Helping people with rare diseases through information and research

PETRA KAUFMANN, M.D., M.SC.
DIRECTOR, DIVISION OF CLINICAL INNOVATION
DIRECTOR, OFFICE OF RARE DISEASES RESEARCH

NCATS

NIH National Center for Advancing Translational Sciences

> 6,000 rare diseases

25 M affected in US

Many undiagnosed

< 500 have any treatment

National Center for Advancing Translational Sciences

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?

- collaborating
- Leveraging technology
- partnering
- Speaking with one voice

How do we get to new treatments?

Discovery

Pre-clinical

Trial Readiness

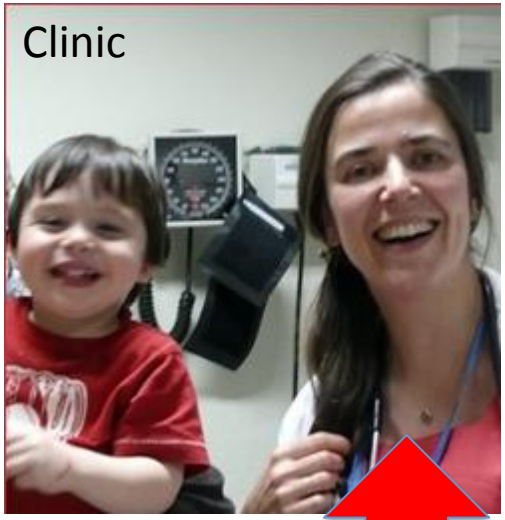
Trials

Post-approval

For rare diseases:

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

How can we get there faster?



Integrate care and research

Integrate data from multiple sources

- Harmonizing efforts
- Streamlining use of data
- COLLABORATION!

How can we make sure our data count?

Photo credit: Michael and Rachel Harris

Registry

Natural History Study

Biomarkers

Trials

FDA APPROVED

Post-approval

For rare diseases:

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

Partnering for Success

Patient Groups

Academics

Industry

Government
(in US: NIH, FDA)

What do we do at the NIH/NCATS?

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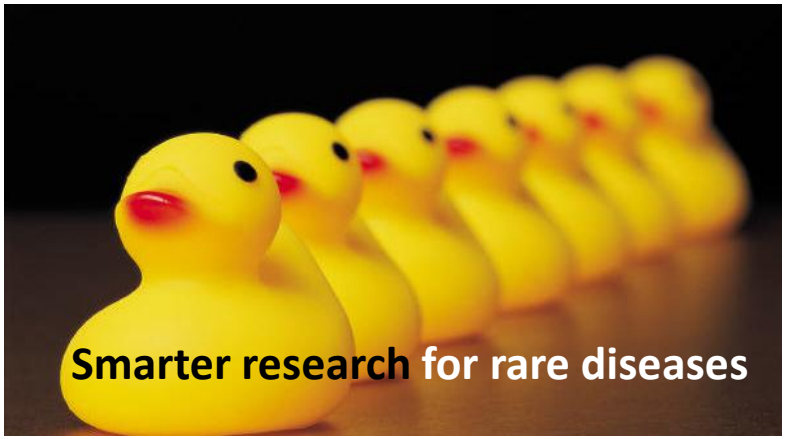


- Single IRB for multi-site study
- Pre-negotiated contracts
- Study participant recruitment
- Linking EMR and research data

