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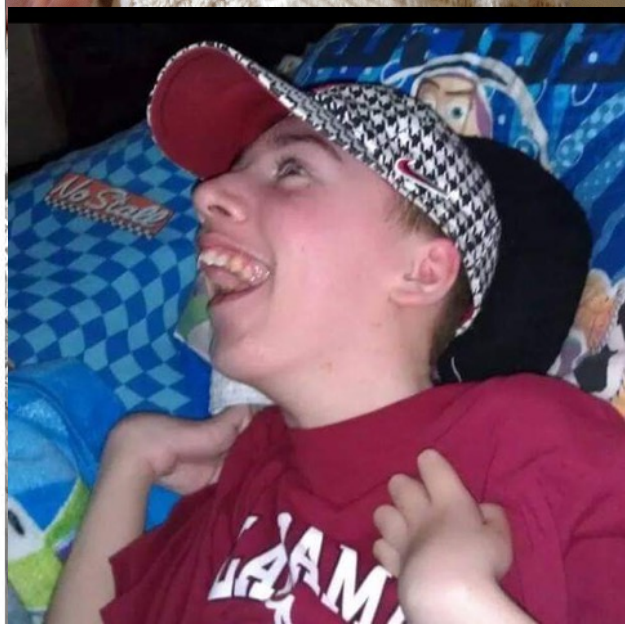
KrabbeConnect
CONNECTING PATIENTS & RESEARCH

Advancing Research Through the Krabbe **C**ommunity **U**nited **R**esearch and **E**ngagement **S**tudy (KrabbeCURES)



Presented by: KrabbeConnect and NORD

December 15, 2020

Alone we are rare. Together we are strong.®

Overview

- Background and Overview of KrabbeConnect
- Distinction between GLIA-CTN and KrabbeCURES
- Background and Overview of NORD
- NORD IAMRARE™ Platform
- The Importance of KrabbeCURES
- Getting involved in KrabbeCURES

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KrabbeConnect

- KrabbeConnect incorporated on Feb 5th, 2018
- The idea for KrabbeConnect originated from the 2015 Family Centered-Krabbe Translational Research Network meeting (FC-KTRN), a collaborative meeting between researchers and families to aid in solving the uncertainties of Krabbe disease.
- KrabbeConnect, provides a platform to amplify the voice of patients, aiding researchers and drug developers in accelerating research for better treatments for Krabbe disease.
- KrabbeCURES came to fruition as a result of many conversations, since inception, with stakeholders in Krabbe disease. The Krabbe disease community needed an effective tool to fill gaps in research. KrabbeConnect's board of directors selected the IAMRARE platform at NORD.

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Studies, Registries, and Survey's-Oh My!

Collection of information about individuals

- Specific disease or diagnosis
- General patient demographics
- Genetic Information
- Patient-reported outcomes
- Comprehensive medical data
- Clinical trial matching

Main types of registries

- Contact registries
- Patient registries

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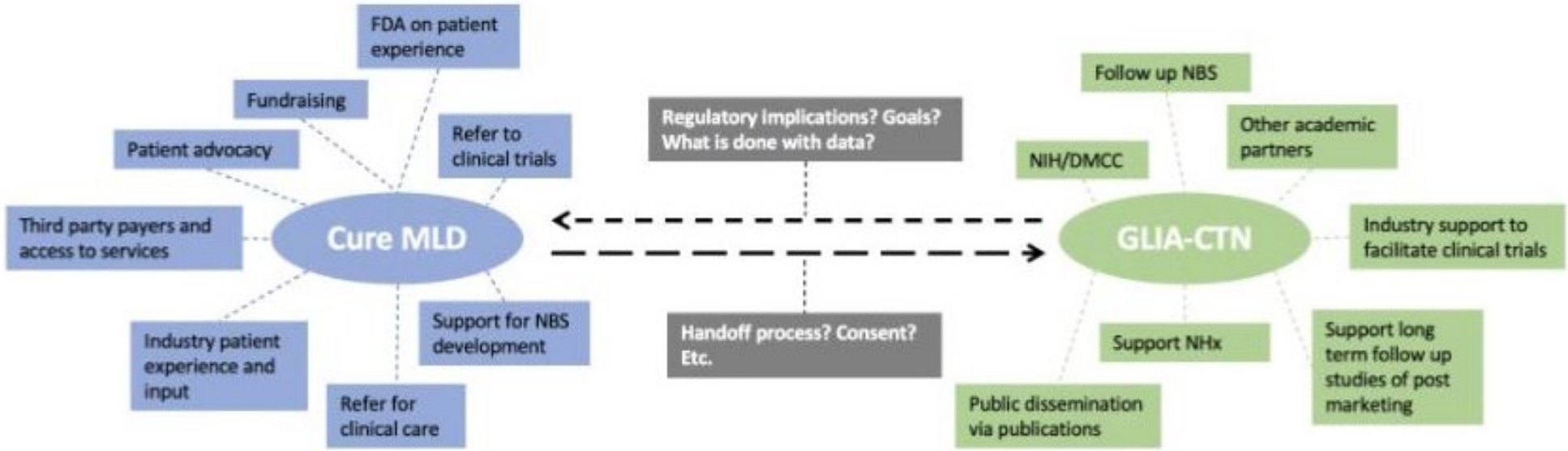
GLIA-CTN Registry



