

OFFICE OF RARE DISEASES UPDATE

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NCATS



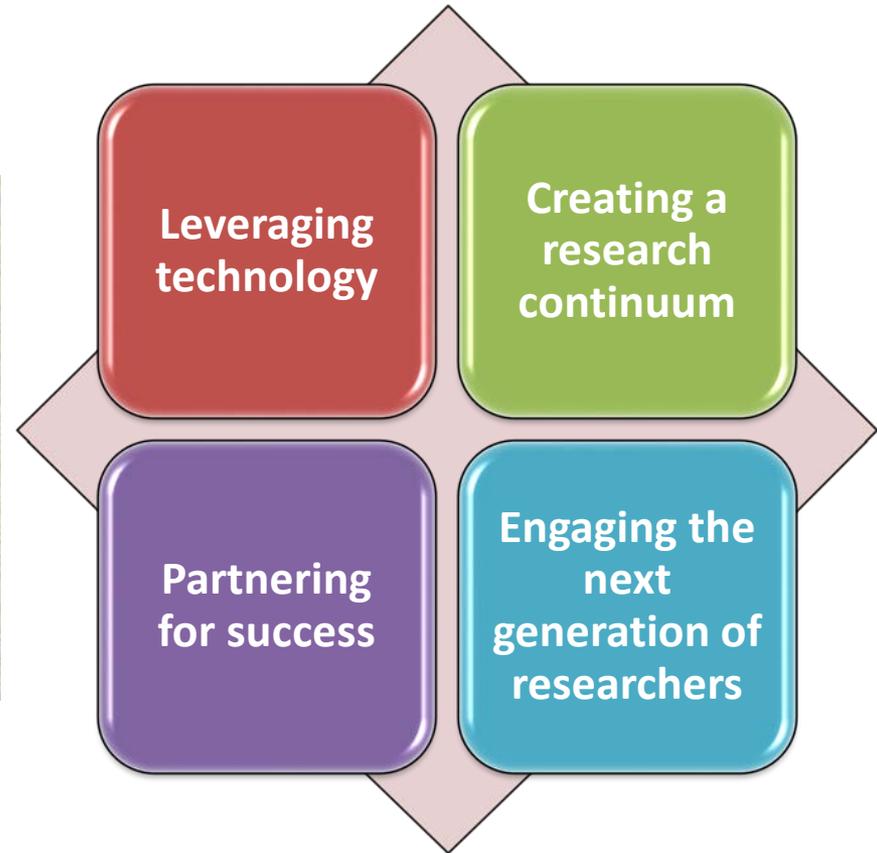
> 7,000 rare diseases

25 M affected in US

Many undiagnosed

< 500 have any treatment

How can we all make a difference for rare diseases?



How can we accelerate the path from discovery to health benefit?



How can we make sure our data count?



Photo credit: Michael and Rachel Harris

Registry

Natural History Study

Biomarkers

Trials



Post-approval



For rare diseases:

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

Partnering for Success



Trans-NIH Rare Diseases Working Group

- Rare Diseases affect many organs, and are often multi-system diseases.
- Therefore, strong partnerships with the categorical NIH ICs are important.
- Coordination and cooperation stipulated in Rare Diseases Act of 2002.
- One way by which the ORDR facilitates coordination and cooperation is through the Trans-NIH Rare Diseases Working Group.
- The re-constituted working group met on December 12, 2016 and will meet quarterly with rotating meeting venue.

