September 17, 2019

Alone we are rare. Together we are strong.®



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



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Madilyn Yang (far left) has been battling a rare breathing disorder since birth called Central Congenital Hypoventilation Syndrome (CCHS) or Ondine's Curse.

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.



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Intersecting Programmatic Areas

148

Meetings attended on Capitol Hill in 2018

280 Member Organizations

150K+

Phone calls answered annually from patients and caregivers



12.7 Million

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

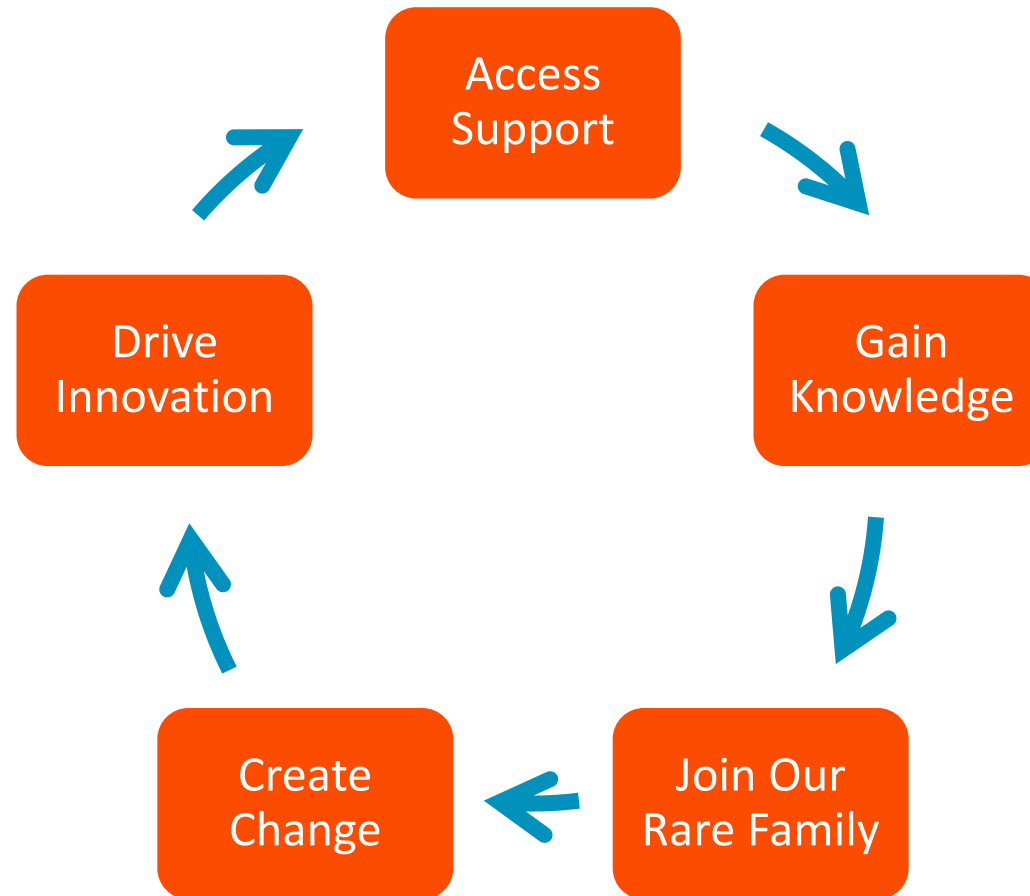
800+

attendees at NORD's Rare Summit in 2018



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Intersecting Programmatic Areas



