

# Helping people with rare diseases through information and research

2016



**ICORD**

International Conference on  
Rare Diseases & Orphan Drugs

19-22 OCTOBER 2016

CAPE TOWN | SOUTH AFRICA

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NCATS



> 6,000 rare diseases

25 M affected in US

Many undiagnosed

< 500 have any treatment



# Many unanswered questions for people living with rare diseases



“How can we get a diagnosis?”

“How do we transition from pediatric to adult care?”

“How do we connect with other families?”

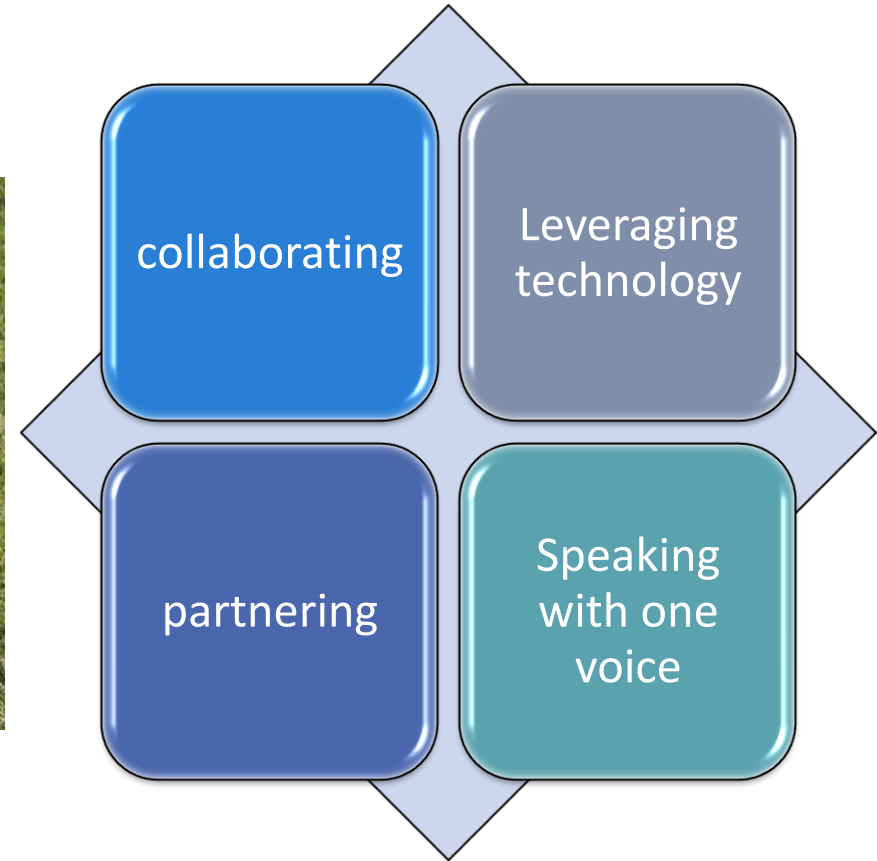
“What does it take to advance research?”

“When will we have a cure?”





# How can we all make a difference for rare diseases?



# How do we get to new treatments?



Discovery

Pre-clinical

Trial Readiness

Trials



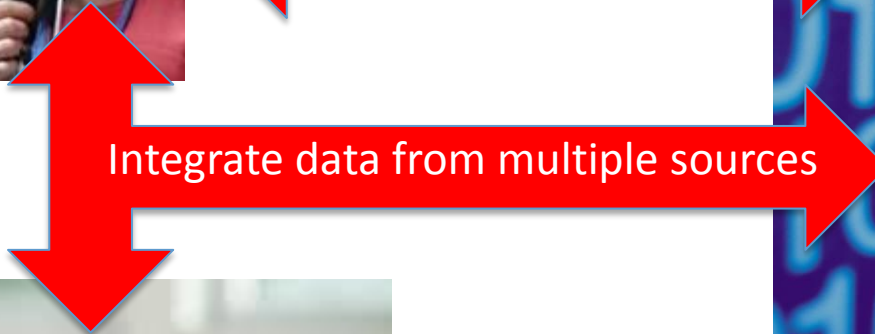
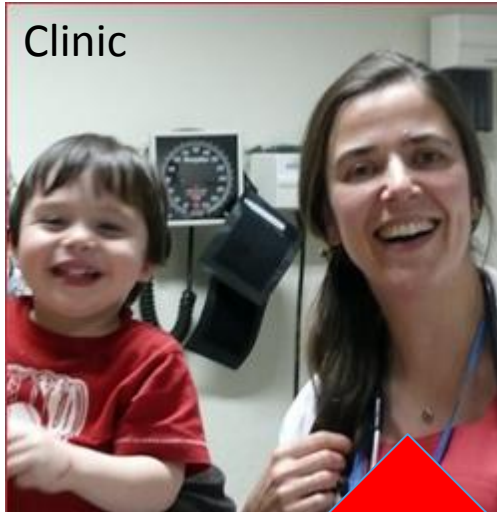
## For rare diseases:

- Continuity along this path
- Make every patient count
- Make every visit count

Post-approval



# How can we get there faster?



- Harmonizing efforts
- Streamlining use of data
- COLLABORATION!

# How can we make sure our data count?



Photo credit: Michael and Rachel Harris

For rare diseases:

- Continuity of data
- Data standards
- Keep “end-user” in mind

Registry

Natural History Study

Biomarkers

Trials

Post-approval





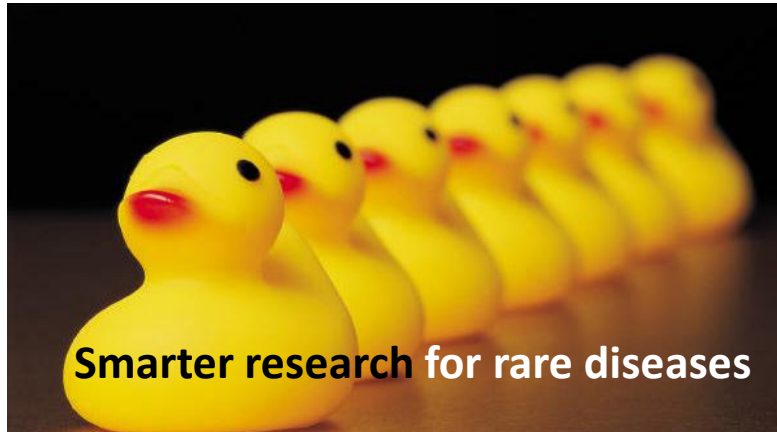
# Partnering for Success





# What do we do at the NIH/NCATS?

## National Center for Advancing Translational Sciences



- Single IRB for multi-site study
- Pre-negotiated contracts
- Study participant recruitment
- Linking EMR and research data



- National network
- Accelerates translation
- Promotes excellence and innovation  
in research and training

- Collectively, the RDCRN is studying **200 rare diseases** at **266 clinical sites** located in **18 countries, including the U.S.**
- There are more than **90 active protocols**
- **41,519 patients** have enrolled in clinical studies
- **208 trainees**
- **3,316 collaborative consortium members**
- **144 patient advocacy groups (PAGs)** as research partners, which collectively formed a Coalition (RDCRN-CPAG)

# RDCRN Website



Working together to find better treatments  
& improve the quality of life for  
individuals with rare diseases

The **Rare Diseases Clinical Research Network (RDCRN)** an initiative of the Office of Rare Diseases Research, NCATS, is made up of **22 research consortia** and a **Data Management and Coordinating Center** that are working together to improve availability of rare disease information, treatment, clinical studies, and general awareness for both patients and the medical community. The RDCRN provides up-to-date information for patients and assists in connecting patients with advocacy groups, expert doctors, and clinical research opportunities. **More about the RDCRN >**

## Rare Disease Information



### Find a disease

Find a description, consortium information, patient advocacy and study information for

### Find a study

Find information about the RDCRN studies.

### Who we are

Get to know the 22



### Are YOU Interested in Research on Rare Diseases?

Stay Connected - Join the Contact Registry!