CTSA Clinical & Translational Science Awards®

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

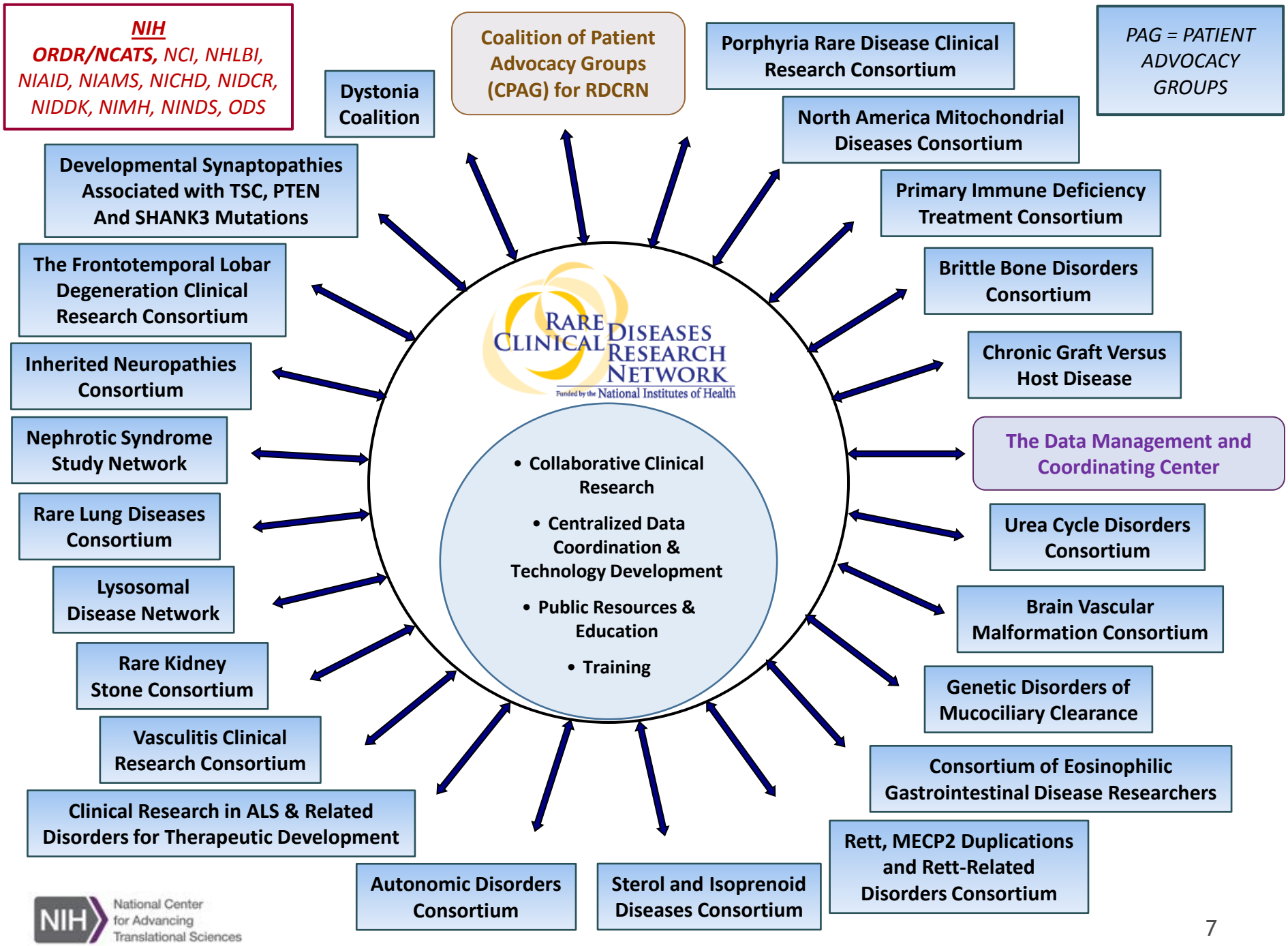
RARE CLINICAL DISEASES RESEARCH NETWORK

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

RDCRN

- 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


NIH
ORDR/NCATS, NCI, NHLBI, NIAID, NIAMS, NICHD, NIDCR, NIDDK, NIMH, NINDS, ODS
Coalition of Patient Advocacy Groups (CPAG) for RDCRN
PAG = PATIENT ADVOCACY GROUPS
Dystonia Coalition
Porphyria Rare Disease Clinical Research Consortium
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
