# A 35 Year Journey: Advocating for the Rare Disease Community

Suzanne Rossov

Research Programs Manager

National Organization for Rare Disorders

Alone we are rare. Together we are strong.®

**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.

# NORD's Reach


NORD's primary website receives **14 million hits** per year.

More than **8.2 million people** turn to NORD for information on rare diseases.

Visitors come from more than **215 countries**, the top 5 being the U.S., U.K., Canada, Australia and India.

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# Core Programs & Services

## Education

- Medical Professional Education
- Medical and PharmD Student Education & University Chapter Program
- Patient and Caregiver Education
- Annual Rare Diseases & Orphan Products Breakthrough Summit

## Patient Services

- Insurance Navigation
- Co-Pay, Premium and Medication Assistance
- Emergency Relief
- Ancillary Services
- Clinical Trial Travel & Lodging

## Membership Services

Advocacy, Capacity Building, Connections, Mentorship, Education, Visibility, Credibility

## Policy & Advocacy

- Advancing Basic and Translational Science
- Strengthening Drug & Device Development
- Ensuring Access to Affordable Treatment
- Ensuring Access to Safe, Effective Medical Care

## Research

- IAMRARE™ Registry Program
- Rare Disease Research Grant Program
- Clinical Trial Awareness & Education
- Data Collection & Analysis for Original Research

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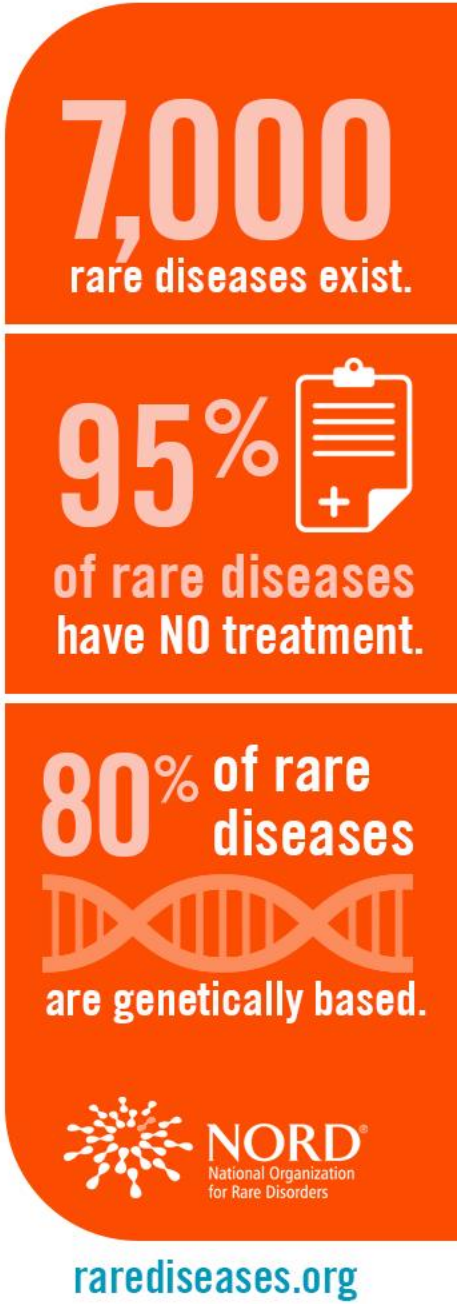
# Challenges for Rare Patients

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



# Education Initiatives

NORD provides educational resources for patients, caregivers, medical professionals, students and the public.

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# NORD RareEdu®

- Webinars
  - Free to the public
- Patient & Caregiver videos
  - “Gene Therapy: Your Questions Answered”
    - Patient organizations are translating to other languages

- Rare Disease Database
  - 85% of the nearly 1 million monthly visitors to NORD’s website
  - Grant from the Anthem Foundation
    - 45 new reports for NORD’s Rare Disease Database
    - Enhance search engine functionality

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# Educating Future Medical Professionals

- 10 NORD Chapters & 9 NORD Rare Disease Clubs at universities and medical schools
- NORD is partnering with “Osmosis” to develop short, animated videos about rare diseases for medical education
- Bi-monthly student newsletter promotes news about Rare Disease Day & other events

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# Patient Services

NORD pioneered Patient Assistance Programs in the U.S., launching the first program in 1987. Today, **RareCare®** serves **over 7,000 patients annually**