IAMRARE™ Registry Program: History, Development, and Growth

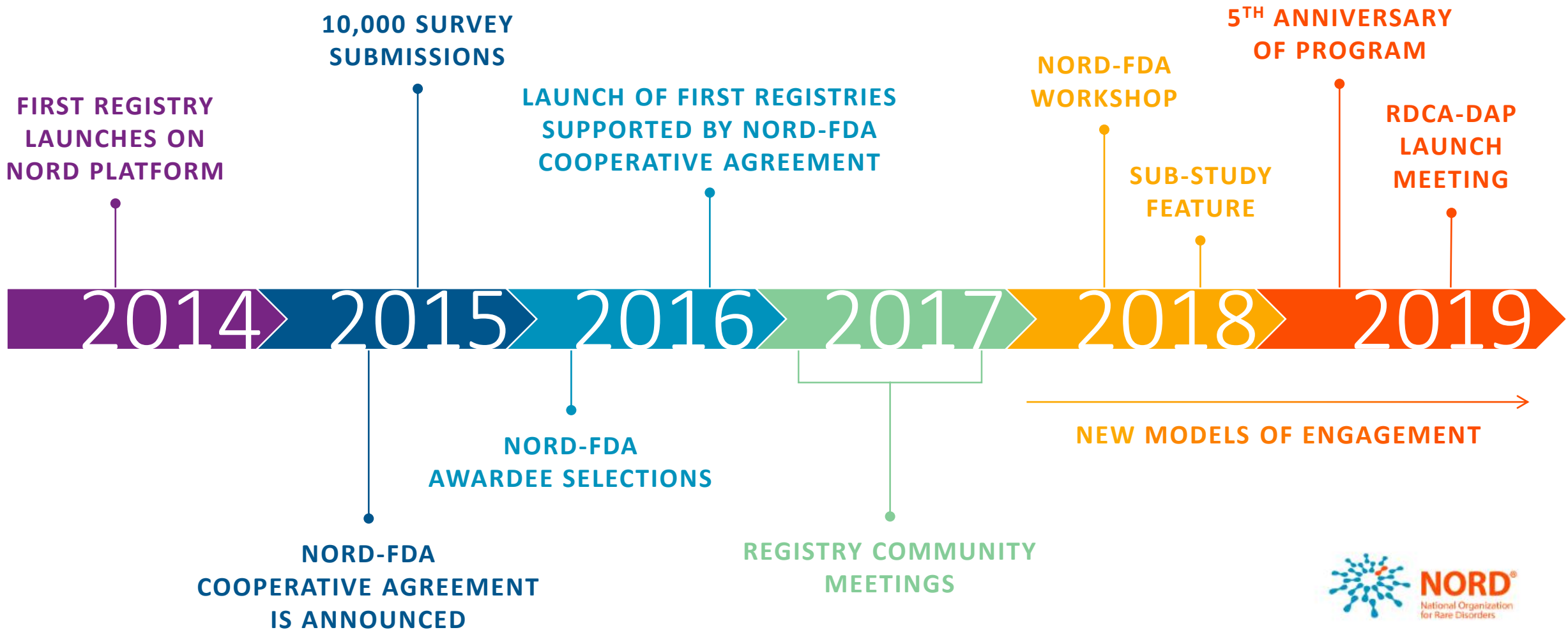


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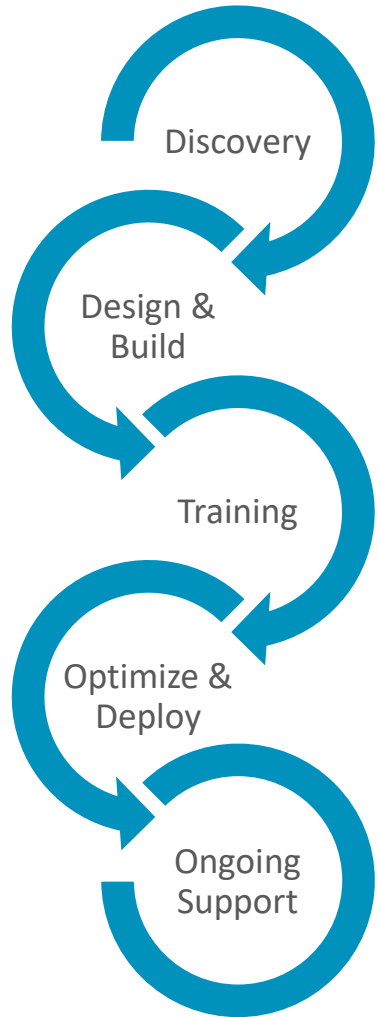
*Stacy McCaroll and
daughter, Cystic Fibrosis*

History of the IAMRARE™ Program



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



IAMRARE™ Program

POWER OF PATIENTS
REGISTRY

[For Patients](#) | [For Researchers](#) | [News](#) | [FAQs](#)

[Contact us](#)

[Join Today!](#)

[Log in](#)

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

[Join Today!](#)

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

[IAMRARE™ Registry Program >](#)

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IAMRARE™ Program

POWER OF PATIENTS
REGISTRY

For Patients

For Researchers

Community

News

Contact us

+ Add Participant

Log out

Your Registry Dashboard

Welcome back, Mary!

