



**NORD<sup>®</sup>**

National Organization for Rare Disorders

# The Importance of Natural History Studies for Rare Diseases

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**30** Million Americans: that's **1** in **10** people

# NORD

The National Organization for Rare Disorders (NORD) is the primary patient advocacy organization dedicated to addressing the needs of individuals and families impacted by rare diseases through programs of advocacy, education, research and financial assistance services.



**NORD<sup>®</sup>**

National Organization for Rare Disorders

**Advocacy**

**Patient  
Services**

**Education**

**Research**

**Membership (250+ Patient Advocacy Groups + 1,000 + Individuals)**

# Challenges in Rare Disease Communities

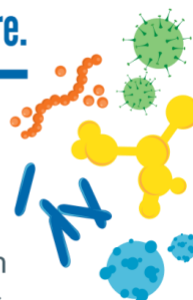
Barriers to care include access to

- Information
- Clinicians
- Treatment
- Support



**All childhood cancers are classified as rare.**

They are the leading cause of death by disease past infancy among children in the United States.



# Natural History of a Disease

“The natural course of a disease from the time immediately prior to its inception, progressing through its pre-symptomatic phase and different clinical stages to the point where it has ended and the patient is either cured, chronically disabled, or dead without external intervention.”

Posada de la Paz M; Groft SC Adv Exp Med Biol 2010; 686: 3-14

# Natural History Study (NHS)

- Identifies demographic, genetic, environmental and other variables associated with the disease
- Provides an avenue for biospecimen collection
- Is most informative when data are available early in the drug development process
- Succeeds when patients and caregivers play a role in the design, implementation and management in NHS

# Benefits of Natural History Studies

- Educate patients, caregivers, researchers and other stakeholders
- Provide opportunities for researchers to collaborate on projects
- Provide access to treatments
- Empower the patient community

# Patient Centered Outcomes Research

APBDRF FAN/NHS

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## APBDRF FAN Natural History Study

*Welcome to the APBDRF FAN Natural History Study.*

[Learn more »](#)

### Rare Disease Research

This is a unique rare disease patient registry. Are you interested in using our data to further your rare disease research?

[Researchers »](#)

### Participating in this Study

Information collected during this study may be used to help provide opportunities for patients and researchers to collaborate in the rare disease community.

[Patients »](#)

### Join the Registry

Please create an account and provide consent to participate in the study.

[Register »](#)

**APBD FAN NHS**  
ADULT POLYGLUCOSAN BODY DISEASE

powered by  
**NORD**  
National Organization for Rare Disorders

# Patient Centered Outcomes Research

## Treatment

*Please enter all of the Participant's past and present medications (prescription, over the counter, homeopathic, other) by clicking the "add another record" button for each medication.*