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# RareCare®

31+ years of white glove assistance with over 1.5 million patients served

- Across the service continuum
  - Access to treatment
  - Travel programs
  - Consultations
  - Diagnostic testing


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# RareCare®

- Across the disease continuum
  - Clinical trials
  - Medication and medical foods
- Technology investment
  - Electronic services
  - RareAccess™ Portal
  - RareCare® 5th generation case management system
- Human capital investment
  - Leadership and staff

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BATTEN DISEASE℠ Support and Research Association

Celebrating NORD 35TH ANNIVERSARY

# Membership Services

- *While we can't always be present for all of the wonderful projects NORD creates for its members knowing that they are there, and that we have a wide context to work from is empowering. I always leave NORD's Summit as a new person.*
- *Thank you for all that you do for us. We are so proud to be NORD members*
  - *- PSC Partners Seeking a Cure*

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# Patient Organization Members


- **280+** nonprofit disease specific patient organizations with missions that align with NORD's and meet pre-determined criteria
- Networking & Community engagement opportunities
  - 1200 patient advocacy organizations in NORD's Organizational Database
- Key Membership Benefits:
  - ✓ Peer networking and guidance
  - ✓ Support promoting and managing research grants
  - ✓ Educational opportunities – live and virtual
  - ✓ Collaboration on campaigns and events
  - ✓ Representation in key regulatory and legislative forums
  - ✓ Promotion of news, information and resources

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# Policy & Advocacy

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# Areas of Focus

**Research**

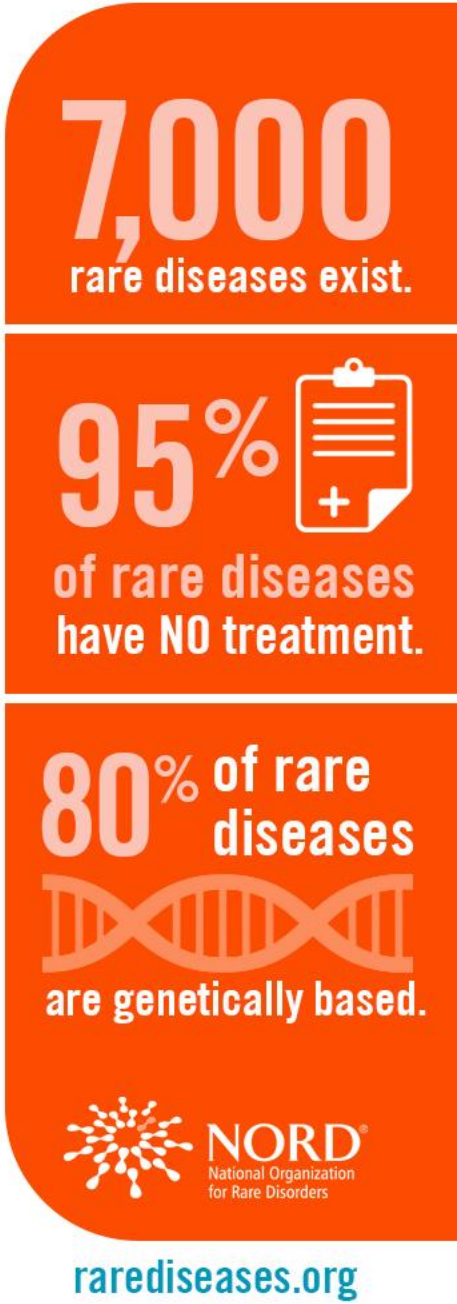
- NIH Funding
- RD Research
- Research protections and governance

**Drug Development**

- Orphan Incentives
- FDA Review of orphan products
- Medical Devices
- Patient Integration
- Expanded Access

**Access to Therapies**

- Patient Protections
- Medicaid
- Cost-sharing
- Newborn Screening
- Benefit Design



# Policy Priorities 2018

## Federal

- Protecting the Orphan Drug Act
- Defending Insurance Patient Protections and Medicaid Coverage
- Stabilizing the Insurance Marketplaces
- Implementing the 21st Century Cures Act, FDARA, the Orphan Drug Modernization Plan, and the Office of Patient Affairs
- Enacting the Medical Nutrition Equity Act
- Improving Access to Pre-Approval Therapies
- Encouraging Responsible Value Assessments
- Opposing harmful “welfare reform”
- Supporting Patient Assistance Programs
- And more!

