NORD®
National Organization for Rare Disorders

IAMRARE Registry & Patient-Centered Research Program Overview

Vanessa Boulanger, Director of Research

Presentation for the Advisory Committee on Heritable Disorders in Newborns and Children, March 22 2019

Alone we are rare. Together we are strong.®

Overview

- Brief Introduction to NORD
- IAMRARE Registry Program: Overview of Growth and Impact
- Registry Partnerships and Models of Engagement
- Opportunities to Engage: Research Studies and Data Resources

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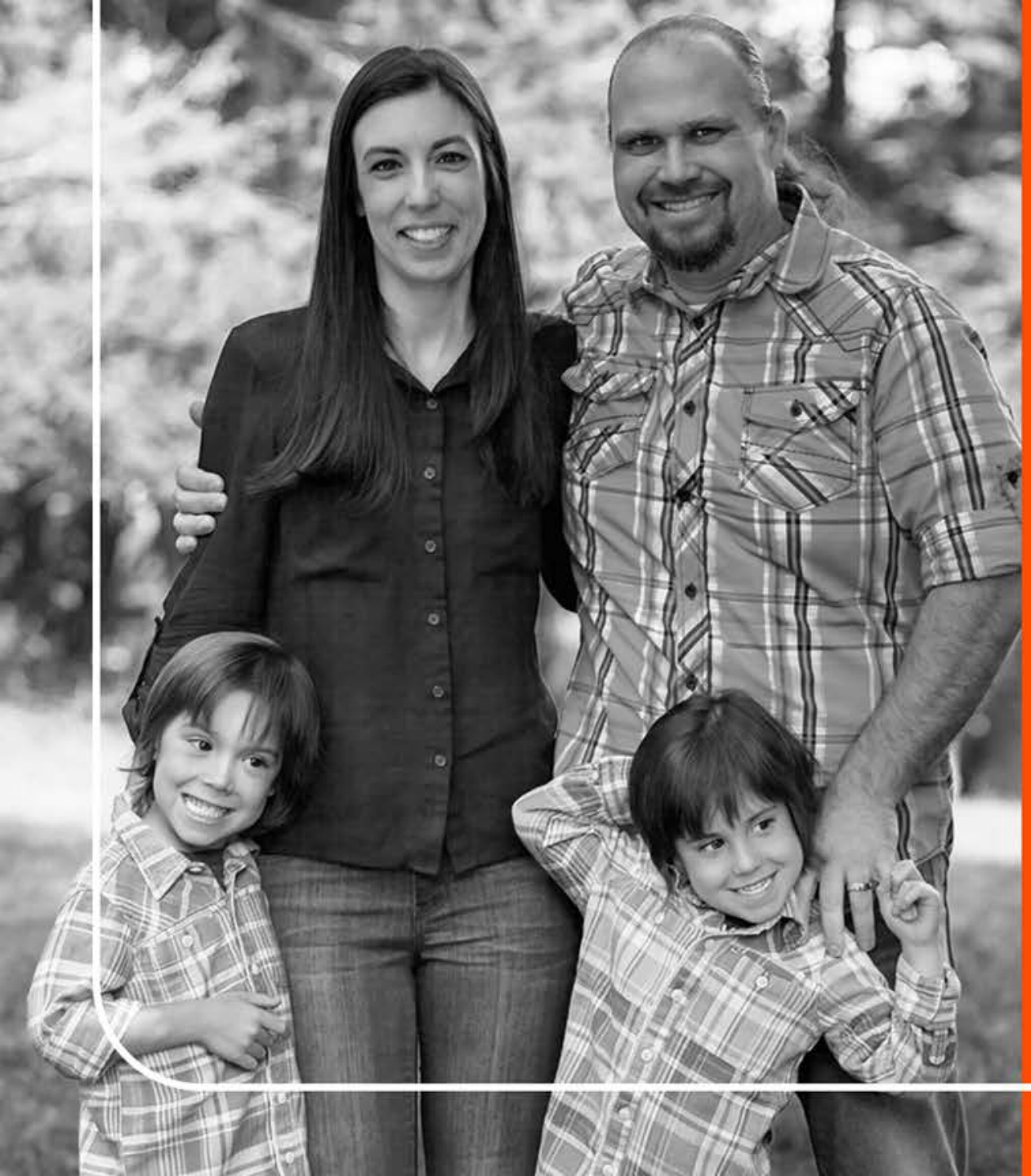

Introduction to NORD

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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.