Select a medication: \*

+ Add another record

**Did the Participant take any dietary supplements during the past year, at least once a week?**

☐ Yes

☐ No

**Does the Participant take any medical foods or follow a special diet for treatment of his/her rare disease?**

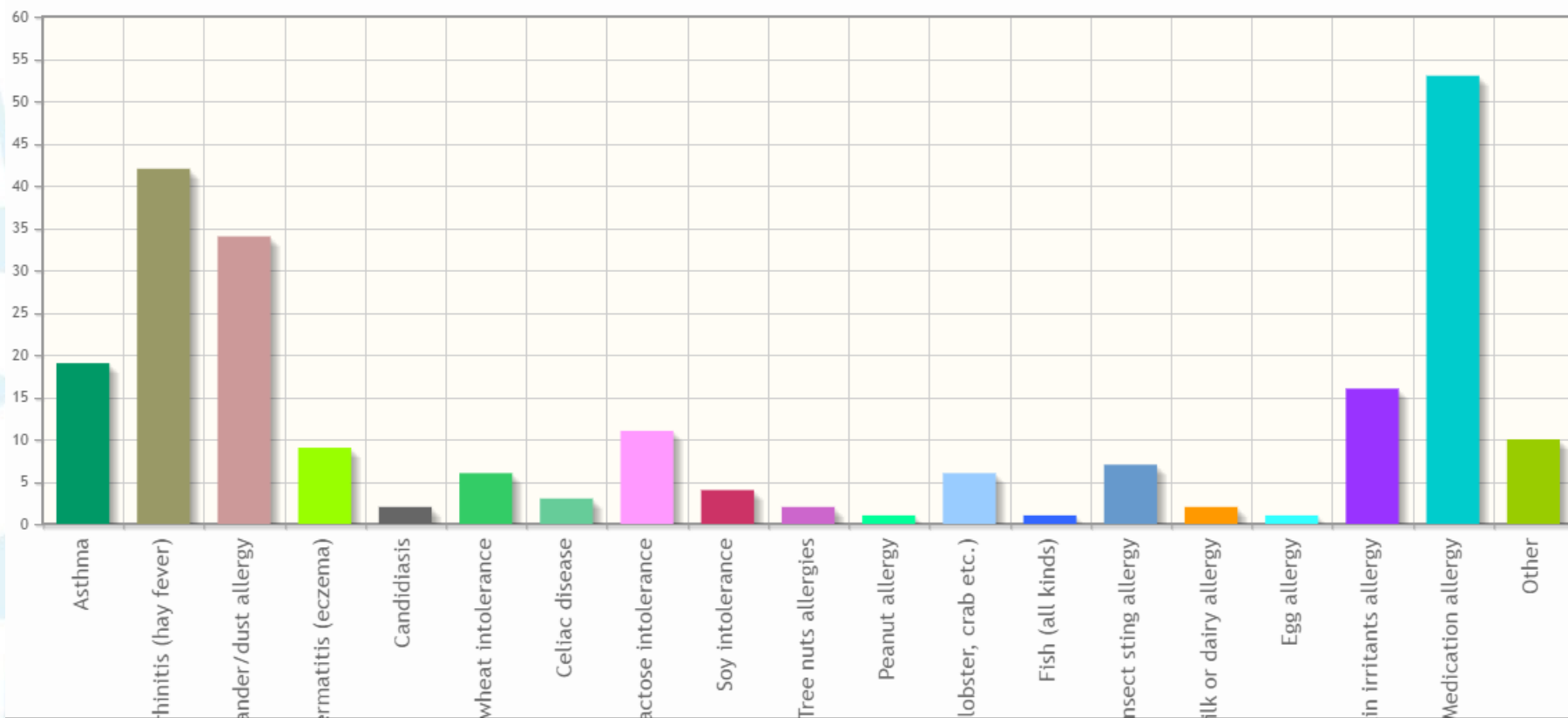
☐ Yes

☐ No

☐ Don't know

# Access to Data

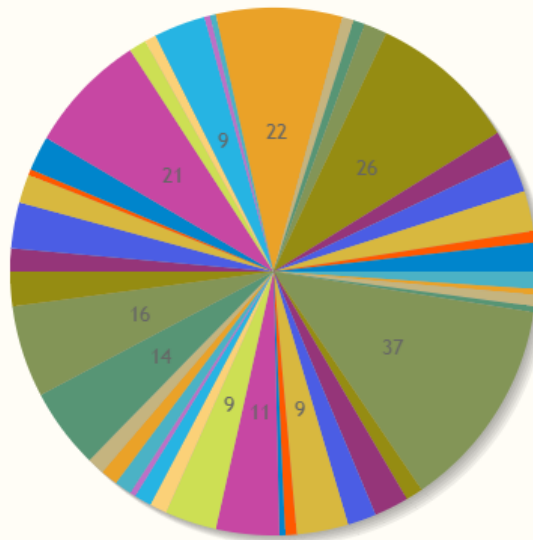
Do you have any allergies or dietary restrictions? (Responses: 229)



[Scroll to Question](#) ▲ [Scroll to Top](#)

# Access to Data

In which state or territory was the participant born, as shown on his/her birth certificate? (Responses: 284)



Alabama (3)	Alaska (1)	Arizona (2)	Arkansas (1)	California (37)	Colorado (3)
Connecticut (6)	District of Columbia (5)	Florida (9)	Georgia (2)	Hawaii (1)	Illinois (11)
Indiana (9)	Iowa (3)	Kansas (3)	Kentucky (1)	Louisiana (3)	Maine (3)
Maryland (3)	Massachusetts (14)	Michigan (16)	Minnesota (6)	Mississippi (4)	Missouri (8)
Nebraska (5)	New Hampshire (1)	New Jersey (6)	New York (21)	North Carolina (3)	North Dakota (2)
Ohio (9)	Oklahoma (1)	Oregon (1)	Pennsylvania (22)	Rhode Island (2)	South Carolina (2)
Tennessee (4)	Texas (26)	Utah (5)	Virginia (6)	Washington (7)	West Virginia (2)
Wisconsin (5)					

# Final Thoughts

## Natural history studies

- Increase the understanding of rare diseases by providing a platform for fundamental research into disease processes.
- Provide the incentive of leveraging PCOR to optimize the use of existing drugs and/or creating novel treatments
- Give patients the flexibility to participate in research regardless of where they live

# Natural History Study Patient Registries



HEREDITARY  
NEUROPATHY  
FOUNDATION



THE DESMOID TUMOR  
RESEARCH FOUNDATION



APBDRF  
ADULT POLYGLUCOSAN BODY DISEASE RESEARCH FOUNDATION



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Organic Acidemia  
Association  
[www.oaanews.org](http://www.oaanews.org)



Scleroderma  
RESEARCH FOUNDATION  
[WWW.SRFCURE.ORG](http://WWW.SRFCURE.ORG)



a common hope | an uncommon bond | [www.pemphigus.org](http://www.pemphigus.org)

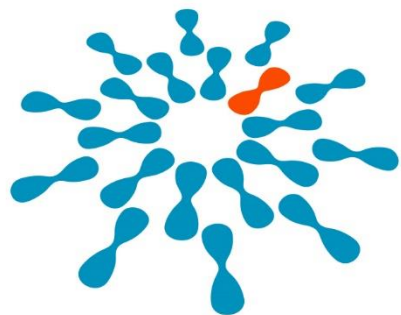
Lipoprotein  
FOUNDATION



Educating, Empowering  
and Saving Lives



Education & Research Foundation



**NORD<sup>®</sup>**

National Organization for Rare Disorders

**Alone we are rare.  
Together we are strong.<sup>®</sup>**

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*Thank You!*