CURRENT ORDR PROGRAMS

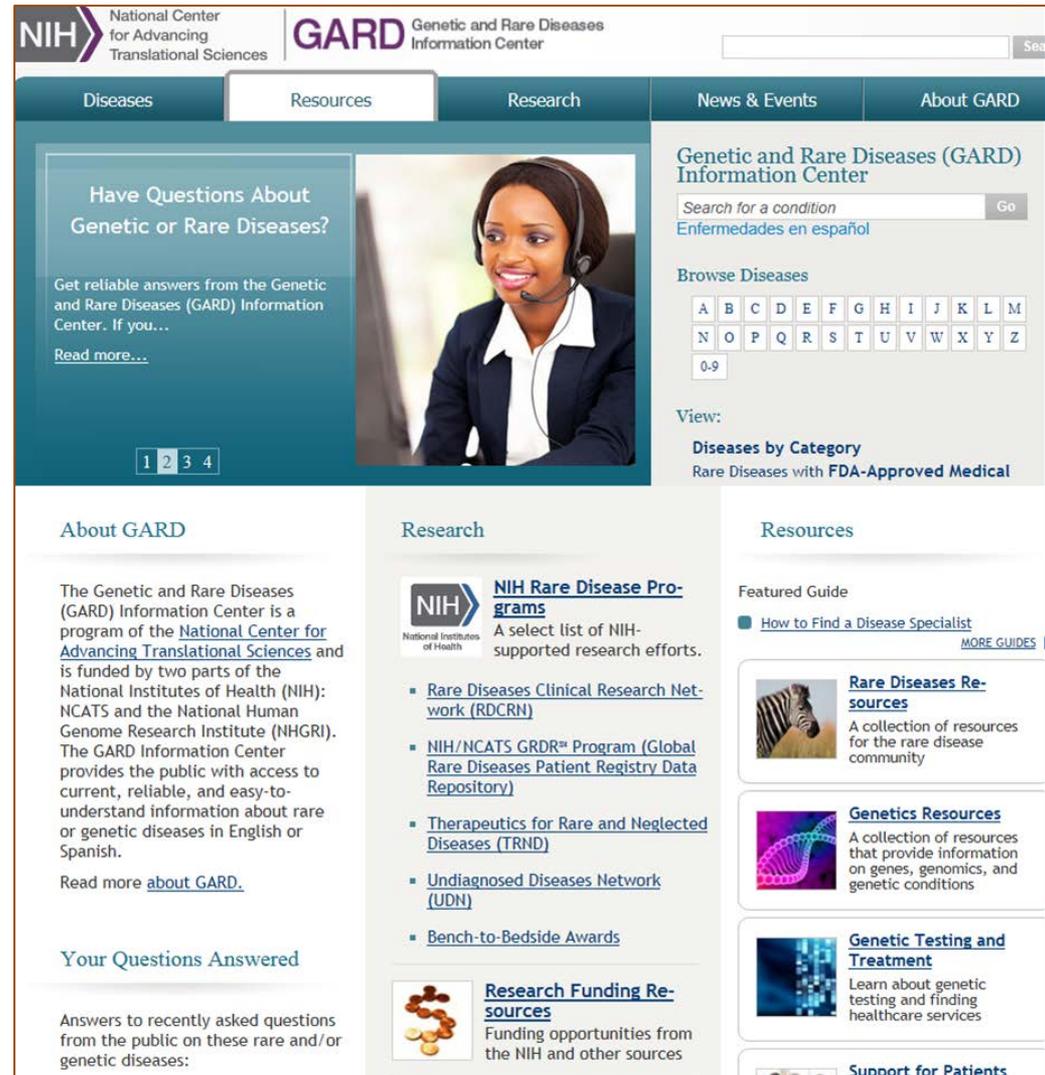
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NCATS Genetic and Rare Diseases Information Center (GARD)

<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



The screenshot shows the homepage of the Genetic and Rare Diseases (GARD) Information Center. The header includes the NIH logo and the text 'National Center for Advancing Translational Sciences' and 'GARD Genetic and Rare Diseases Information Center'. The main navigation bar has tabs for 'Diseases', 'Resources', 'Research', 'News & Events', and 'About GARD'. A large banner on the left asks 'Have Questions About Genetic or Rare Diseases?' and features a photo of a woman wearing a headset. To the right, there is a search bar, a 'Go' button, and a link to 'Enfermedades en español'. Below the search bar is a 'Browse Diseases' section with a grid of letters A-Z and a '0-9' button. The main content area is divided into three columns: 'About GARD' (describing the center's mission and funding), 'Research' (listing programs like RDCRN, GRDR, TRND, UDN, and Bench-to-Bedside Awards), and 'Resources' (featuring guides like 'How to Find a Disease Specialist', 'Rare Diseases Resources', 'Genetics Resources', and 'Genetic Testing and Treatment').

NIH/NCATS Rare Diseases Registry (GRDR) Program

NIH National Center for Advancing Translational Sciences

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



Home > About NCATS > NCATS Programs & Initiatives > The NIH/NCATS G

The NIH/NCATS GRDR® Program

The aim of the GRDR program is to develop a Web-based resourc secures and stores de-identified patient information from many rare diseases, all in one place.

[About the GRDR](#)

[Common](#)

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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



Image courtesy of NCATS

In August 2015, the National Center for Advancing Translational Sciences awarded DBMI a supplemental grant to support the **Rare Diseases Patient Registry Data Repository** (GRDR) project. The GRDR is a Web-based SURE platform (Patient-Centered Research Elements), which is a product of the **NIH's Big Data to Knowledge** program.

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 other information in a secure place.