Very **different data** being accumulated by **GLIA-CTN** versus **KrabbeCURES**.

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Aim of KrabbeCURES

To be a **data-collection study for patients** around the world to share information about globoid cell leukodystrophy (Krabbe disease) **that will fill current and future gaps in research**.

- Understand the burden of Krabbe disease
- Provide a convenient online platform for participants (or caregivers) to self-report cases of globoid cell leukodystrophy (Krabbe disease).
- Characterize and describe the globoid cell leukodystrophy population as a whole, enhancing the understanding of disease prevalence and phenotype as well as the rate of progression of disease characteristics.
- Develop a communications study within KrabbeCURES (e.g., to notify patients of research studies and clinical trials).
- Assist the globoid cell leukodystrophy community with the development of recommendations and standards of care.
- Be a case-finding resource to be used for researchers who seek to study the pathophysiology of Krabbe disease, retrospectively collate intervention outcomes, and design prospective trials of novel treatments.

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About KrabbeCURES

Launched on **August 27, 2020** and includes surveys to capture data on:

- Demographics
- Medical, Diagnostic and Treatment Data
- Quality of Life
- Developmental Milestones
- Therapy
- Symptoms and side effects

Principal Investigator:

- Vanessa Boulanger, NORD

Co-Investigator:

- Stacy Pike-Lagenfeld, KrabbeConnect

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Important Insights about KrabbeCURES

1. It's IRB approved. This means informed consent is obtained from participants to allow the participants de-identified data to be utilized for future research.

2. The accumulation of quality data under an IRB will produce peer-reviewed publications. Peer-reviewed articles have become the trusted form of scientific communication to stakeholders (industry, NBS committees, FDA, researchers, etc.).

3. It's representative of the patient voice because it's patient reported data/results.

4. Krabbe disease community owns the engagement study and will be available to other researchers, nonprofit org., academic institutions, etc.

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NORD, an independent nonprofit, is leading the fight to improve the lives of **rare disease patients and families.**

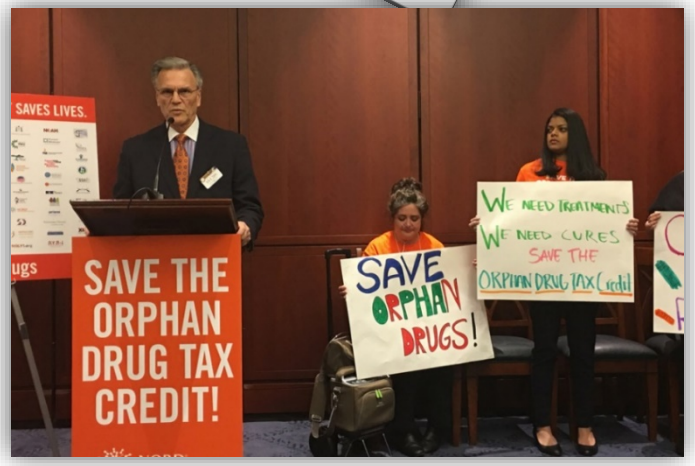
We do this by supporting patients and organizations, accelerating research, providing education, disseminating information and driving public policy.

The Orphan Drug Act

- From 1967 to 1983, only 34 drugs approved by the Food and Drug Administration (FDA) were for rare diseases.

- Only 10 of the products brought to market by the pharmaceutical industry in the decade before 1983 would have qualified under today's ODA as orphan drugs.

