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

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

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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.

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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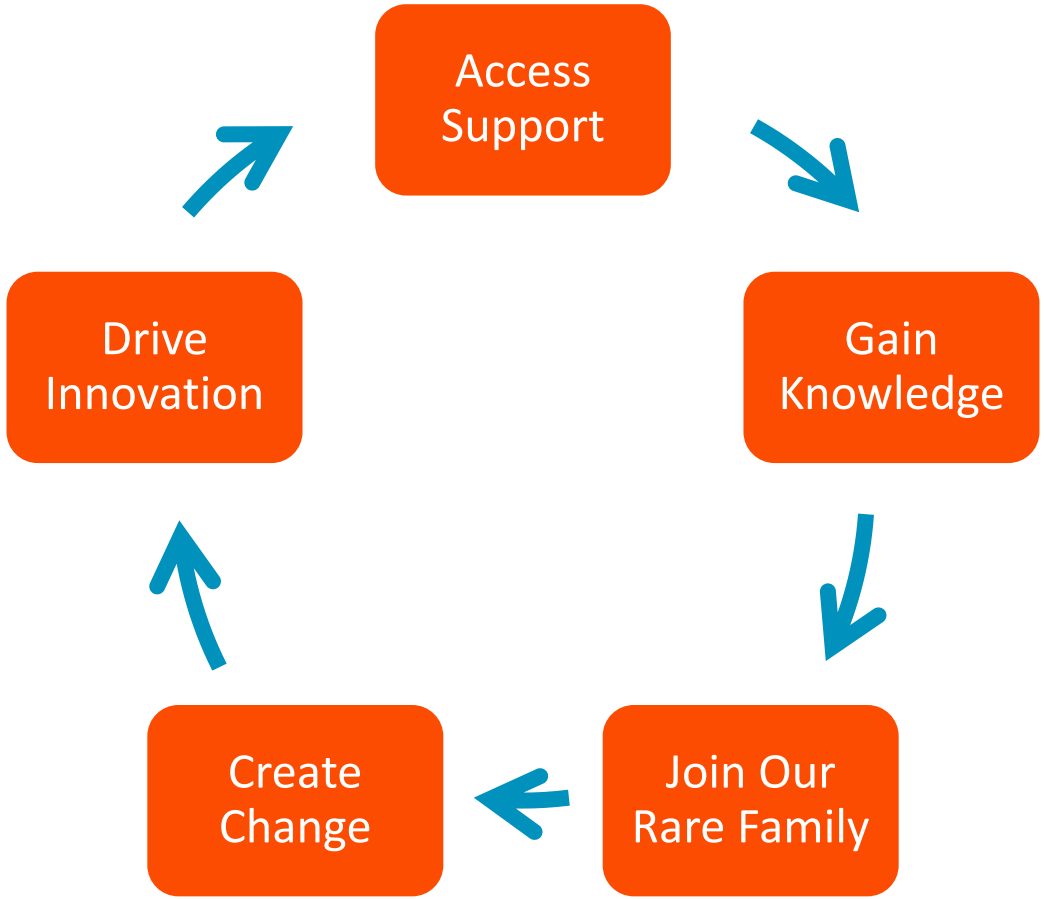
Intersecting Programmatic Areas



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IAMRARE™ Registry Program: History, Development, and Growth

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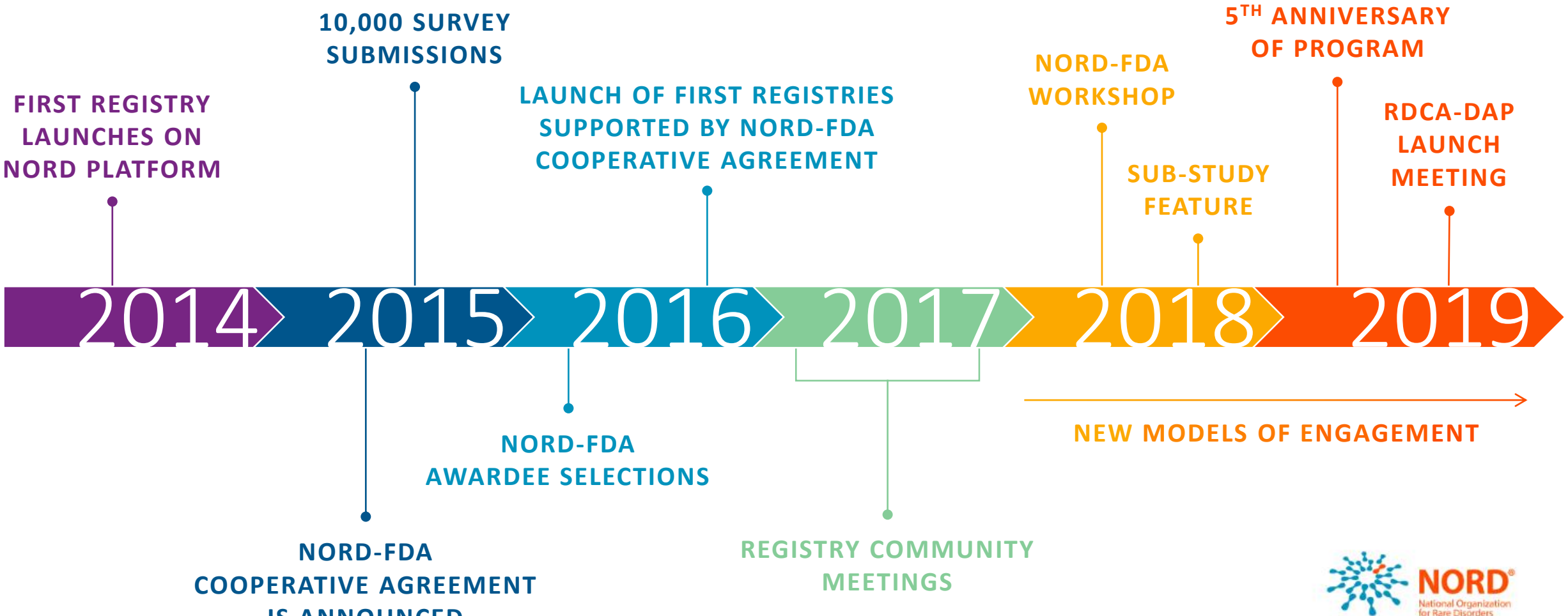
Stacy McCaroll and daughter, Cystic Fibrosis