## State

- Rx Out-of-Pocket Costs
- State Rare Disease Advisory Councils
- Health Insurance Protections and Affordability
- Medicaid and CHIP
- Coverage for Medical Nutrition
- Expansion of Newborn Screening
- EMS/ER rare disease treatment protocols

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# Policy Status Report 2018

## Federal

- Sent or Joined >15 letters to Congress
- 40 Comment documents/letters to the Administration
- Tracks over 25 pieces of legislation on Capitol Hill

## State

- >85 Bills tracked
- 36 State Ambassadors/Community Engagement Liaisons across 28 states
- RAN members in all 50 states

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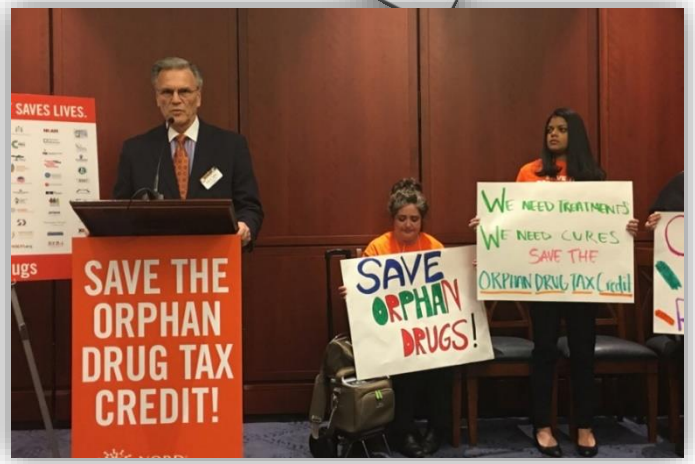
# The Orphan Drug Act

- 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.**


NORD was founded in **1983** along with the Orphan Drug Act

**260+** NORD member organizations (& growing each year)

**3,000+** RareAction® advocates across the country

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# NORD® RARE ACTION NETWORK®

The mission of the Rare Action Network℠ (RAN) is to connect and empower a unified network of individuals and organizations with tools, training, and resources to become effective advocates for rare diseases through national and state based initiatives across the United States.

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**rareaction.org**

# Rare Action Network

- **RareAction.org**
- State Ambassador Program®
- Regional meetings
- Legislative Tracker®
- Policy education
- Legislative (hill) days
- Rare Disease Day®
- Legislative action alerts
- Social media hosting
- Education & Awareness

**State Specific Components**

- NORD State Policy Report Card®
- State Action Center

**Join today!**

**RareAction.org**

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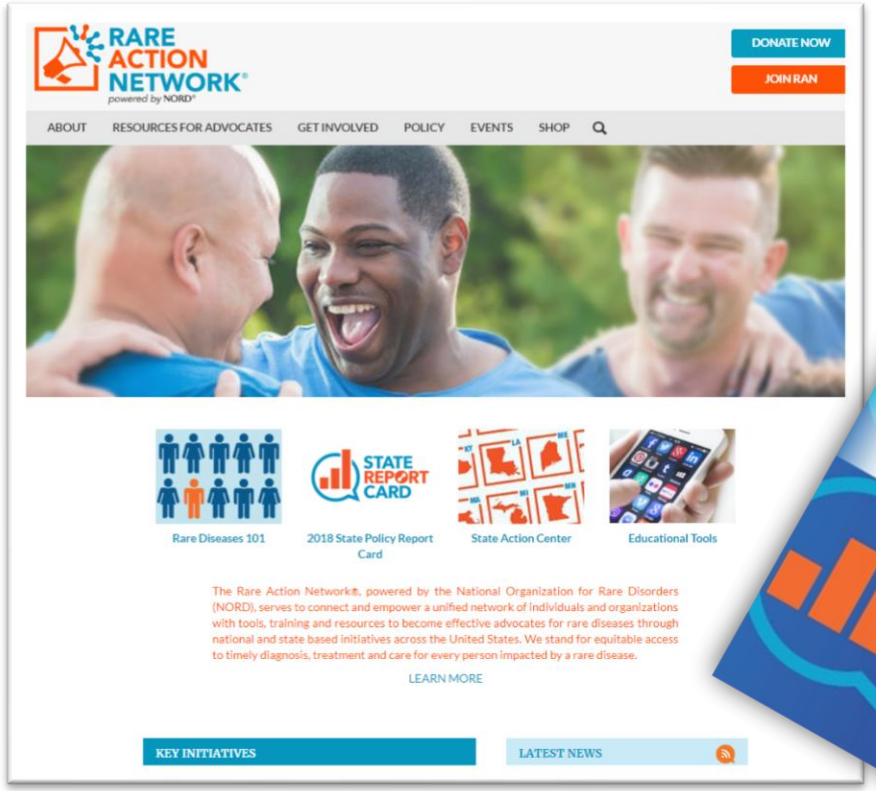
# Rare Action Online