- A task force, whose members included staff of the FDA and National Institutes of Health (NIH), considered individuals with rare diseases to be an **underserved patient community** and felt that the plight of these patients and their families was a **public health issue.**



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Core Programs & Services

RESEARCH

- Research grants for translational or clinical studies
- IAMRARE™ Registry Platform & NHS 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

MEMBERSHIP SERVICES

- Advocacy
- Capacity Building
- Mentorship & Education
- Visibility & Credibility

EDUCATION

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

POLICY AND ADVOCACY

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

Our Stakeholders

PATIENTS & FAMILIES

Undiagnosed
Newly Diagnosed
Caregivers

MEDICAL PROFESSIONALS

LEGISLATORS

House & Senate
HELP Committee
Ways & Means Committee
Energy & Commerce Committee

INDUSTRY

Biotechnology
Pharmaceutical
Research Organizations
Service Providers

PATIENT ORGANIZATIONS

Research Foundations
Disease-Specific Advocacy Organizations
Support Groups

PAYORS

Pharmacy Benefit Managers (PBMs)
Private Insurers
Medicaid/Medicare

REGULATORS

Food and Drug Administration (FDA)
National Institutes of Health (NIH)
Centers for Medicaid and Medicare (CMS)
Social Security Administration (SSA)

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Accelerating Research

> The IAMRARE team at NORD is absolutely committed to helping rare disease patient organizations run low-cost, high-quality natural history studies. NORD's understanding of the needs, goals and challenges of a small nonprofit like ours is in a league of its own.
>
> - Member, Fibrous Dysplasia Foundation

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Challenges for Rare Patients

- Little known about disease progression and burden
- Few medical experts
- Delays in diagnosis
- Limited FDA-approved treatments
- Extensive, life-long medical needs
- High cost of care and treatment
- Small, scattered patient populations
- Social isolation

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Core Research Objectives

- Advancing basic and translational research for rare diseases.
- Supporting data collection for discovery, review and approval of new treatments and orphan products.
- Aid researchers with clinical trial design, endpoints and biomarkers.
- Original research and publication to inform legislative and regulatory decision-makers on the challenges and opportunities in rare diseases.
- Empower patients to contribute to research for their disease.
- Galvanize and support organizations in launching research grant programs and natural history studies.

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Marcia Galan and her daughter, Alena, diagnosed with Mucopolysacchardosis type VI (MPS VI)

The IAMRARE™ Platform

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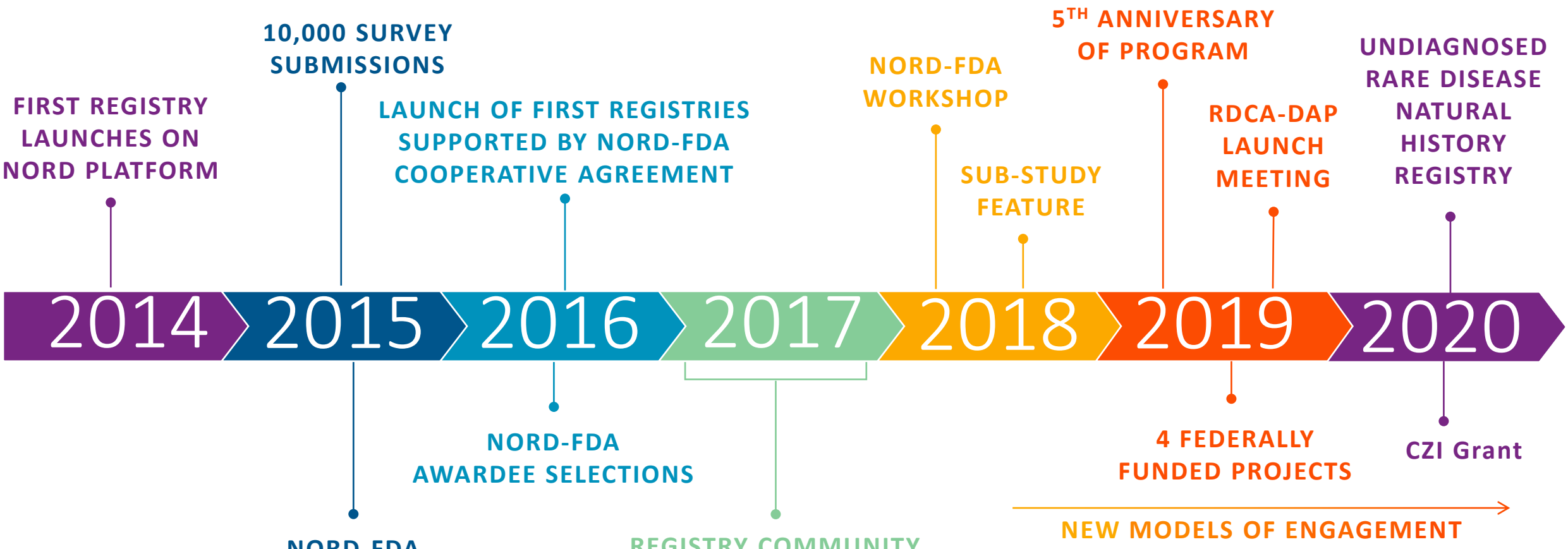
History of the IAMRARE Program