History of the IAMRARE™ Program



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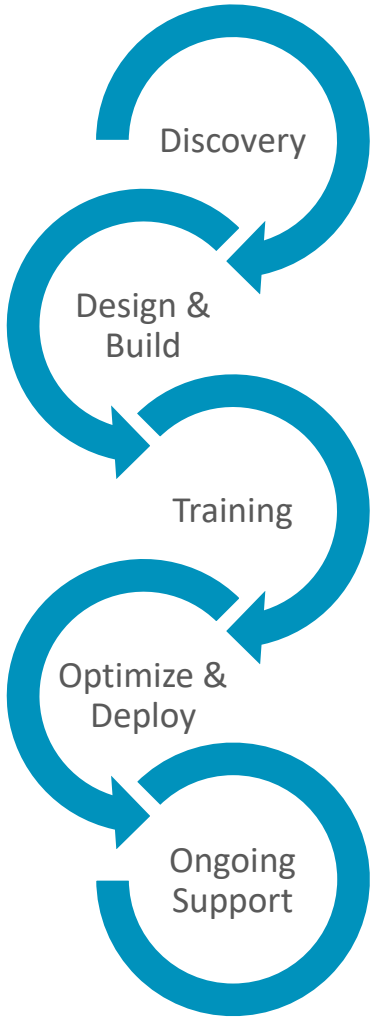
IAMRARE™ Program



- 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

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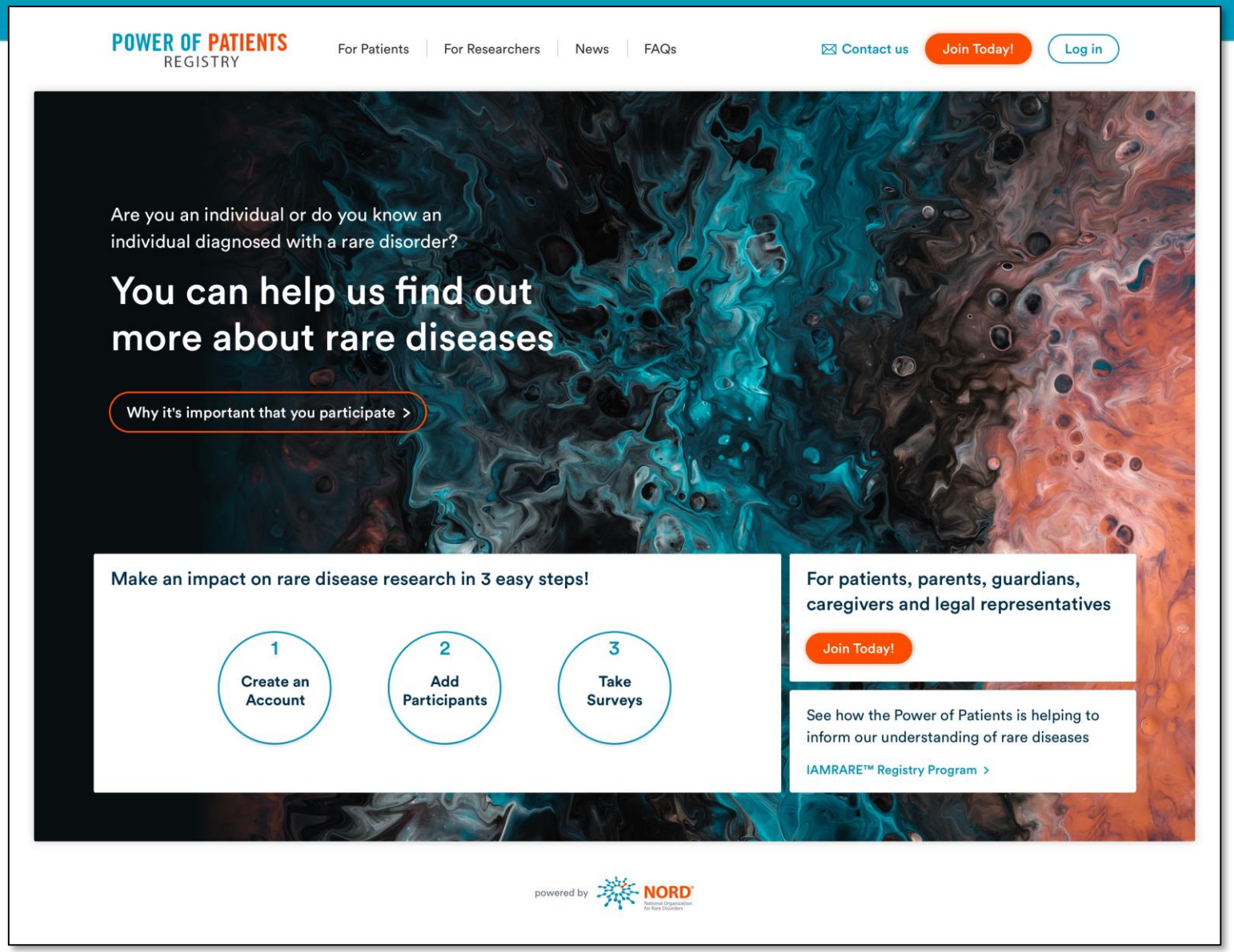
IAMRARE™ Program