Pillars of NORD

Tracking > 150 bills

Hosted over 200 legislative meetings

Servicing >7,000 patients/yr

Fielding 114k phone calls/yr

Policy & Advocacy

- Federal and State Policy
- Regulatory Affairs
- Rare Action Network® Grassroots advocacy coalition present in all 50 states
- Advocate training workshops

Research

- Research grants for translational or clinical studies
- IAMRARE® registry platform and natural history study development program
- Original research and publications

Patient Services

- Premium, copay and coinsurance support
- Diagnostic and genetic testing
- Ancillary services
- Clinical trial recruitment and travel & lodging
- Emergency relief programs

Education

- Patient and family education
- Medical professional education
- Annual Rare Diseases & Orphan Products Breakthrough Summit
- Rare disease mentorship & workshops for patient advocacy groups

Over 30 Registry Partnerships

2 FDA Approved Products

1,200+ rare disease reports

Summit Draws 700+ Attendees

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IAMRARE Registry Program

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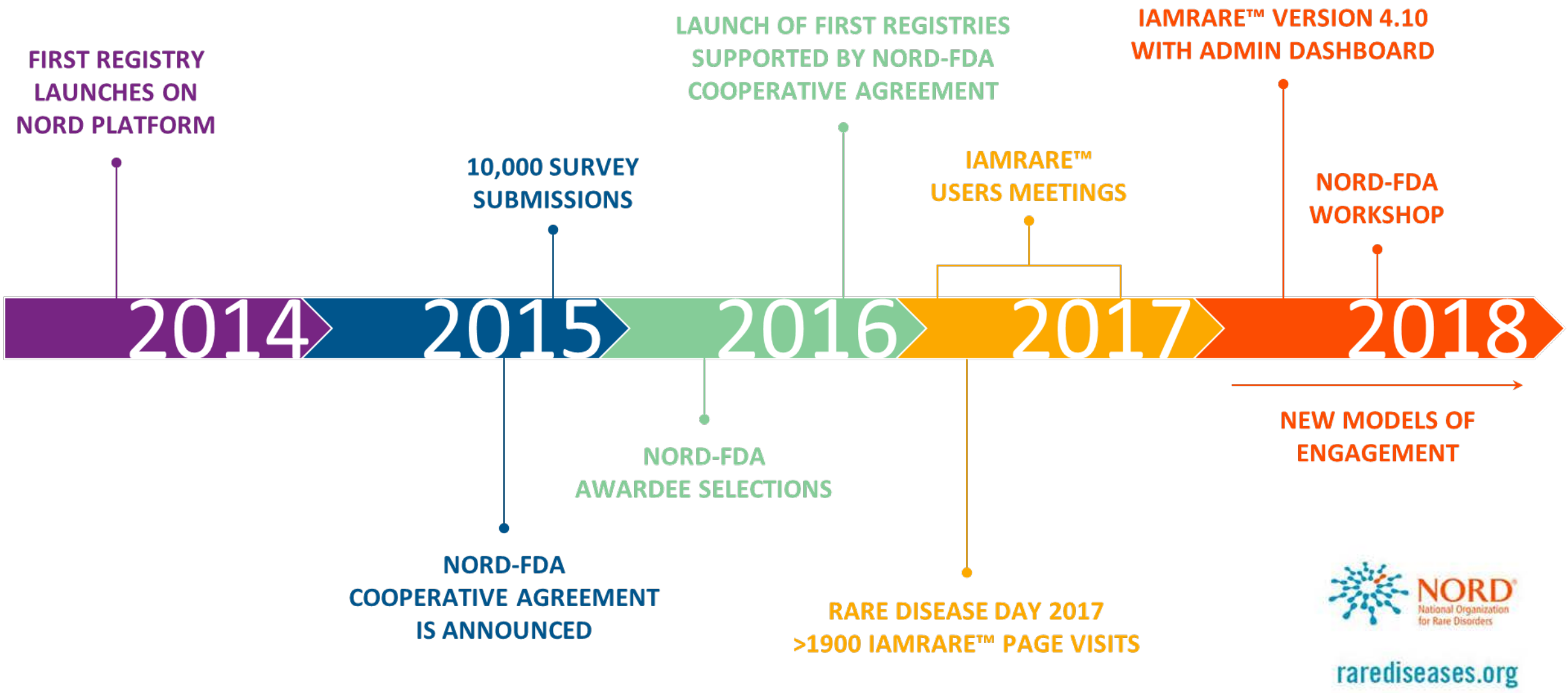
History of the IAMRARE Program