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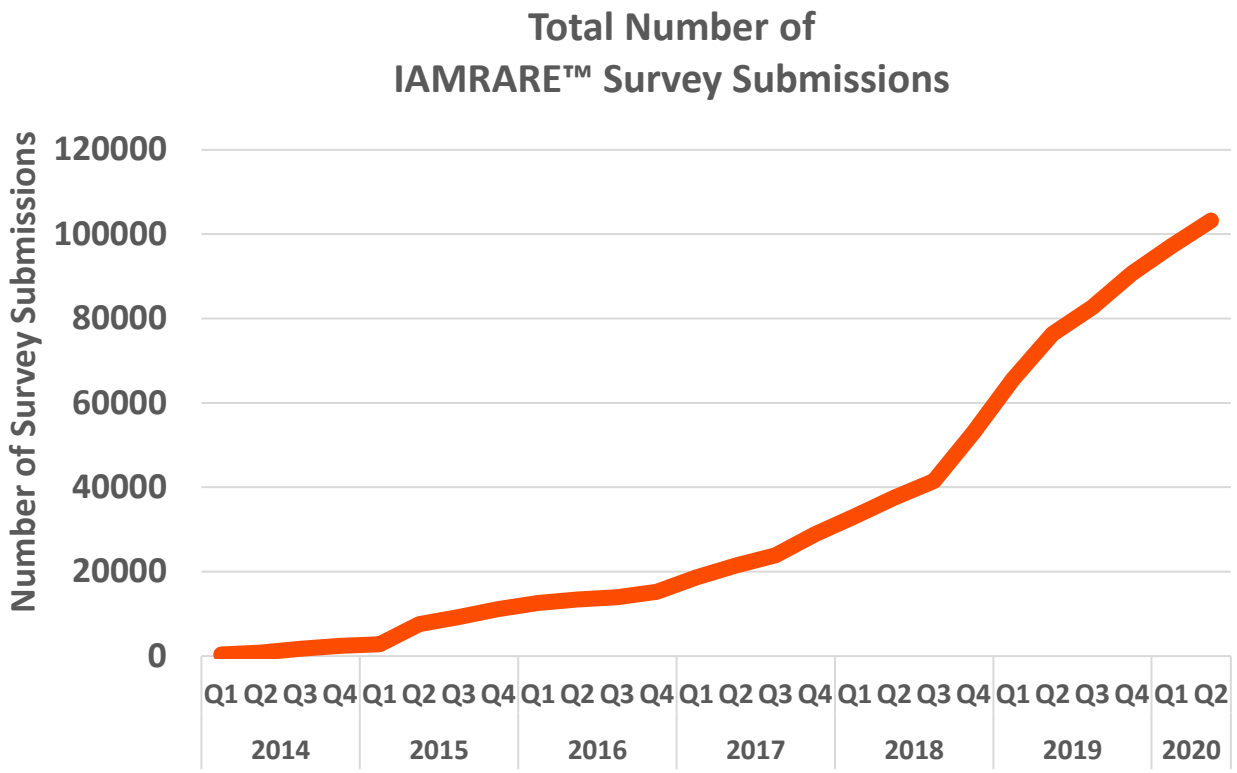
IAMRARE Platform

Since **2014** Launch:

40+
Rare Conditions

11,000+
Participants

100,000+
Survey Submissions



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Using the Data

Data collected through the IAMRARE Platform has been presented through many venues and formats:

- Patient/family conferences
- Medical conferences
- Webinars
- Patient Focused Drug Development meetings

- Publications
  - Medical journal articles
  - Peer-reviewed papers
- Posters
- Community Reports
- Research collaborations