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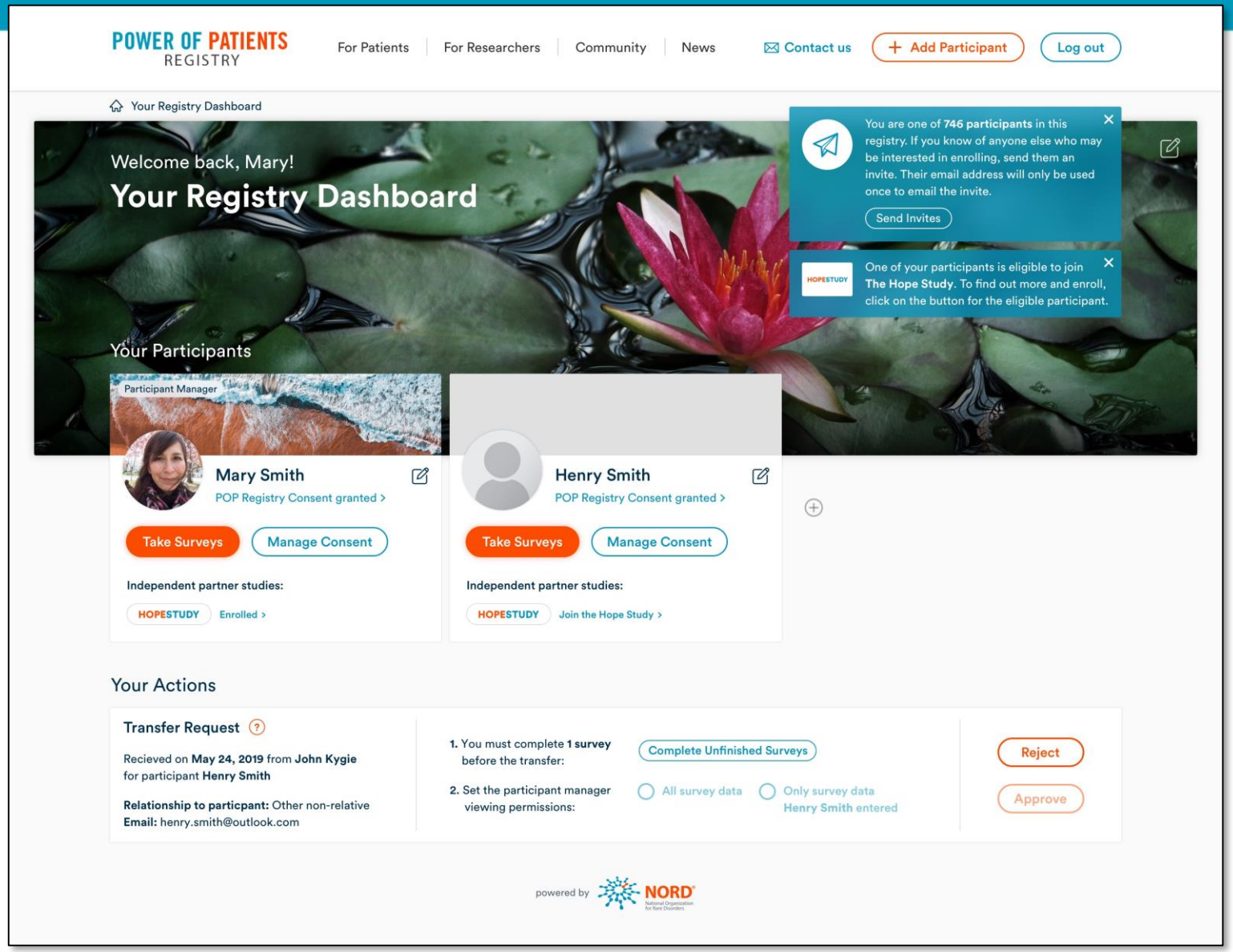
IAMRARE™ Program