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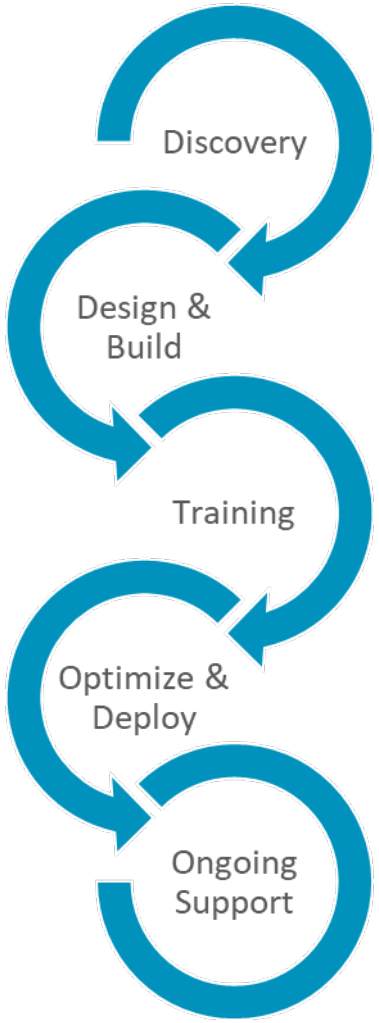
IAMRARE Program



- IAMRARE™ Platform
  - Training, User Guides
- Study Resources
  - Core Survey Library
    - Custom Survey Support
  - Templates (e.g. consent, marketing)
  - IRB Partnership
- IAMRARE™ Community
  - Portal, Meetings, Webinars, Newsletters, Videos

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IAMRARE Registry Community Members

- Arachnoiditis
- Autoimmune Polyglandular Syndrome type 1 (APS-1, APECED)
- Cat Eye Syndrome
- Congenital Central Hypoventilation Syndrome (CCHS)
- Congenital Hyperinsulinism (CHI)
- Dercum's Disease
- Desmoid Tumors
- Fibrous Dysplasia and McCune–Albright Syndrome (FD/MAS)
- Galactosemia
- Idiopathic Thrombocytopenic Purpura (ITP)
- Leukodystrophy
- Moyamoya
- Necrotizing Enterocolitis (NEC)
- Opsoclonus Myoclonus Syndrome (OMS)
- Organic Acidemia
- Pemphigus and Pemphigoid
- Phenylketonuria (PKU)
- Prader-Willi Syndrome
- SMARD 1
- SPG47
- Syngap1
- Syringomyelia & Chiari
- Tyrosinemia Type 1
- Undiagnosed
- Von Hippel Lindau
- 17q12

Example question sets - PKU

- About participant - demographics
- Diagnosis – date and type, NBS, PAH mutations, family relationships/history
- Treatment – age at PKU diet, diet start/stop, medical foods, adherence, Phe levels, Tyrosine levels, complete metabolic blood tests, essential fatty acids, supplement use
- Medical history –physical function and activity, vaccinations, serious illness history, neurological, dermatological, ADD/ADHD, autism, psychiatric, bone density, oral health, stress, anxiety, depression
- Insurance information and medical costs
- Education – family, educational services/assistance
- Mood - Hospital Anxiety and Depression Scale
- Maternal – pregnancy history, birth history and assessments