Receive the most current information

**NIH**

**ORDR/NCATS, NCI, NHLBI,  
NIAID, NIAMS, NICHD, NIDCR,  
NIDDK, NIMH, NINDS, ODS**

**Coalition of Patient  
Advocacy Groups  
(CPAG) for RDCRN**

**Porphyria Rare Disease Clinical  
Research Consortium**

**PAG = PATIENT  
ADVOCACY  
GROUPS**

**Dystonia  
Coalition**

**North America Mitochondrial  
Diseases Consortium**

**Developmental Synaptopathies  
Associated with TSC, PTEN  
And SHANK3 Mutations**

**Primary Immune Deficiency  
Treatment Consortium**

**The Frontotemporal Lobar  
Degeneration Clinical  
Research Consortium**

**Brittle Bone Disorders  
Consortium**

**Inherited Neuropathies  
Consortium**

**Chronic Graft Versus  
Host Disease**

**Nephrotic Syndrome  
Study Network**



- Collaborative Clinical Research
- Centralized Data Coordination & Technology Development
- Public Resources & Education
- Training

**The Data Management and  
Coordinating Center**

**Rare Lung Diseases  
Consortium**

**Urea Cycle Disorders  
Consortium**

**Lysosomal  
Disease Network**

**Brain Vascular  
Malformation Consortium**

**Rare Kidney  
Stone Consortium**

**Genetic Disorders of  
Mucociliary Clearance**

**Vasculitis Clinical  
Research Consortium**

**Consortium of Eosinophilic  
Gastrointestinal Disease Researchers**

**Clinical Research in ALS & Related  
Disorders for Therapeutic Development**

**Rett, MECP2 Duplications  
and Rett-Related  
Disorders Consortium**

**Autonomic Disorders  
Consortium**

**Sterol and Isoprenoid  
Diseases Consortium**



# NCATS Genetic and Rare Diseases Information Center (GARD)

The screenshot shows the GARD website interface. At the top, the NIH logo is on the left, followed by the text 'National Center for Advancing Translational Sciences' and 'GARD Genetic and Rare Diseases Information Center'. Below this is a navigation bar with tabs for 'Diseases', 'Resources', 'Research', 'News & Events', and 'About GARD'. The 'Resources' tab is currently selected. The main content area is divided into several sections. On the left, there's a teal box with the text 'Have Questions About Genetic or Rare Diseases?' and a link to 'Read more...'. Next to it is a photo of a woman wearing a headset. To the right of the photo is a search bar with the text 'Search for a condition' and a 'Go' button. Below the search bar is a link to 'Enfermedades en español'. Further down is a 'Browse Diseases' section with a grid of letters A through Z and a '0-9' link. Below this is a 'View:' section with a link to 'Diseases by Category' and a link to 'Rare Diseases with FDA-Approved Medical'. At the bottom of the page, there are three columns of content. The first column is 'About GARD', which describes the center and its funding. The second column is 'Research', which lists several NIH-supported research efforts. The third column is 'Resources', which includes a 'Featured Guide' for 'How to Find a Disease Specialist' and links to 'Rare Diseases Resources', 'Genetics Resources', 'Genetic Testing and Treatment', and 'Support for Patients'.

NIH National Center for Advancing Translational Sciences

GARD Genetic and Rare Diseases Information Center

Diseases Resources Research News & Events About GARD

Have Questions About Genetic or Rare Diseases?

Get reliable answers from the Genetic and Rare Diseases (GARD) Information Center. If you...

Read more...

1 2 3 4

Genetic and Rare Diseases (GARD) Information Center

Search for a condition Go

Enfermedades en español

Browse Diseases

A B C D E F G H I J K L M  
N O P Q R S T U V W X Y Z  
0-9

View:

Diseases by Category  
Rare Diseases with FDA-Approved Medical

About GARD

The Genetic and Rare Diseases (GARD) Information Center is a program of the [National Center for Advancing Translational Sciences](#) and is funded by two parts of the National Institutes of Health (NIH): NCATS and the National Human Genome Research Institute (NHGRI). The GARD Information Center provides the public with access to current, reliable, and easy-to-understand information about rare or genetic diseases in English or Spanish.

Read more [about GARD](#).

Your Questions Answered

Answers to recently asked questions from the public on these rare and/or genetic diseases:

Research

NIH Rare Disease Programs  
A select list of NIH-supported research efforts.