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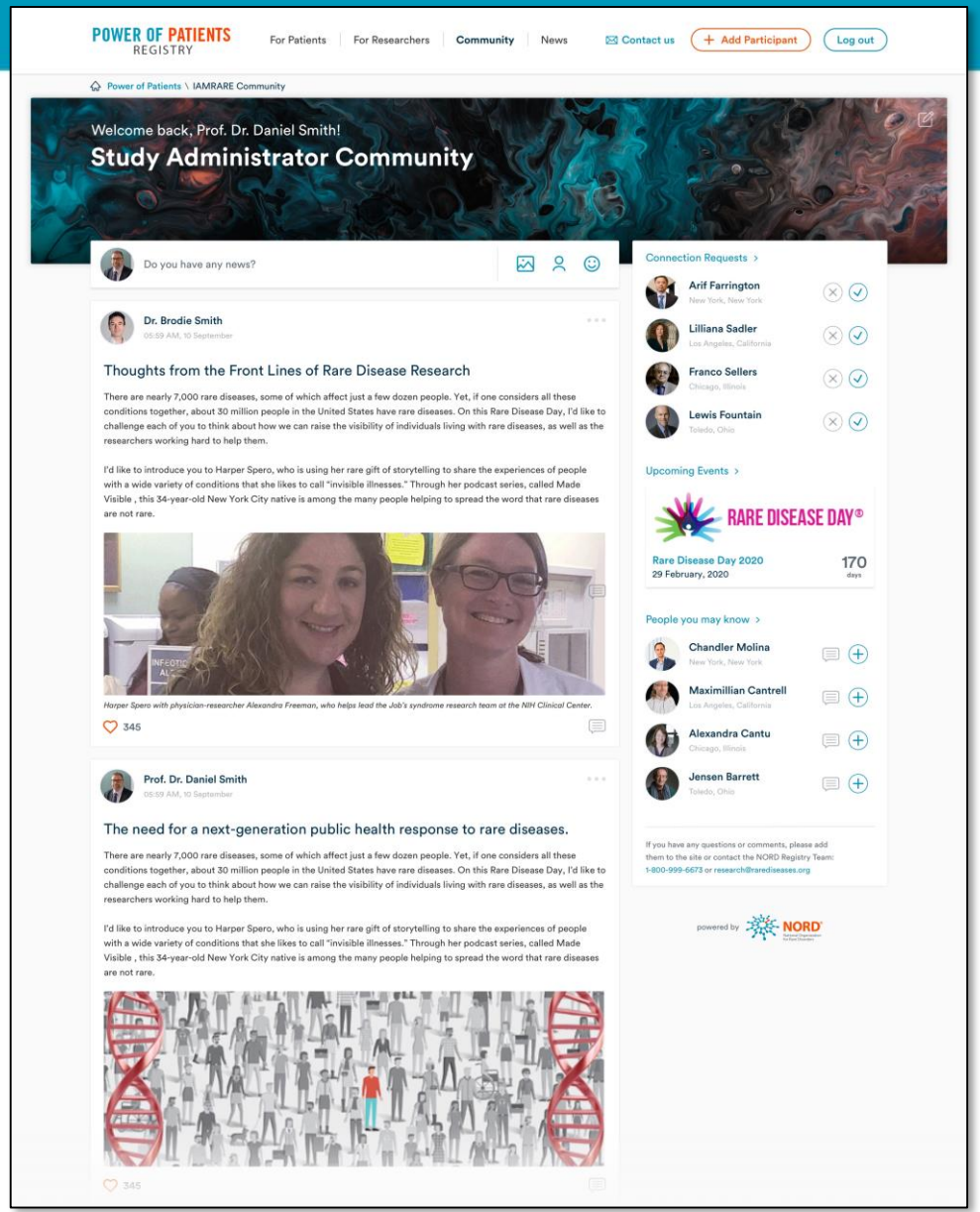
IAMRARE™ Program