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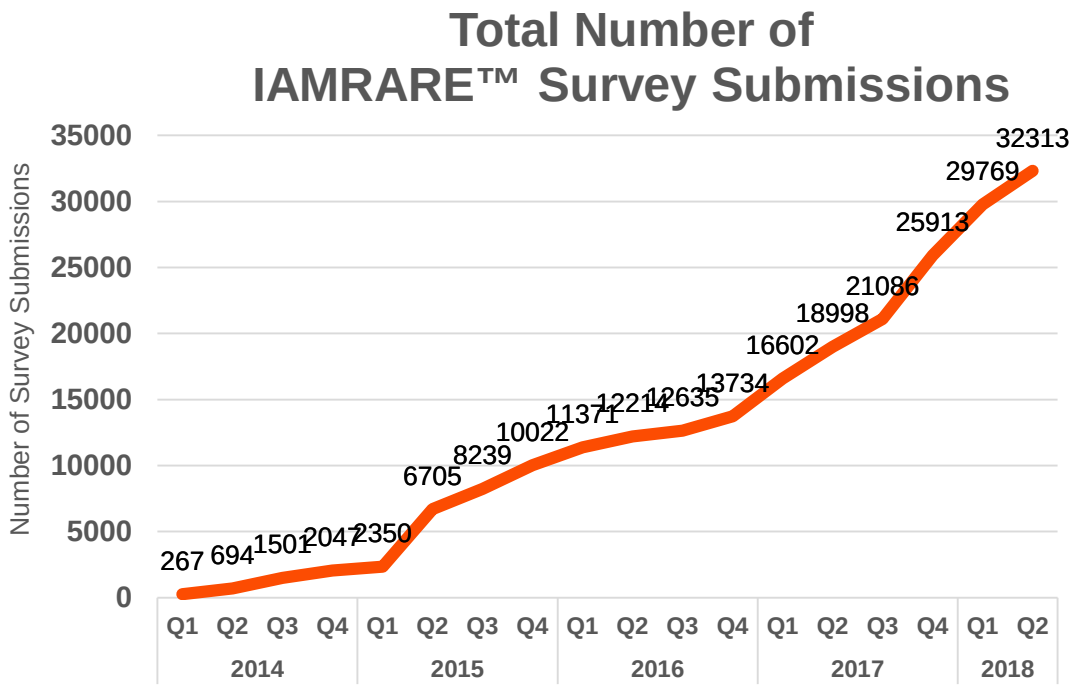
IAMRARE Community Snapshot

- 34 Registry Partnerships
  - Live/Development
- 8,500+ Users; 90% active
  - Respondents
  - Data Curators
  - System Administrators
- 45,000+ Survey Submissions