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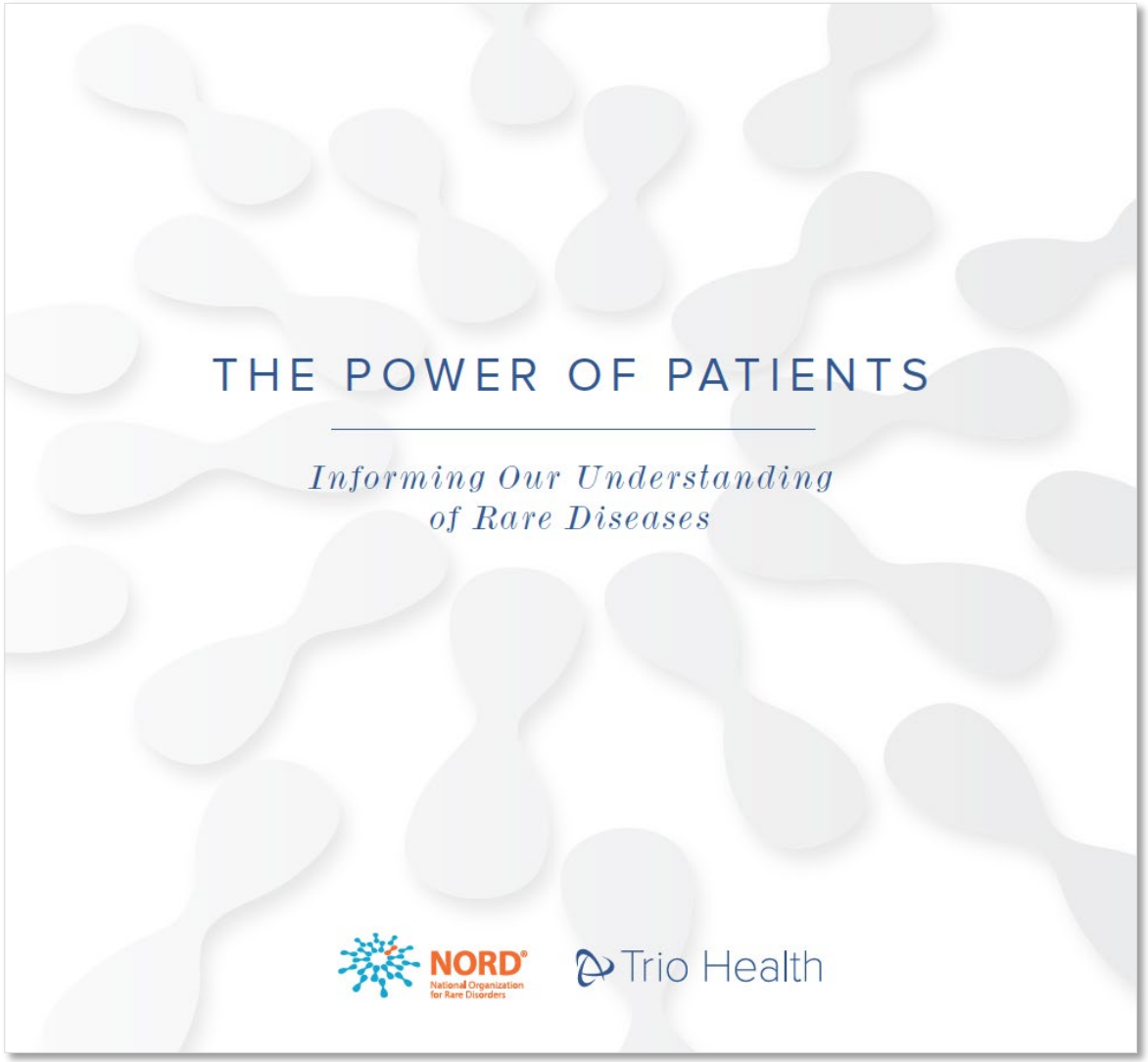
The Power of Patients



The Power of Patients *amplifies the experiences of individuals and families living with rare diseases and demonstrates the importance of registries and natural history data in helping to shape the rare disease landscape.*

National PKU Alliance

ippf International Pemphigus & Pemphigoid Foundation

DTRF THE DESMOID TUMOR RESEARCH FOUNDATION

Platelet Disorder Support Association
Empowering ITP Patients

Bridge the Gap SYNGAP Education & Research Foundation

OMS life Foundation

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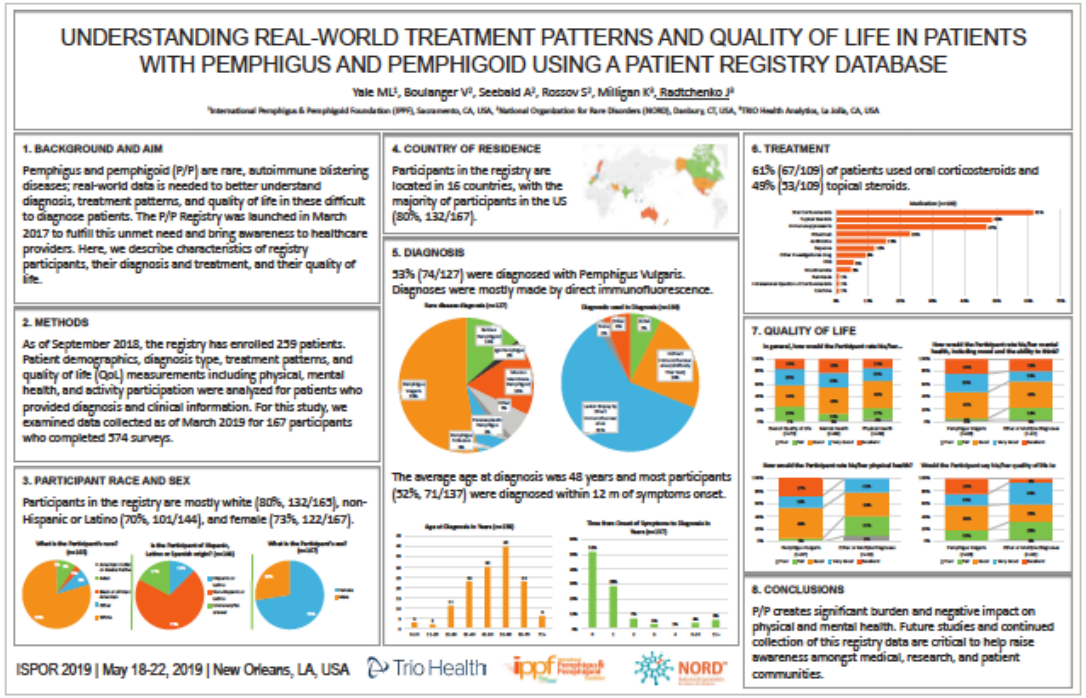
The Power of Patients