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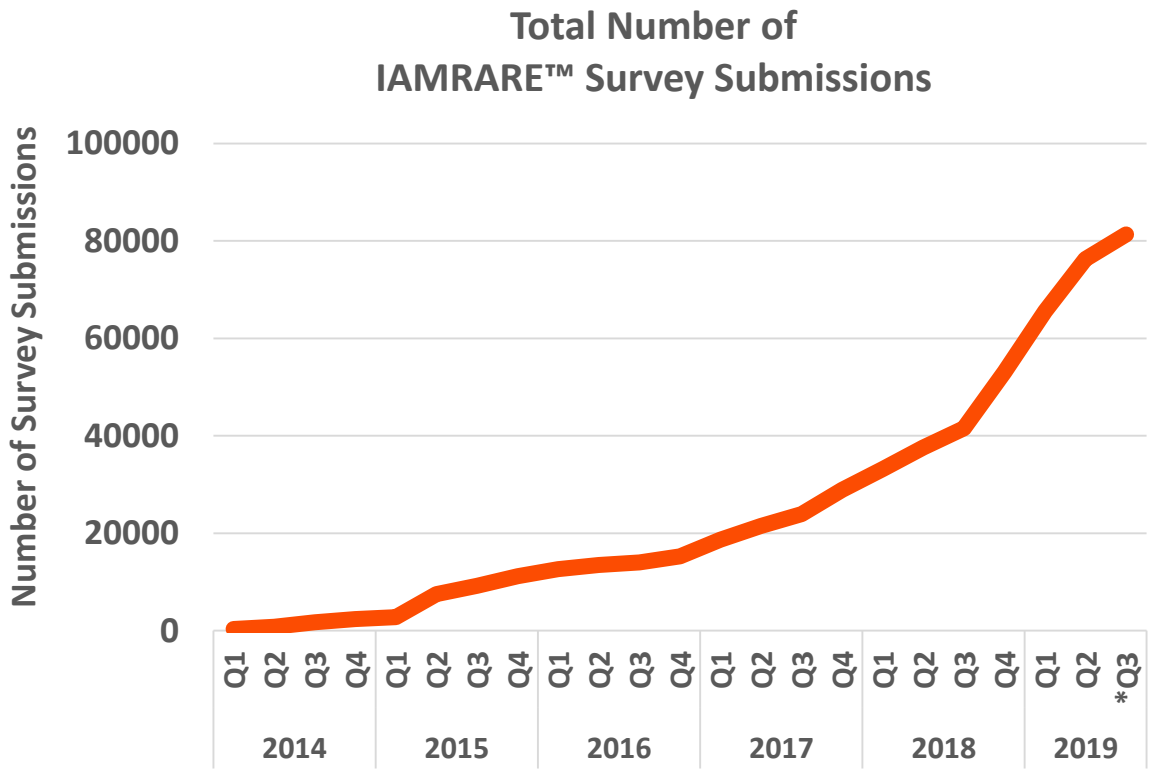
IAMRARE™ Program

Since **2014** Launch:

40+
Registry Partners

10,000+
Participants

80,000+
Survey Submissions



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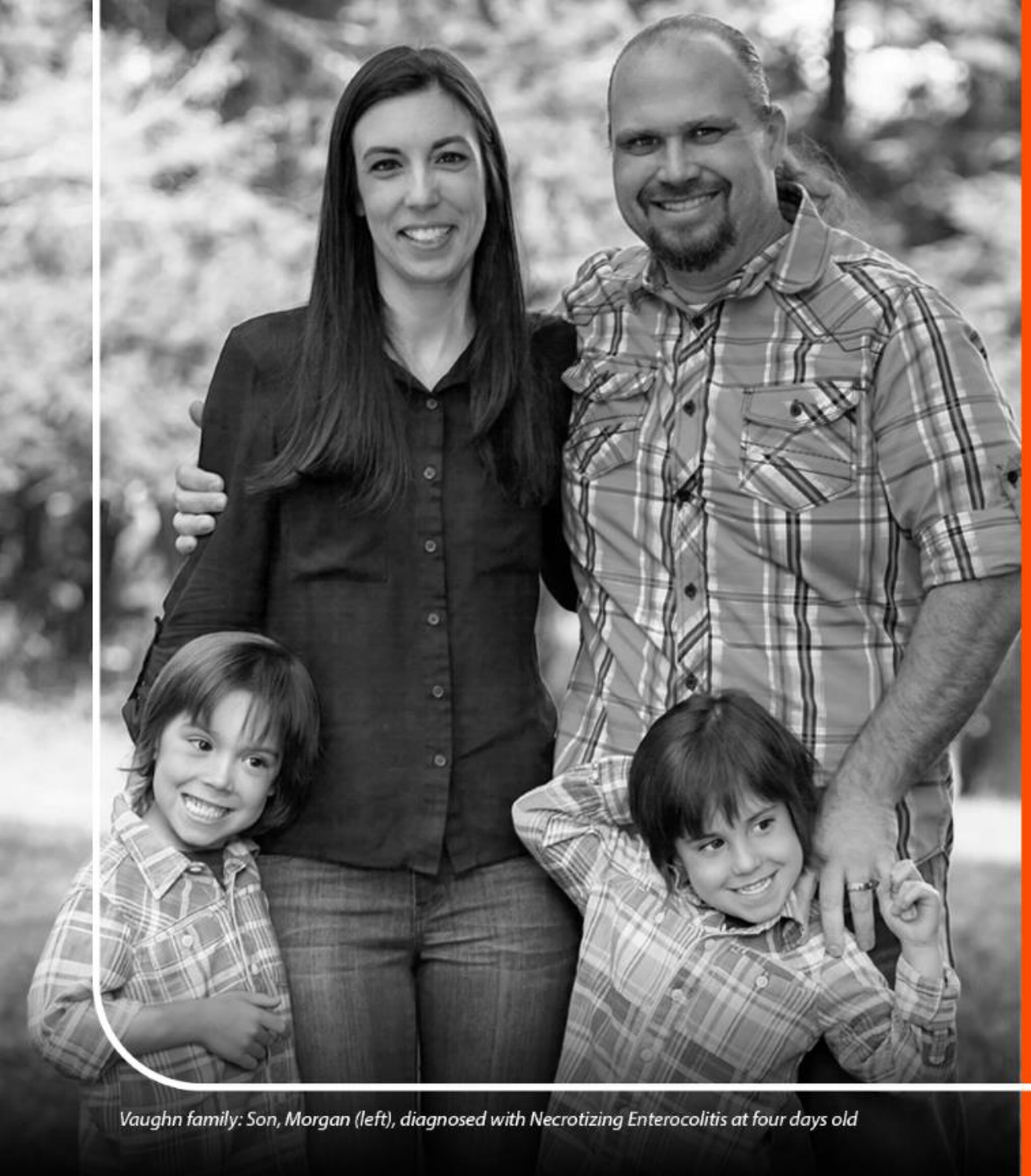