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**RareAction.org**

**@RareAction**

# Research Initiatives

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# Core Research Objectives

- Advance basic and translational research for rare diseases
- Support data collection that can be used in the discovery, review and approval of new treatments and orphan products
- Help researchers with clinical trial design, endpoints and biomarkers
- Develop original research and publications to inform legislative and regulatory decision-makers on the challenges and opportunities in rare diseases (economic, social, political)
- Empower patients to contribute to research for their disease
- Empower and support organizations in launching research grant programs and natural history studies

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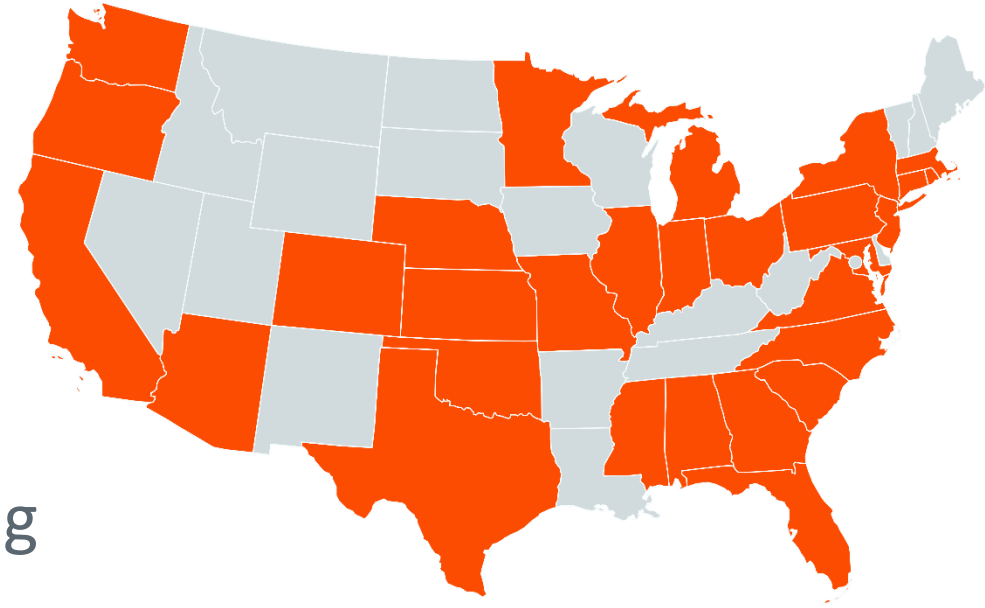
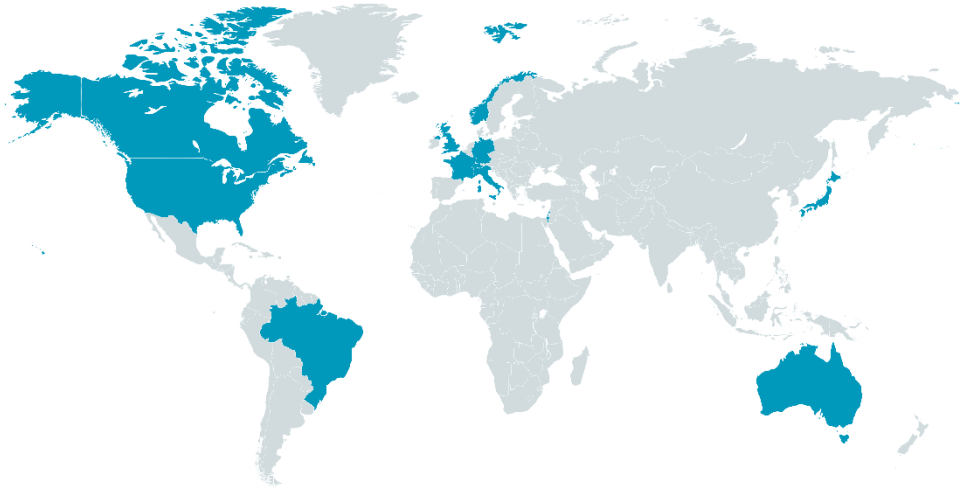
# GRANTS