Your Registry Dashboard


You are one of 746 participants in this registry. If you know of anyone else who may be interested in enrolling, send them an invite. Their email address will only be used once to email the invite.

Send Invites

One of your participants is eligible to join **The Hope Study**. To find out more and enroll, click on the button for the eligible participant.

Your Participants


Participant Manager



Mary Smith
POP Registry Consent granted >

Take Surveys Manage Consent

Independent partner studies:
HOPESTUDY Enrolled >



Henry Smith
POP Registry Consent granted >

Take Surveys Manage Consent

Independent partner studies:
HOPESTUDY Join the Hope Study >

Your Actions

Transfer Request ?

Received on May 24, 2019 from John Kygie for participant Henry Smith

Relationship to participant: Other non-relative
Email: henry.smith@outlook.com

1. You must complete 1 survey before the transfer:

Complete Unfinished Surveys

Reject

2. Set the participant manager viewing permissions:

All survey data Only survey data Henry Smith entered

Approve

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IAMRARE™ Program

POWER OF PATIENTS REGISTRY For Patients | For Researchers | Community | News | Contact us | + Add Participant | Log out

Power of Patients \ IAMRARE Community

Welcome back, Prof. Dr. Daniel Smith!
Study Administrator Community


Do you have any news?

Dr. Brodie Smith
05:59 AM, 10 September

Thoughts from the Front Lines of Rare Disease Research

There are nearly 7,000 rare diseases, some of which affect just a few dozen people. Yet, if one considers all these conditions together, about 30 million people in the United States have rare diseases. On this Rare Disease Day, I'd like to challenge each of you to think about how we can raise the visibility of individuals living with rare diseases, as well as the researchers working hard to help them.

I'd like to introduce you to Harper Spero, who is using her rare gift of storytelling to share the experiences of people with a wide variety of conditions that she likes to call "invisible illnesses." Through her podcast series, called *Made Visible*, this 34-year-old New York City native is among the many people helping to spread the word that rare diseases are not rare.



Harper Spero with physician-researcher Alexandra Freeman, who helps lead the Job's syndrome research team at the NIH Clinical Center.


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Prof. Dr. Daniel Smith
05:59 AM, 10 September

The need for a next-generation public health response to rare diseases.

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Connection Requests

- Arif Farrington (New York, New York) [X] [✓]
- Liliana Sadler (Los Angeles, California) [X] [✓]
- Franco Sellers (Chicago, Illinois) [X] [✓]
- Lewis Fountain (Toledo, Ohio) [X] [✓]

Upcoming Events

RARE DISEASE DAY®

Rare Disease Day 2020
29 February, 2020 **170** days

People you may know

- Chandler Molina (New York, New York) [M] [+
- Maximilian Cantrell (Los Angeles, California) [M] [+
- Alexandra Cantu (Chicago, Illinois) [M] [+
- Jensen Barrett (Toledo, Ohio) [M] [+

If you have any questions or comments, please add them to the site or contact the NORD Registry Team: 1-800-999-6673 or research@rarediseases.org

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National Organization for Rare Disorders