Vaughn family: Son, Morgan (left), diagnosed with Necrotizing Enterocolitis at four days old

Collaborative Research Models

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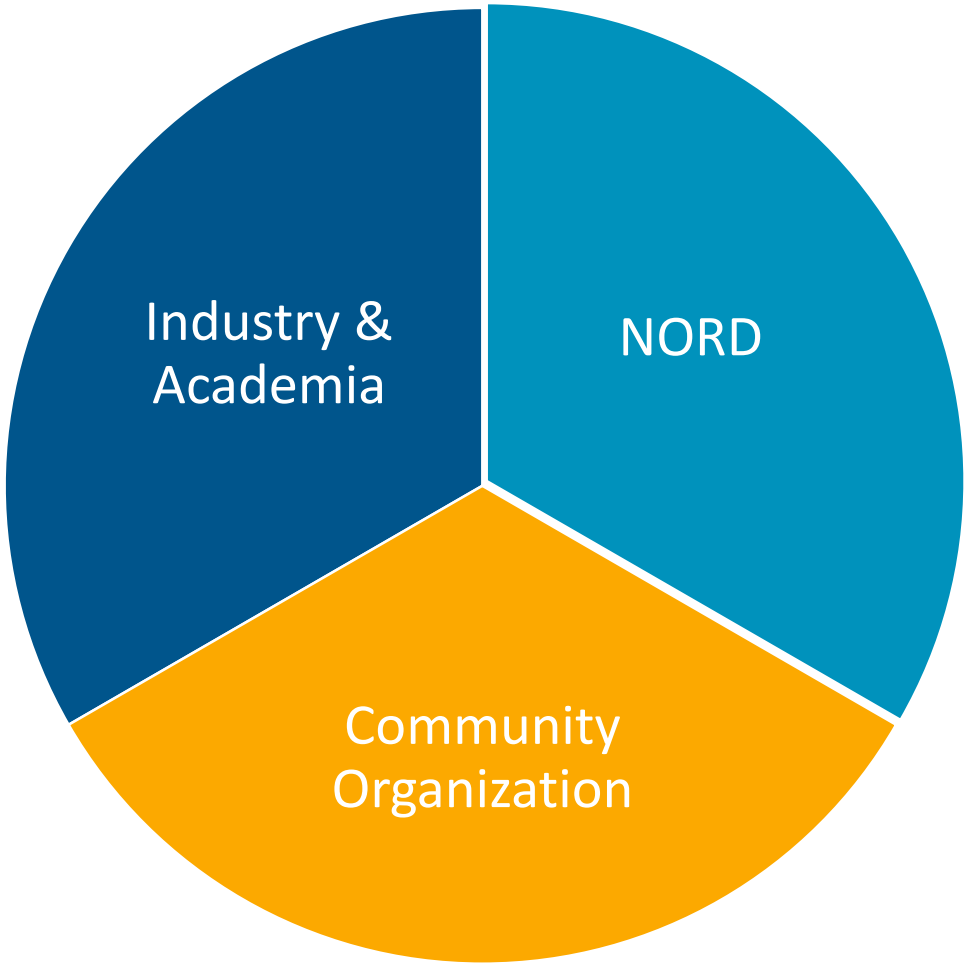
NORD Partnership Models