- First grants awarded in 1989
- 150+ grants and fellowships
- Approx.  $7 million in approved funding
- 29 states
- 13 countries





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PROCESS

## 1 Donations Collected

NORD manages hundreds of research funds to which patients, organizations and companies contribute.

## 2 RFPs Developed & Posted

NORD develops and posts RFPs for the research grants that are available on our website, in our e-newsletter and social media.

## 3 Letters of Intent

Researchers first submit a letter of intent, reviewed by NORD's scientific advisory committee. Select researchers are then invited to submit full proposals.

## 4 Full Proposals

Final proposals are reviewed by the MAC committee, and award decisions are made.

## 5 Awards Provided

Once awards are made, researchers are required to send bi-annual updates to NORD on the status of their research.

## Success Corner

These grants have led to over 150 articles in peer-reviewed journals and at least 2 FDA-approved products.

1. Titanium Rib developed for children with rare disorders resulting in thoracic insufficiency syndrome.

2. Northera capsules for the treatment of neurogenic orthostatic hypertension (NOH)


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# Program Impact

*During this period we received critical seed grant funding from the National Organization of Rare Disorders to support our research. Based on the preliminary clinical trial results made possible by the NORD support, we were able to secure further substantial grant support from the FDA Office of Orphan Product Development that enabled us to complete our sole site FDA study and initiate a multi-center FDA VEPTR trial.*

**Robert M. Campbell, Jr., M.D.**
**Senate Testimony, March 27, 2007**

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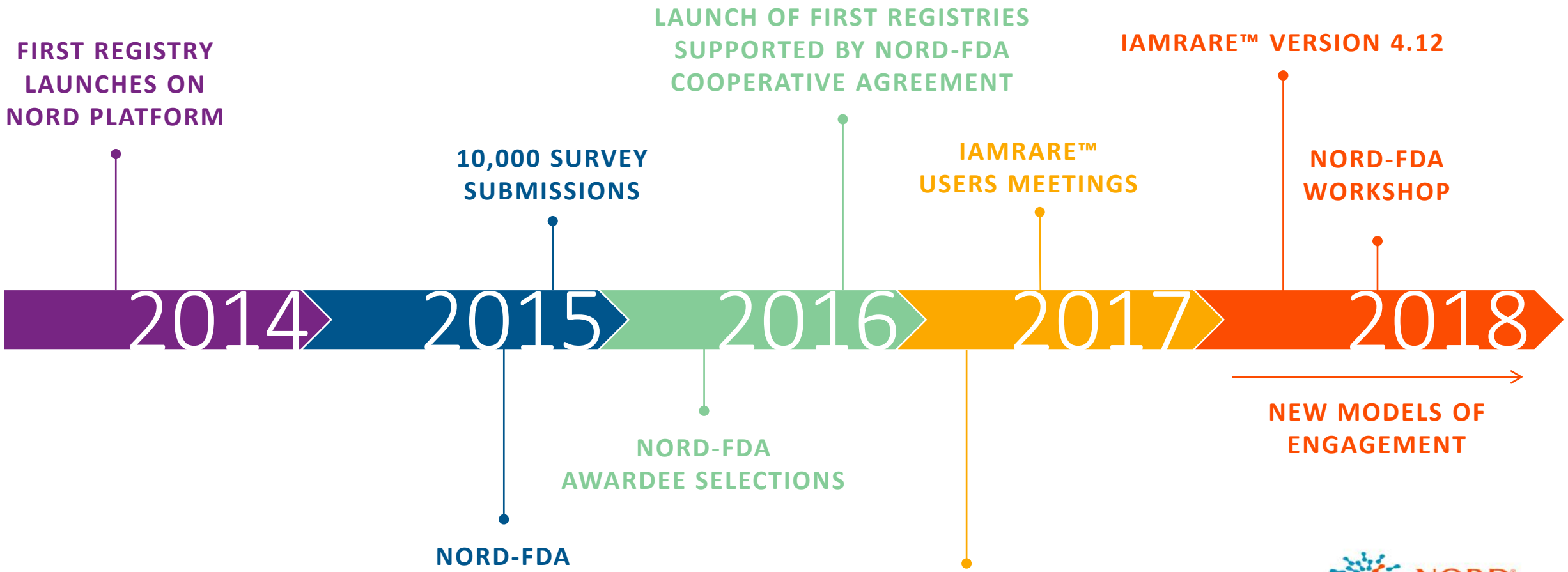
# IAMRARE™ Registry Program