- Rare Diseases Clinical Research Network (RDCRN)
- NIH/NCATS GRDR™ Program (Global Rare Diseases Patient Registry Data Repository)
- Therapeutics for Rare and Neglected Diseases (TRND)
- Undiagnosed Diseases Network (UDN)
- Bench-to-Bedside Awards

Research Funding Resources  
Funding opportunities from the NIH and other sources

Resources

Featured Guide  
How to Find a Disease Specialist MORE GUIDES

Rare Diseases Resources  
A collection of resources for the rare disease community

Genetics Resources  
A collection of resources that provide information on genes, genomics, and genetic conditions

Genetic Testing and Treatment  
Learn about genetic testing and finding healthcare services

Support for Patients

<https://rarediseases.info.nih.gov>

Online resource with:

- Up-to-date, reliable and easy-to-understand information on rare or genetic diseases
- In English or Spanish
- For people with rare or genetic diseases, their families, friends, care providers and wider communities
- Contact information for telephone and email queries

# NIH/NCATS Global Rare Diseases Registry (GRDR®) Program

NIH National Center for Advancing Translational Sciences

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Research Funding & Notices News & Media About Translation About NCATS

**Collaborating to Advance Rare Diseases Research**

NCATS and Harvard are collaborating to advance rare diseases research to benefit patients. [More...](#) [Work with Us](#)

HARVARD MEDICAL SCHOOL DEPARTMENT OF BIOMEDICAL INFORMATICS

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## News

Home / News / DBMI and NCATS to Collaborate on Global Rare Diseases Data Repository

### DBMI and NCATS to Collaborate on Global Rare Diseases Data Repository

September 28, 2015



In August 2015, the National Center for Advancing Translational Sciences (NCATS) awarded DBMI a supplemental grant to develop the **Global Rare Diseases Patient Registry Data Repository** (GRDR®). The GRDR program is designed to facilitate the application of scientific insights gained from the integration of different types of de-identified patient electronic health records, research data, and clinical trial data into a single place. The goal is to develop a Web-based platform that will facilitate the integration of these data to authorized users. Using this approach, data providers can more easily share their data and help get more treatments to more patients more quickly.

The ability to retrieve data from such a repository for comparative and cross-disease analysis, facilitating its use in state-of-the-art collaborative research ultimately may lead to new understandings of disease mechanisms and the development of new drugs and therapeutics for the millions of patients with rare diseases.

This NCATS-HMS collaboration will enable work to integrate different patient registries into the GRDR. The goal is to develop a Web-based platform that will facilitate the integration of these data to authorized users. Using this approach, data providers can more easily share their data and help get more treatments to more patients more quickly.

[About the GRDR](#) [Common Data Elements](#)

- Common Data Elements (CDEs) for collecting data
- Informed consent templates
- Access to GRDR Global Unique Identifier (GUID)
- Map patient data to GRDR CDEs & national standards
- Information and tools

# NCATS Toolkit Project



NCATS

# Rare Diseases Toolkit



## Why?

- Patient involvement and community engagement are vital throughout the translational research process
- A wealth of educational and informational tools have already been developed by and for rare disease community
  - Academia
  - Disease foundations
  - Government agencies
  - Industry
- ***But*** existing resources are dispersed, difficult to discover — especially for newcomers



# What will the Toolkit project do for the rare diseases community?

- Collaboratively create a **well-designed source for online educational and informational research resources and tools**.
- Provide a **single online portal with resources** that patient groups can readily access along with context.
- **Improve coordination** rather than re-create existing resources.
- Facilitate opportunities to **bring groups together, identify gaps** in online resources, and **disseminate information** to patient groups.
- Promote **continuity across the lifecycle of the drug development process**.