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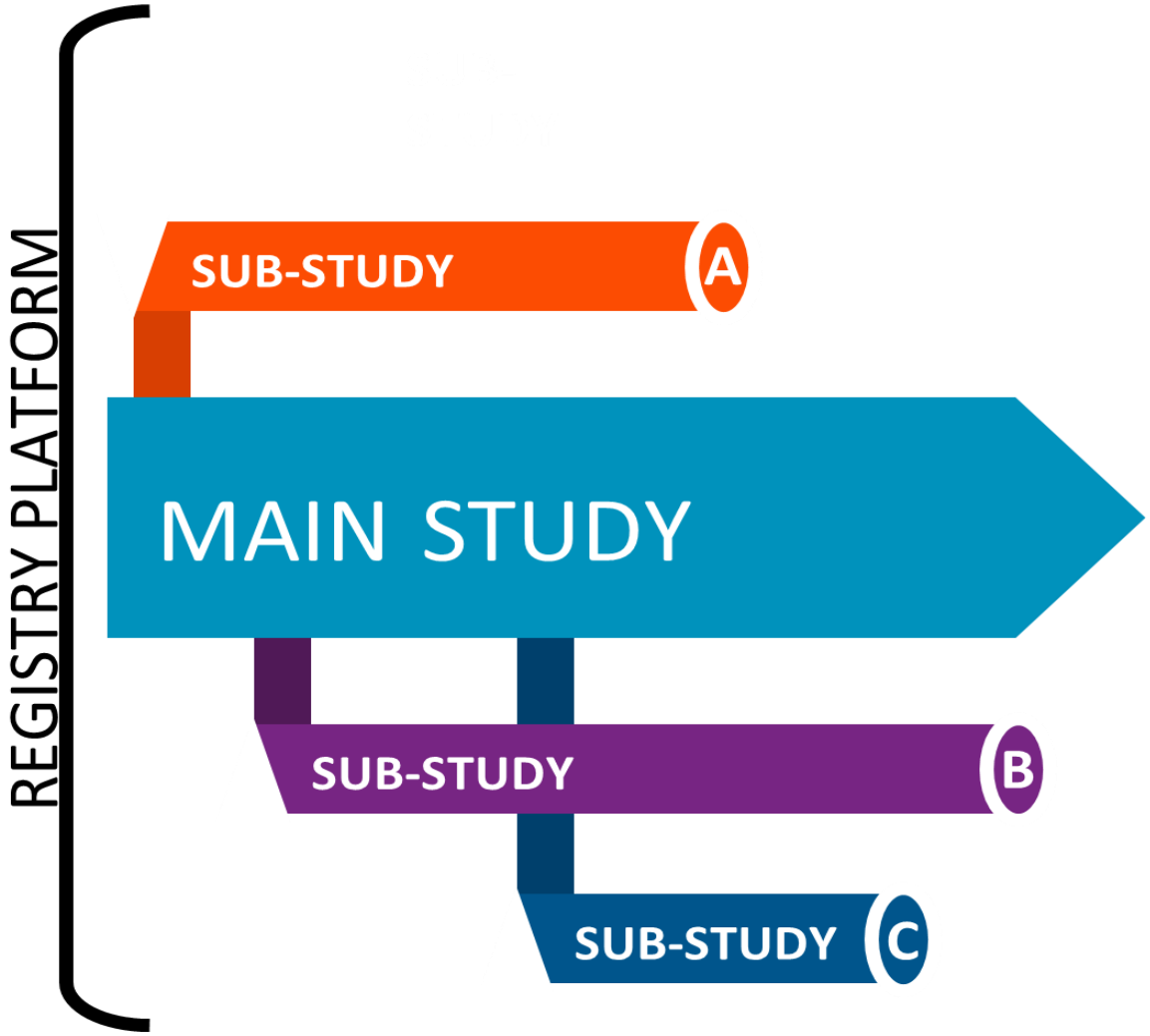
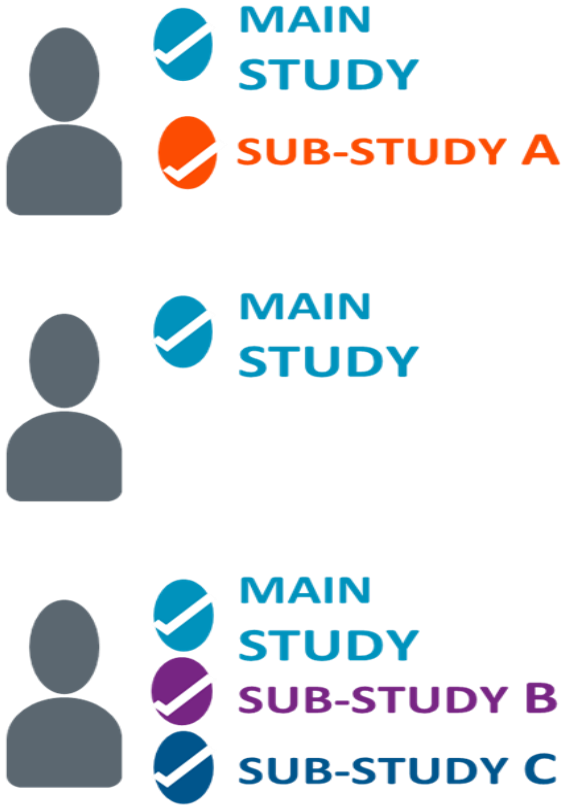
NORD Partnership Models





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Real-World Case Studies from NORD's Registry Community

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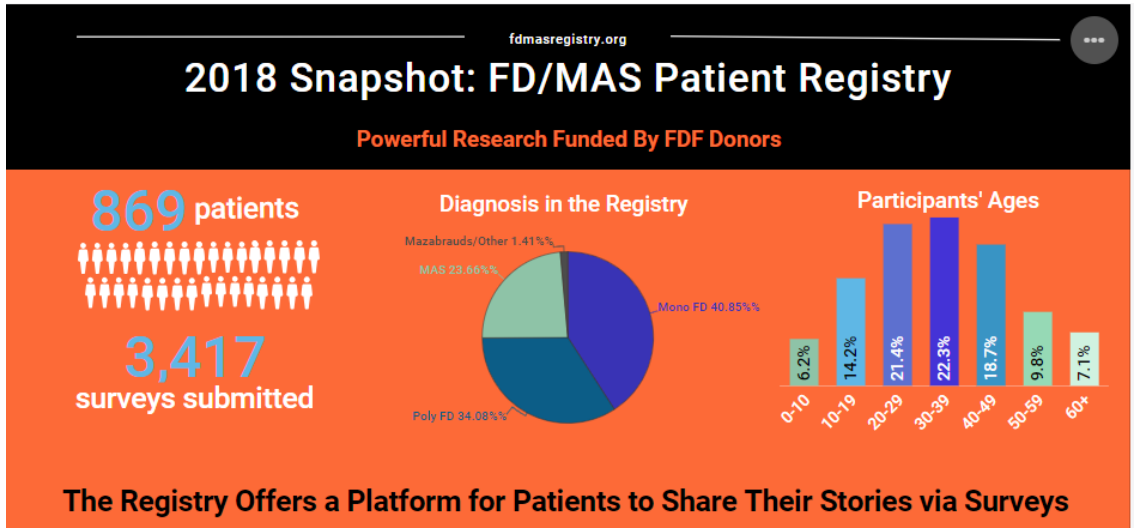
Case Studies