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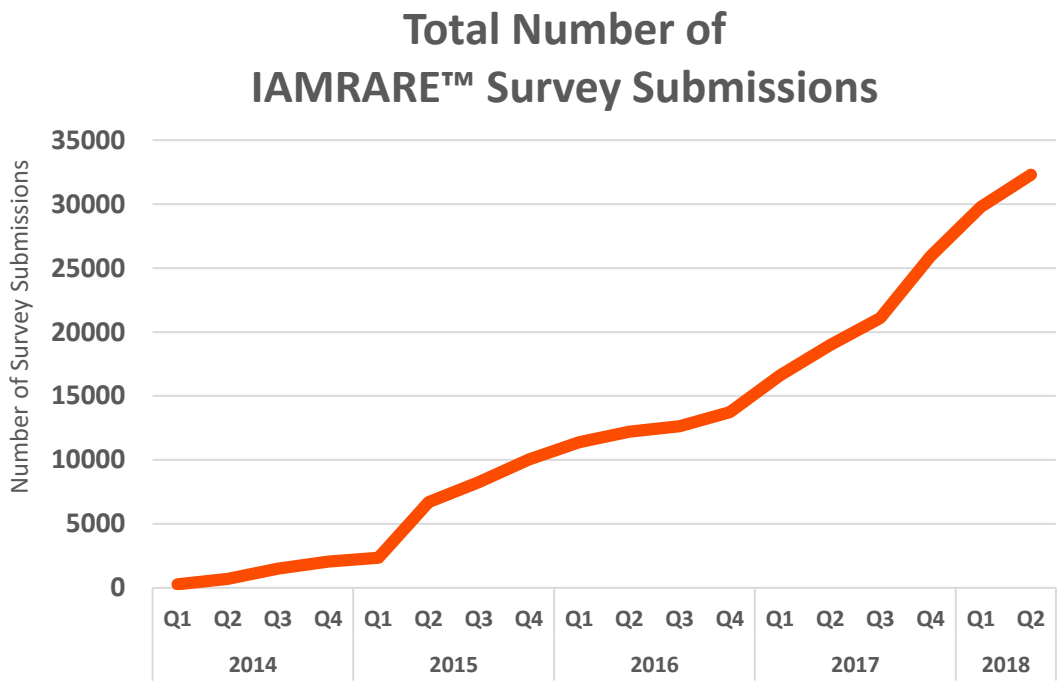
# Key Numbers for IAMRARE™

- 20 Registries (Live/Development)
  - 10+ Additional Planned
- 6,000+ Users
  - Respondents
  - Data Curators
  - System Administrators
- 30,000+ Survey Submissions

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# The Importance of Natural History Study Data

- **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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# What Your Participation Means for OMS Research