Pre-clinical

Trial readiness

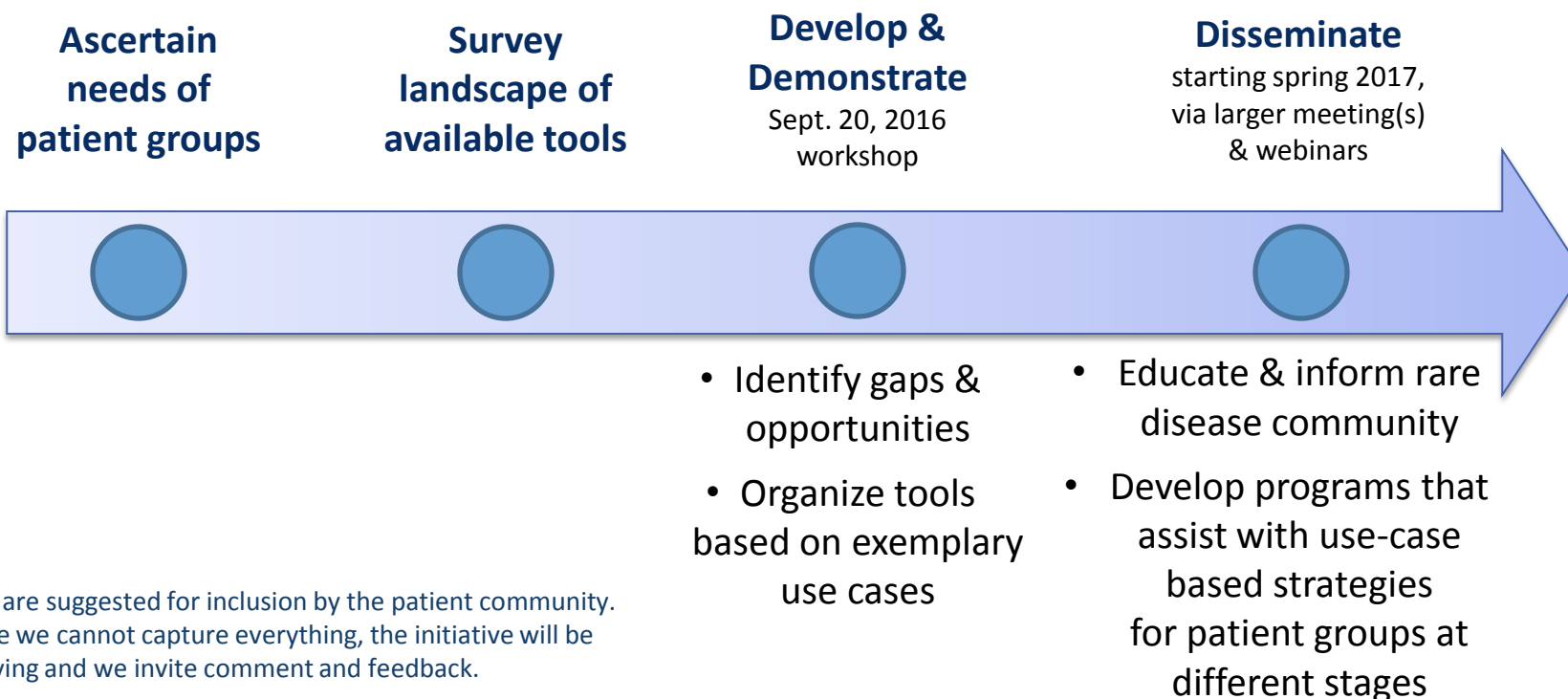
Trials

Post-approval

# How we plan to develop the Toolkit



- Planning group driven by patient group representatives
- Inclusive\*, transparent, collaborative
- Focus on tools that are useful for research, easily accessible and practical



\* Tools are suggested for inclusion by the patient community. While we cannot capture everything, the initiative will be evolving and we invite comment and feedback.

# Take-home Messages



- We need to work smarter so that we can get better treatments to people living with rare diseases
- Strategies for success are:
  - Collaborating
  - Leveraging technologies
  - Partnering among stakeholders
- What you can do:
  - Let your organizations and communities know how critical these strategies are in bringing more research discoveries to more rare diseases patients more quickly.



**Thank you  
& Time for Questions**



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Program Assistant

# Connect With NCATS:

[ncats.nih.gov/connect](https://ncats.nih.gov/connect)

**Website:** [ncats.nih.gov](https://ncats.nih.gov)



**Facebook:** [facebook.com/ncats.nih.gov](https://facebook.com/ncats.nih.gov)



**Twitter:** [twitter.com/ncats\\_nih\\_gov](https://twitter.com/ncats_nih_gov)



**YouTube:** [youtube.com/user/ncatsmedia](https://youtube.com/user/ncatsmedia)



**E-Newsletter:** <https://ncats.nih.gov/enews>

**Announce Listserv:** <http://bit.ly/1sdOI5w>