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

Monica Weldon[2], Courtney A. Miller[1,5] and Gavin Rumbaugh[1,5*]

pcori
PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

GBS | CIDP
Foundation International

Platelet Disorder Support Association
20 Years
Empowering ITP Patients

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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

FOUNDATION FOR PRADER-WILLI RESEARCH

Working Toward an Independent Future

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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
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 Internationa 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 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 , 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

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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*

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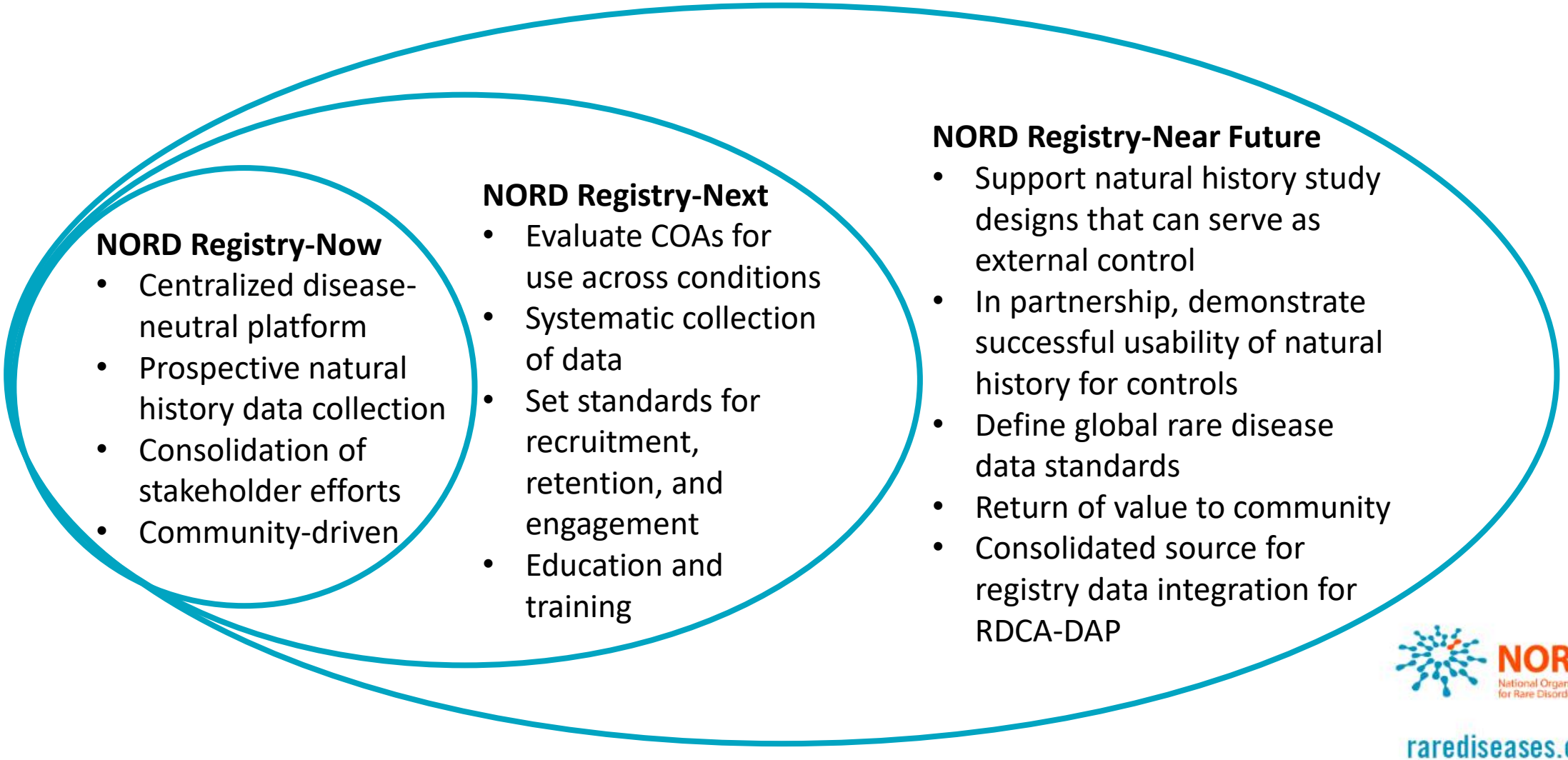
RDCA-DAP in Context



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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!

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Questions?

Contact: research@rarediseases.org

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