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# OMSLife Patient Registry

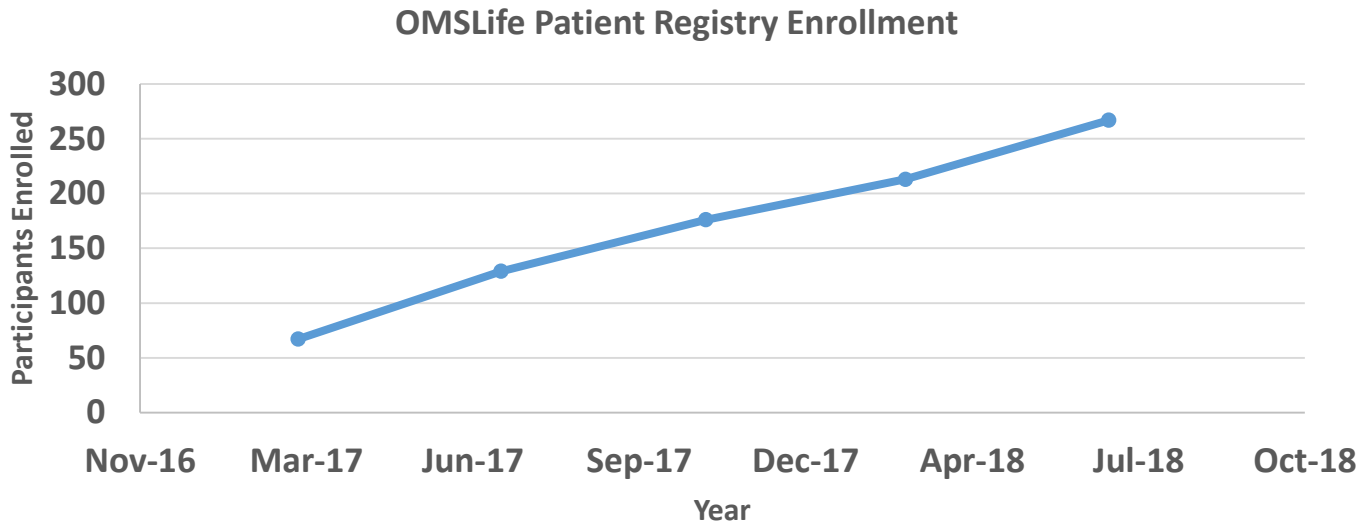
**March 2016**
OMSLife Awarded Grant from NORD

**April 2016**
Registry Development

**February 2017**
Launch of OMSLife Patient Registry

**June 2017**
Preliminary Data Analysis

**March 2018**
**Present Data at Global Conference**




## Key Enrollment Numbers

- March 2017 – 67 enrolled
- July 2017 – 129 enrolled
- August 2018 – 267 enrolled

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# Patients, Families and Caregivers

- Access to data - individual and aggregate
  - Insights into patient outcomes
  - Support for self-management (e.g. symptom tracking)
  - Evidence for insurance claims
  - Resources for conversations with clinicians, educators
    - Planning for clinical visits
    - IEPs
- Direct role in drug development & evaluation
  - Inform clinical trial endpoints
  - Provide patient preference data

**· 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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# Clinicians

- Clinicians
  - Use data as a reference point for patient-reported outcomes (PRO)
    - Targeted treatment
  - Provides alternatives when existing treatment protocols are not effective
  - Access to patient cohorts
  - Use registry data to supplement existing clinical trial data
  - Document conditions
    - No data exists or is lacking
    - Track progression
    - Transitions

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# Researchers

- 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 drug development
  - Regulatory approvals
  - Validate measures
  - Establish new research protocols

*"…With the new funding, the team plans to focus on how this Syngap1 sensitive period regulates developmental processes that link sensory processing to learning, and how harmful Syngap1 mutations may lead to autism-associated behavioral changes through sensory dysfunction…" Grant # R01 MH096847*

*The Scripps Research Institute News and Views, Vol 18. Issue 11. May 2018.*

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# Regulatory Agencies

- Use registry data to  accelerate and improve research
  - Identify areas of unmet need
  - Best practices
- Outcomes from clinical trials
- Transitions in care
  - Pediatric to adult
  - Treatment Protocols
    - No treatment to treatment; new treatment
    - Start/stop medication schedules
    - Medication cocktails
    - Side effects
- Capture barriers to adherence and or compliance to treatment protocols

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# What's Next?