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IAMRARE™ Program

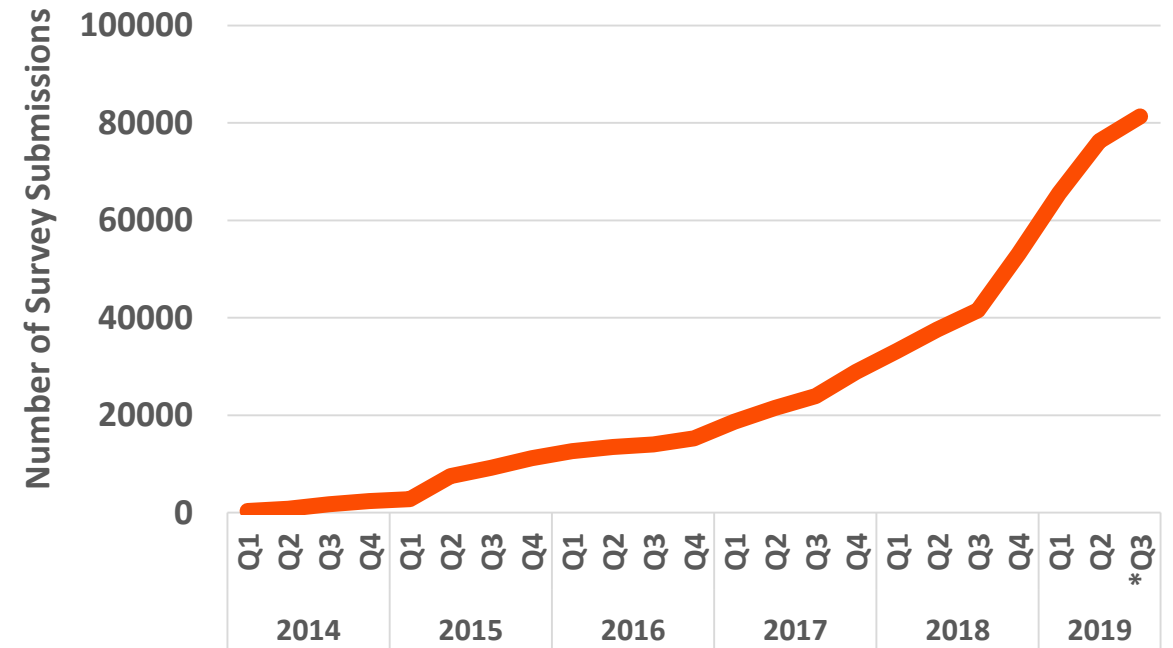
Since **2014** Launch:

40+ Registry Partners
10,000+ Participants

80,000+ Survey Submissions



Total Number of IAMRARE™ Survey Submissions





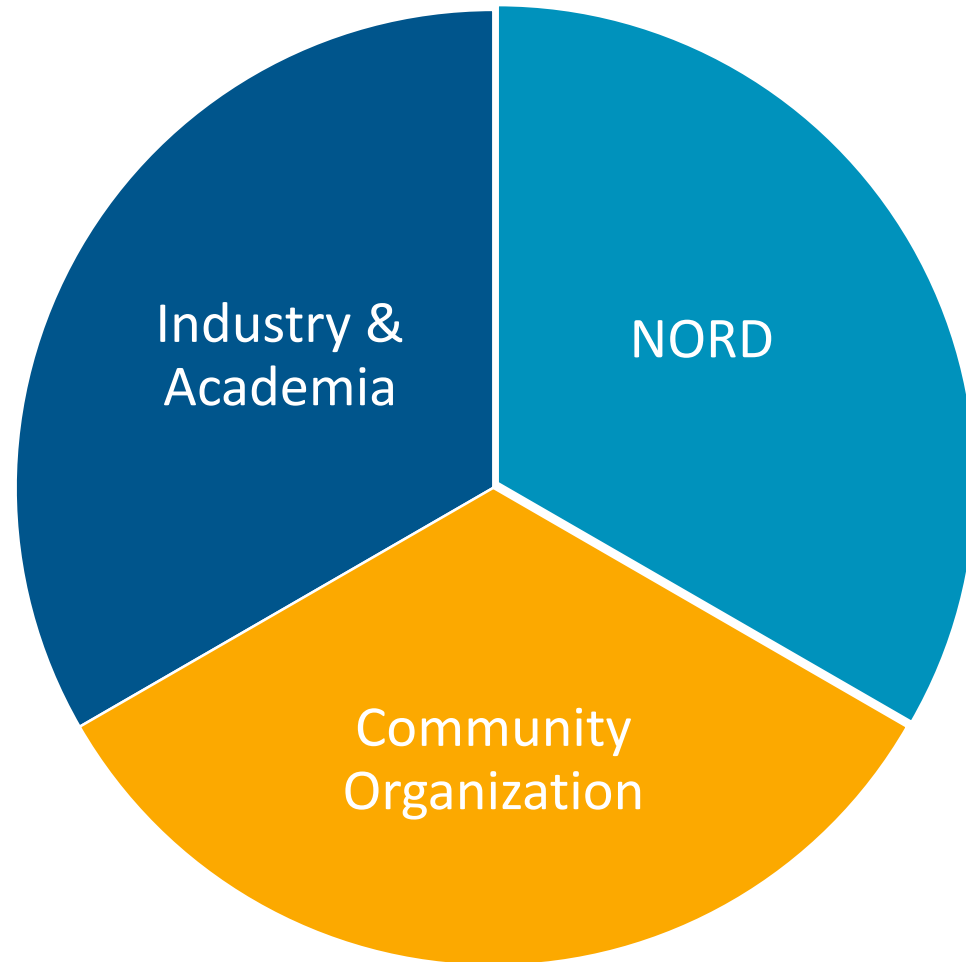
Collaborative Research Models

Vaughn family: Son, Morgan (left), diagnosed with Necrotizing Enterocolitis at four days old