Pemphigus and Pemphigoid |



UNDERSTANDING REAL-WORLD TREATMENT PATTERNS AND QUALITY OF LIFE IN PATIENTS WITH PEMPHIGUS AND PEMPHIGOID USING A PATIENT REGISTRY DATABASE

Yale ML[1], Boulanger V[2], Seebald A[3], Rossov S[3], Milligan K[3], Radtchenko J[3]

[1]International Pemphigus & Pemphigoid Foundation (IPPF), Sacramento, CA, USA, [2]National Organization for Rare Disorders (NORD), Danbury, CT, USA, [3]TRIO Health Analytics, La Jolla, CA, USA

1. BACKGROUND AND AIM

Pemphigus and pemphigoid (P/P) are rare, autoimmune blistering diseases; real-world data is needed to better understand diagnosis, treatment patterns, and quality of life in these difficult to diagnose patients. The P/P Registry was launched in March 2017 to fulfill this unmet need and bring awareness to healthcare providers. Here, we describe characteristics of registry participants, their diagnosis and treatment, and their quality of life.

2. METHODS

As of September 2018, the registry has enrolled 259 patients. Patient demographics, diagnosis type, treatment patterns, and quality of life (QoL) measurements including physical, mental health, and activity participation were analyzed for patients who provided diagnosis and clinical information. For this study, we examined data collected as of March 2019 for 167 participants who completed 374 surveys.

3. PARTICIPANT RACE AND SEX

Participants in the registry are mostly white (80%, 132/165), non-Hispanic or Latino (70%, 101/144), and female (73%, 122/167).

4. COUNTRY OF RESIDENCE

Participants in the registry are located in 16 countries, with the majority of participants in the US (80%, 132/167).

5. DIAGNOSIS

53% (74/127) were diagnosed with Pemphigus Vulgaris. Diagnoses were mostly made by direct immunofluorescence.

The average age at diagnosis was 48 years and most participants (52%, 71/137) were diagnosed within 12 m of symptoms onset.

6. TREATMENT

61% (67/109) of patients used oral corticosteroids and 49% (53/109) topical steroids.