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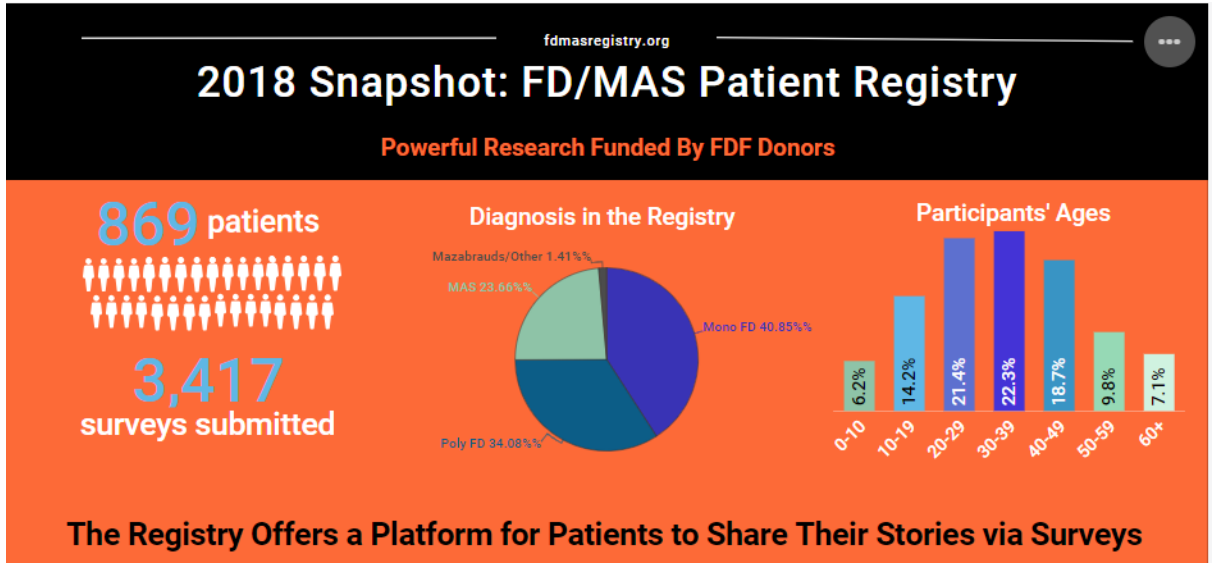
IAMRARE Community Impact

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

Sheldon D. Michaelson[1,7], Emin D. Ozkan[1,7], Massimiliano Aceti[1,6], Sabyasachi Maity[1], Nerea Llamosas[1], Monica Weldon[2], Elisa Mizrachi[1], Thomas Vaissiere[1], Michael A. Gaffield[3], Jason M. Christie[3], J. Lloyd Holder Jr.[4], Courtney A. Miller[1,5] and Gavin Rumbaugh[1,5]*



Platelet Disorder Support Association
20 Years
Empowering ITP Patients

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Partnerships and Models of Engagement

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Partnership Models

NORD Announces Ten New Rare Disease Registries, Thanks to Multi-Year Grant from Shire

Posted by Laura Mullen

Washington, D.C., September 11, 2018—The National Organization for Rare Disorders (NORD), the leading independent nonprofit organization representing the 30 million Americans with rare diseases, has announced the addition of ten new rare disease registries, made possible through a multi-year grant award from Shire.

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.

PATH for PWS™

Paving the way for Advances in Treatments & Health for PWS

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.

NORD Rare Summit

a new era of patient-focused innovation

RARE DISEASES & ORPHAN PRODUCTS BREAKTHROUGH SUMMIT®

NORD-TRIO HEALTH PARTNERSHIP

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Trio Health

NORD is pleased to present, in partnership with Trio Health, new findings from the **IAMRARE™ Patient Registry Program**, in collaboration with:

The International Pemphigus and Pemphigoid Foundation, ippf International Pemphigus & Pemphigoid Foundation

The National PKU Alliance, National PKU Alliance

The OMS Life Foundation OMS Life Foundation

Opportunities to Engage

- As registry partners
- As data partners
- As research project collaborators

- Speak at a NORD event
- Join NORD's Scientific and Medical Advisory Committee
- Write or review rare disease reports
- And more!

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NORD-FDA Patient Listening Sessions

- The FDA "Request to Connect" portal is now live: www.fda.gov/requesttoconnect
- This new portal is an opportunity for the patient and caregiver community to submit a question or request a meeting with the FDA in order to share their disease experience or better understand FDA's regulatory work.
- Requests for the rare disease listening sessions co-hosted by NORD and the FDA can also be submitted through the portal!

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Get Connected at NORD's Major Events

LIVING RARE
LIVING STRONGER™
NORD® PATIENT
AND FAMILY FORUM

featuring the
Rare Impact
2019 Awards

HOUSTON ✪ JUNE 21-23, 2019

NORD®
Rare Summit®

Washington, DC
October 21-22 2019

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Vanessa Boulanger
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National Organization for Rare Disorders (NORD)

Questions?

Email: VBoulanger@rarediseases.org or Research@rarediseases.org

NORD homepage: https://rarediseases.org/

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Thank you

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