The ability to retrieve data from such a secure, comparative and cross-disease and cross-institutional place

facilitating its use in state-of-the-art collaborative research ultimately may lead to new understandings of the biology of rare diseases, 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 ability to retrieve these data to authorized users. Using this approach, data providers can more easily share their data, and patients can help get more treatments to more patients more quickly.

- 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

The logo consists of several overlapping, curved yellow and white shapes that form a circular, abstract pattern. The text "RARE DISEASES CLINICAL RESEARCH NETWORK" is overlaid on this pattern in a dark blue, serif font. The words "RARE DISEASES" and "RESEARCH NETWORK" are stacked on the right side, while "CLINICAL" is on the left. There are horizontal lines under "CLINICAL" and "RESEARCH NETWORK".

RARE DISEASES CLINICAL RESEARCH NETWORK

Initiative of the National Center for Advancing
Translational Sciences (NCATS)

ORDR/NCATS

(NCI, NHLBI, NIAID, NIAMS, NICHD, NIDCR, NIDDK, NIMH, NINDS, ODS)

Coalition of Patient
Advocacy Groups
(CPAG)

Porphyria Rare Disease Clinical
Research Consortium

PAG

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

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

Autonomic Disorders
Consortium

Sterol and Isoprenoid
Diseases Consortium

Rett, MECP2 Duplications
and Rett-Related
Disorders Consortium



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

About the RDCRN Program

- Collectively, the RDCRN is studying 200 rare diseases in natural history and clinical trials at 418 active clinical sites located in the US and in 24 countries.
- There are more than 90 active protocols.
- 41,519 patients have enrolled in clinical studies.
- There have been 265 trainees.
- There are 3,545 collaborative consortium members.
- There are 144 PAGs as research partners, collectively formed a Coalition (RDCRN-CPAG).

<http://rarediseasesnetwork.epi.usf.edu/>

International Coordination

- Given the “rarity” of patients and investigators, the ORDR is engaged in coordination with international partners, including:
 - IRDiRC
 - GA4GH
 - ICORD
- ORDR participated in a collaborative project with the NCATS New Therapeutic Uses (NTU) program and the European E-Rare initiative:
 - RDCRN investigators were invited to seek support for participation in international clinical trials to repurpose drugs in collaboration with E-Rare (European investigators)
<http://grants.nih.gov/grants/guide/pa-files/PA-16-183.html>
 - Applications were received in July, 2016 (including two RDCRN applications).

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 the rare disease community
 - Academia
 - Disease foundations
 - Government agencies
 - Industry
- **But** existing resources are dispersed and 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