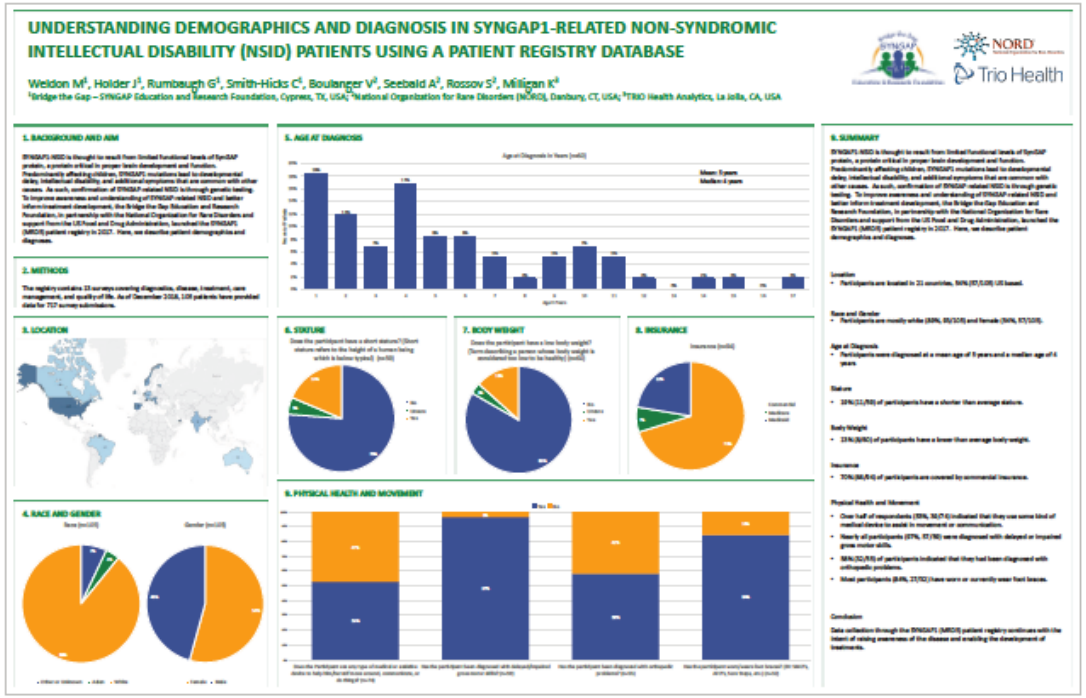
7. QUALITY OF LIFE

8. CONCLUSIONS

P/P creates significant burden and negative impact on physical and mental health. Future studies and continued collection of this registry data are critical to help raise awareness amongst medical, research, and patient communities.

ISPOR 2019 | May 18-22, 2019 | New Orleans, LA, USA

| *SYNGAP1-Related NSID*



UNDERSTANDING DEMOGRAPHICS AND DIAGNOSIS IN SYNGAP1-RELATED NON-SYNDROMIC INTELLECTUAL DISABILITY (NSID) PATIENTS USING A PATIENT REGISTRY DATABASE

Weldon M[1], Holder J[1], Rumbaugh G[1], Smith-Hicks C[1], Boulanger V[2], Seebald A[2], Rossov S[2], Milligan K[2]

[1]Bridge the Gap – SYNGAP Education and Research Foundation, Cypress, TX, USA; [2]National Organization for Rare Disorders (NORD), Danbury, CT, USA; [3]TRIO Health Analytics, La Jolla, CA, USA

1. BACKGROUND AND AIM

2. METHODS

3. LOCATION

4. RACE AND GENDER

5. AGE AT DIAGNOSIS

6. STATURE

7. BODY WEIGHT

8. INSURANCE

9. PHYSICAL HEALTH AND MOVEMENT

9. SUMMARY

The Power of Data from KrabbeCURES

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KrabbeCURES Data will

- **Inform** patient care and best practices
- **Assess** patient and caregiver experiences and preferences
- **Contribute** to disease understanding
- **Identify** research priorities such as genetic, molecular and physical basis of rare diseases
- **Estimate** the number of affected patients and patients potentially available to participate in research
- **Evaluate** the individual and global economic burden of disease
- **Inform** drug development
- **Provide** an avenue for bio-specimen collection

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The Power of KrabbeCURES

Patient Organizations

- Research partners and leaders
- New models of engagement
  - New members/collaborations
  - Expand/strengthen existing partnerships
  - Unite divided communities

Industry

- Collaborating with patient groups and supporting meaningful patient centered product development
- Models of partnerships
- Pair registry data with clinical trial data
- Clinical trial recruitment

Patients, Families & Caregivers

- Access to data
- Insights into patient outcomes
- Symptom tracking
- Resources for conversations with clinicians, educators

Clinicians

- Data as a reference point for patient reported outcomes (PRO)
- Access to patient cohorts
- Registry data to supplement existing clinical trial data
- Document conditions (no data, progression, transitions in care)
- Inform new and revised standards of care

· USER FEEDBACK ·

What impact has your natural history study had on your organization and/or the community at large?

- ✓ *It has empowered our community to know that they have the power to influence research and knowledge about our disease.*
- ✓ *It has created excitement and hope within our community. Patients are beginning to understand that they have the ability to influence research.*

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The Power of KrabbeCURES

Researchers

- Compare and validate experimental findings with PRO data
- Use trends from preliminary data to explore new areas of a disease
- Provide real world evidence when applying for research grants
- Inform product development
- Regulatory approvals
- Validate measures
- Establish new research protocols

Regulatory Agencies

- Registry data to accelerate and improve research
- Outcomes from clinical trials
- Transitions in care
  - Pediatric to adult
  - Treatment protocols
- Barriers to adherence and or compliance to treatment
- Contextualize clinical trial data
- New models of engagement
  - New members/collaborations
  - Expand/strengthen existing partnerships
  - Unite divided communities

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How can you get involved with KrabbeCURES?