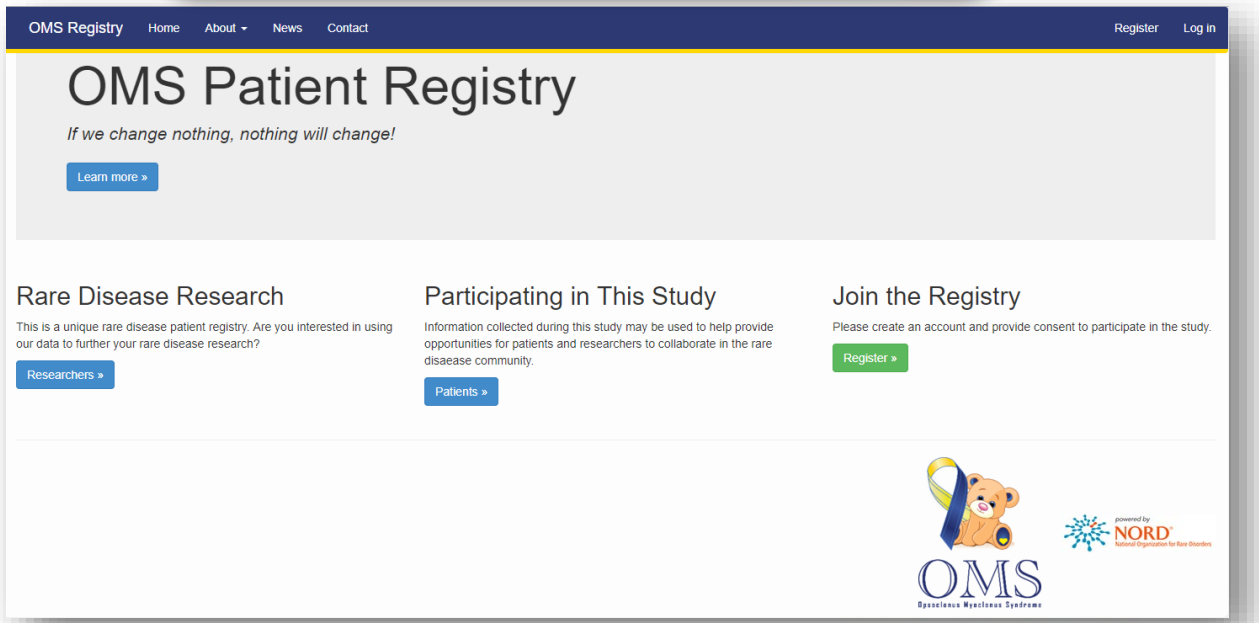
- Meaningful use of the data
  - Industry
  - Regulators
  - Share with patient community
  - Research Collaborations
    - Worldwide registry and biobank
    - Publication of manuscripts
- Build OMS Community

· 2016 ·

NORD-FDA AWARDEE SELECTIONS

April 21, 2016 at 5:24 pm

How wonderful! My son was diagnosed with OMS almost 15 years ago. Hardly any information or research was available. We will be more than happy to help & share our experiences since diagnosis. This is such good news for all the rare diseases that received the grant.



# 5 MYTHS About Orphan Drugs & the Orphan Drug Act

NORD® RARE INSIGHTS™

MYTH 1:
*7 of the 10 Top-Selling Drugs in the United States Are Orphan Drugs*

**FACT:** These drugs have multiple indications, both orphan and non-orphan. For example, Humira has 12 indications, four of which are orphan. Of its $13.6 billion in total sales in 2016, only 3.8% were for orphan indications.

MYTH 2:
*Blockbuster drugs are protected from competition by seeking added orphan indications and reaping the benefit of market exclusivity for the entire drug.*

**FACT:** If a drug is already on the market and the company gains approval for an additional orphan indication, the benefit of seven-years of exclusivity under the Orphan Drug Act applies only to the new orphan indication, not the entire drug.

MYTH 3:
*Orphan Drugs Are a Major Contributor to Rising Drug and Healthcare Costs.*

**FACT:** Of the total drug sales of $450 billion in the US in 2016, only 7.9% were for orphan designations of approved drugs.

MYTH 4:
*Specialty Drugs Are the Same as Orphan Drugs*

**FACT:** The two are not the same. Specialty drugs are defined by special requirements (i.e., for storage or handling); how they are administered (i.e., by a professional or as an infusion); and how much they cost. While an orphan drug may be a specialty drug, not all specialty drugs are orphans.

MYTH 5:
*The benefits of the Orphan Drug Act distort the marketplace and bias research away from diseases affecting more people.*

**FACT:** Studying rare diseases has led to increased understanding of the body's biochemical pathways and to major breakthroughs in discovering how our genes interact with other factors to cause disease. The Orphan Drug Act has helped drive innovation in many fields within medicine, including cancer treatment.

# Resources

- NORD
  - https://rarediseases.org/

- Educational Videos – NORD YouTube Channel
  - https://www.youtube.com/user/raredisorders/videos
  - Gene Therapy https://youtu.be/5ChXI6cSQs0



- Papers
  - www.rarediseases.org/nord-white-paper-quintilesims-report-download/

- Rare Action Network®
  - Rareaction.org

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# NORD Membership and International Partnerships










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# THANK YOU

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