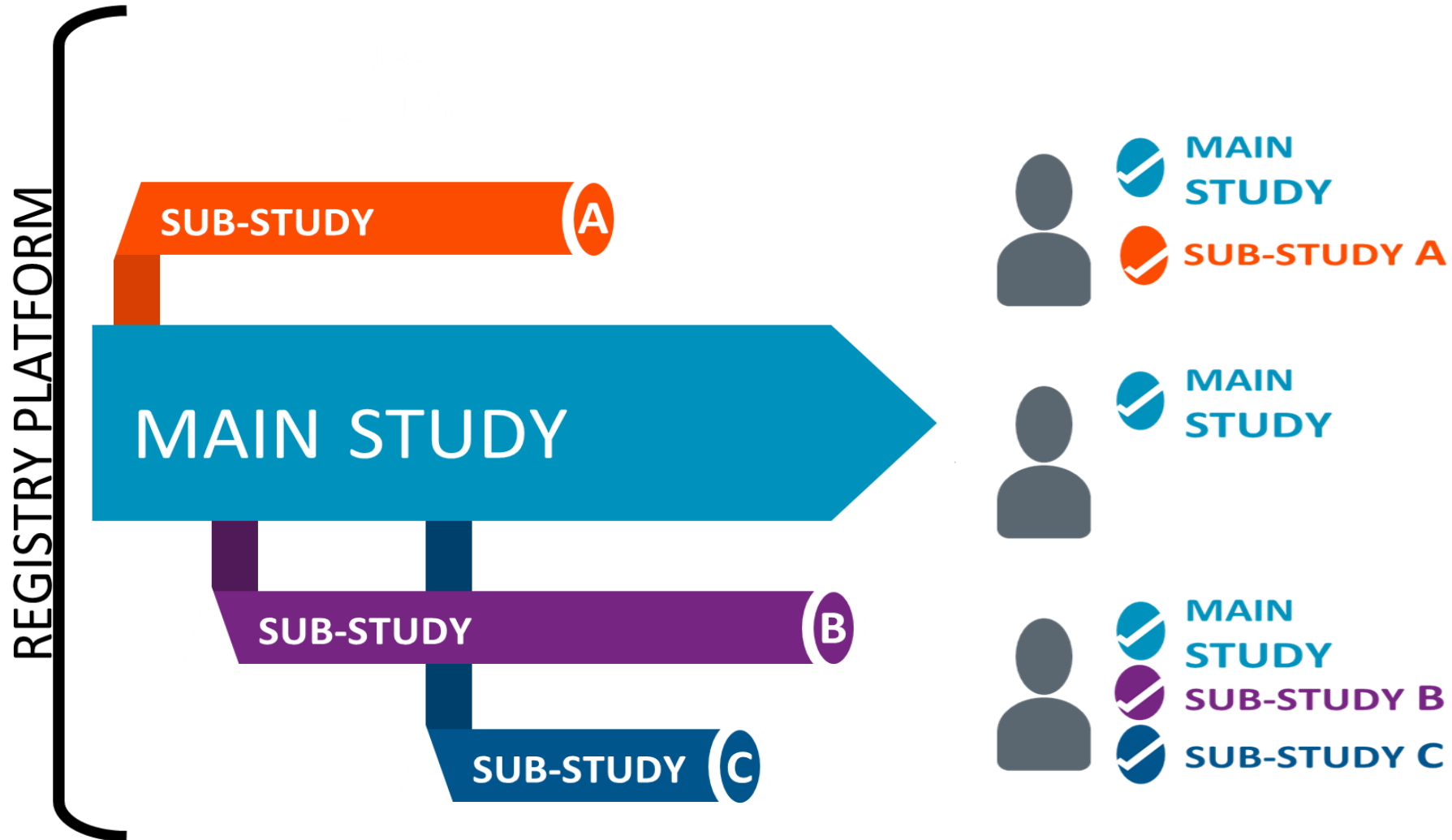


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NORD Partnership Models



NORD Partnership Models



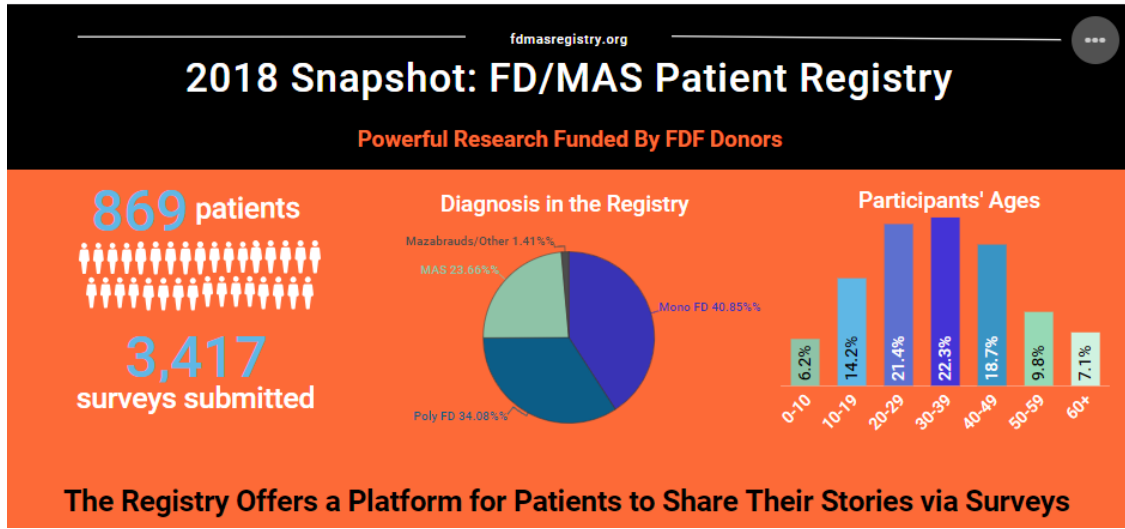
Real-World Case Studies from NORD's Registry Community



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Case Studies



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

Shelley D. Mitchell^{1,7}, Emin D. Ozkan^{1,7}, Massimiliano Aceti^{1,6}, Sabyasachi Maity¹, Nerea Llamosas¹, **Monica Weldon²**, Elisa Mizrachi¹, Thomas Vaissiere¹, Michael A. Gaffield³, Jason M. Christie³, J. Lloyd Holder Jr., Courtney A. Miller^{1,5} and Gavin Rumbaugh^{1,5*}



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Partnerships to Conduct Research (PaCR) within PCORnet

Case Study

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.

PATH
for PWS™
Paving the way for Advances
in Treatments & Health for PWS



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Case Study



OMSLife Foundation OMS – Opsoclonus-Myoclonus Syndrome

OMS is a rare, orphan disease affecting as few as 1 in 5,000,000 and primarily impacting children 1-5 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 SYNGAP1 (MRD5) 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

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 (P/P) 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 Association 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 , IVIG, 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



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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



Summary Points

- 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



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Thank you.



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