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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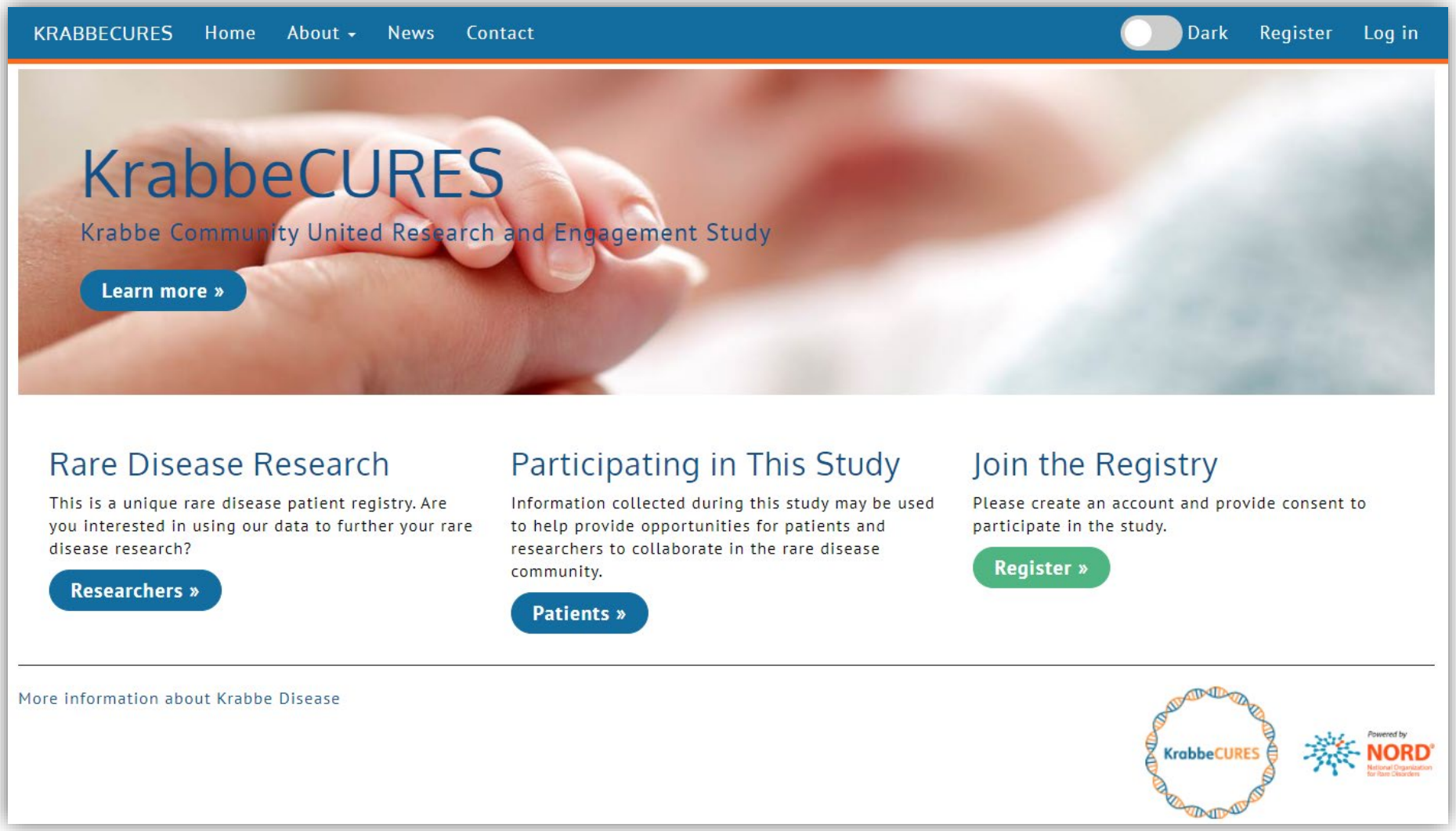
krabbecures.iamrare.org



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KrabbeCURES Resources

- **Informational Brochures**
- **User guides**
- **FAQ Document**
- **Study Staff**

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Learn more at krabbeconnect.org

KrabbeCURES

Be the first to make a difference. Sign up today to be a part of KrabbeCURES.

Join the initiative at krabbecures.iamrare.org

Become a Partner

Are you looking to help make a difference for patients and families living with Krabbe disease? KrabbeConnect wants to collaborate with you! **LEARN MORE**

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Questions?

- For information about **KrabbeCURES** go to

  krabbecures.iamrare.org

  Or email info@krabbeconnect.org

- For information about **KrabbeConnect** go to

  Krabbeconnect.org

  Or email info@krabbeconnect.org

- For information about **NORD** go to

  https://rarediseases.org/

  Or email orphan@rarediseases.org

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