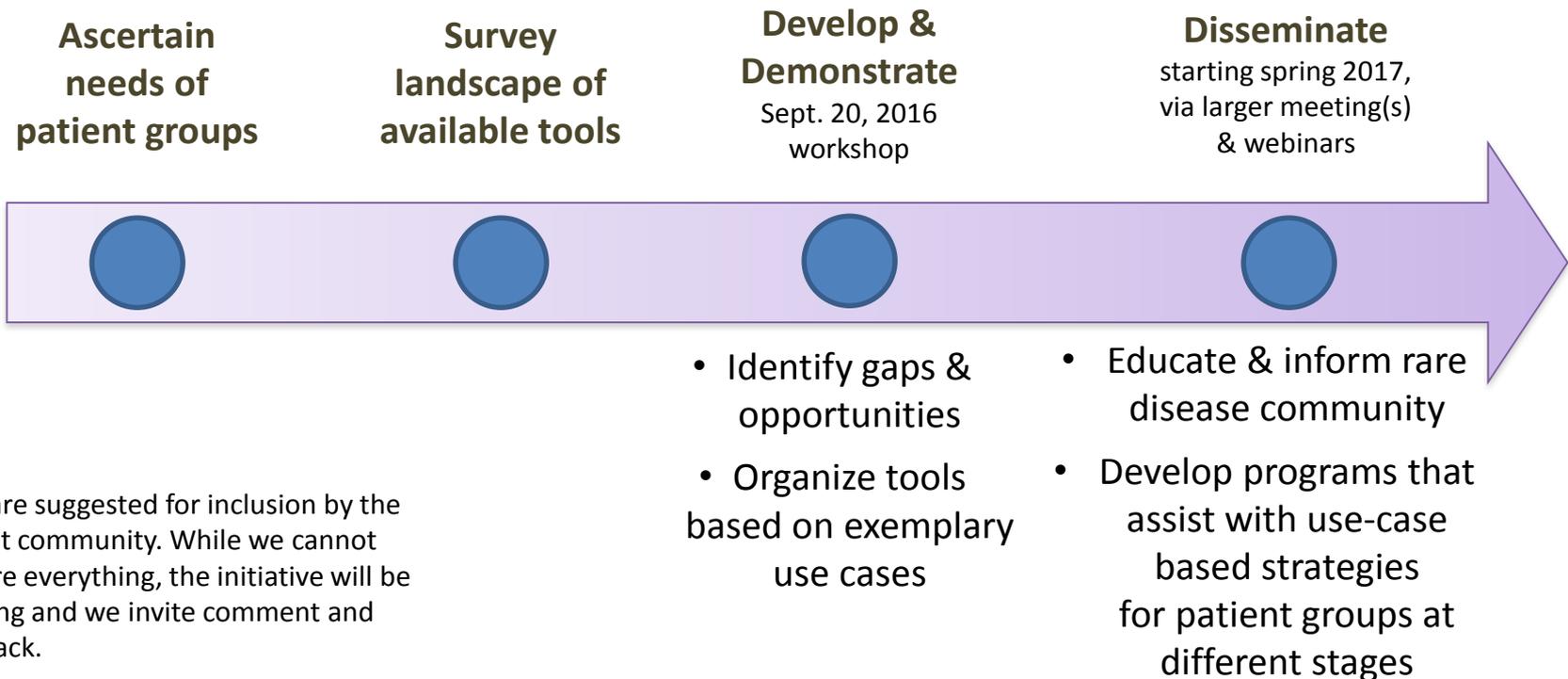
Trials

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

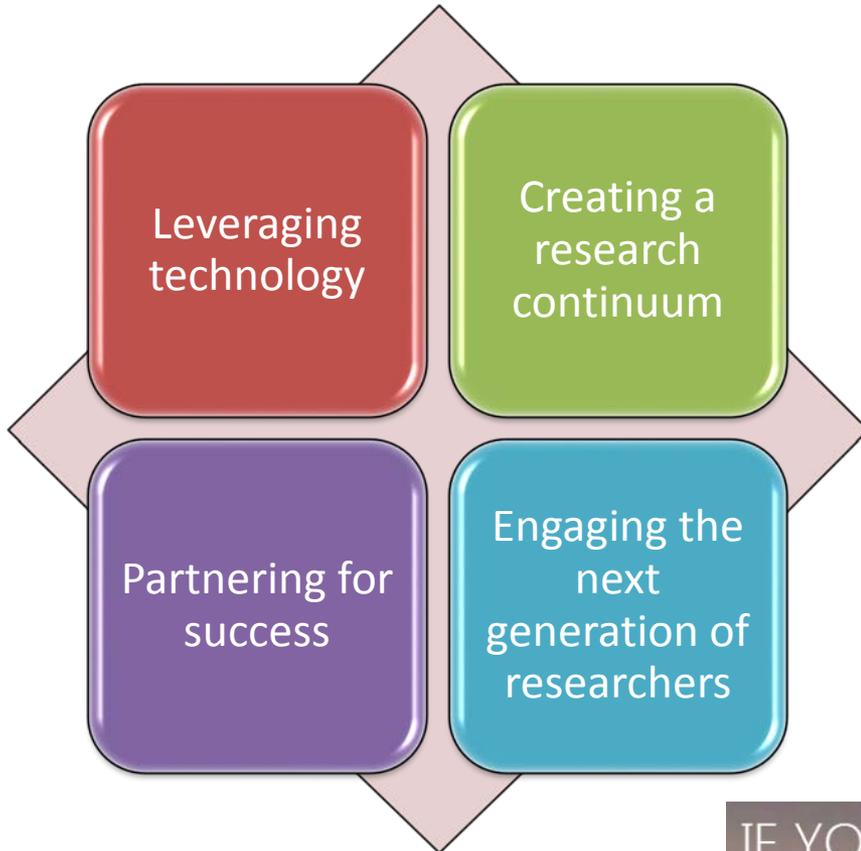


* Tools are suggested for inclusion by the patient community. While we cannot capture everything, the initiative will be evolving and we invite comment and feedback.

Potential Future Directions

- Increasing collaborations between the ORDR and NCATS DPI programs such as TRND
- Shifting from “one-disease-at-a-time” to “rare disease-ome” approach
- Stimulating partnerships for rare diseases
- Registries and natural history studies that are trial and regulatory-ready
- Promoting new therapeutic modalities such as gene therapy and editing for rare diseases
- Harmonizing and internationalizing rare diseases clinical research networks

Take-home Messages



IF YOU WANT TO GO FAR,
GO TOGETHER.

- African Proverb

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DISCUSSION

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