RARE CLINICAL DISEASES RESEARCH NETWORK
Funded by the National Institutes of Health
Nephrotic Syndrome Study Network
The Data Management and Coordinating Center
• Collaborative Clinical Research
• Centralized Data Coordination & Technology Development
• Public Resources & Education
• Training
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
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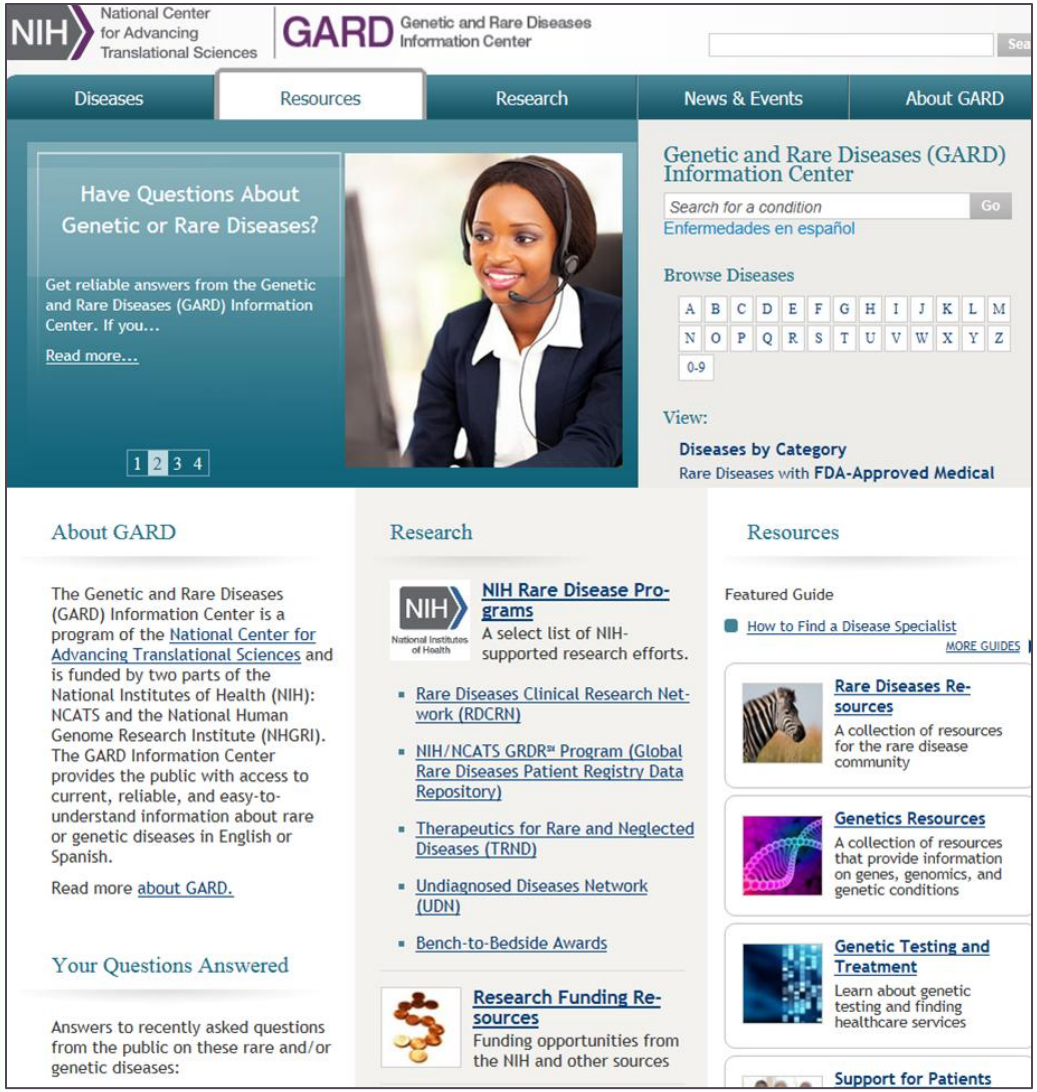
NCATS Genetic and Rare Diseases Information Center (GARD)



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

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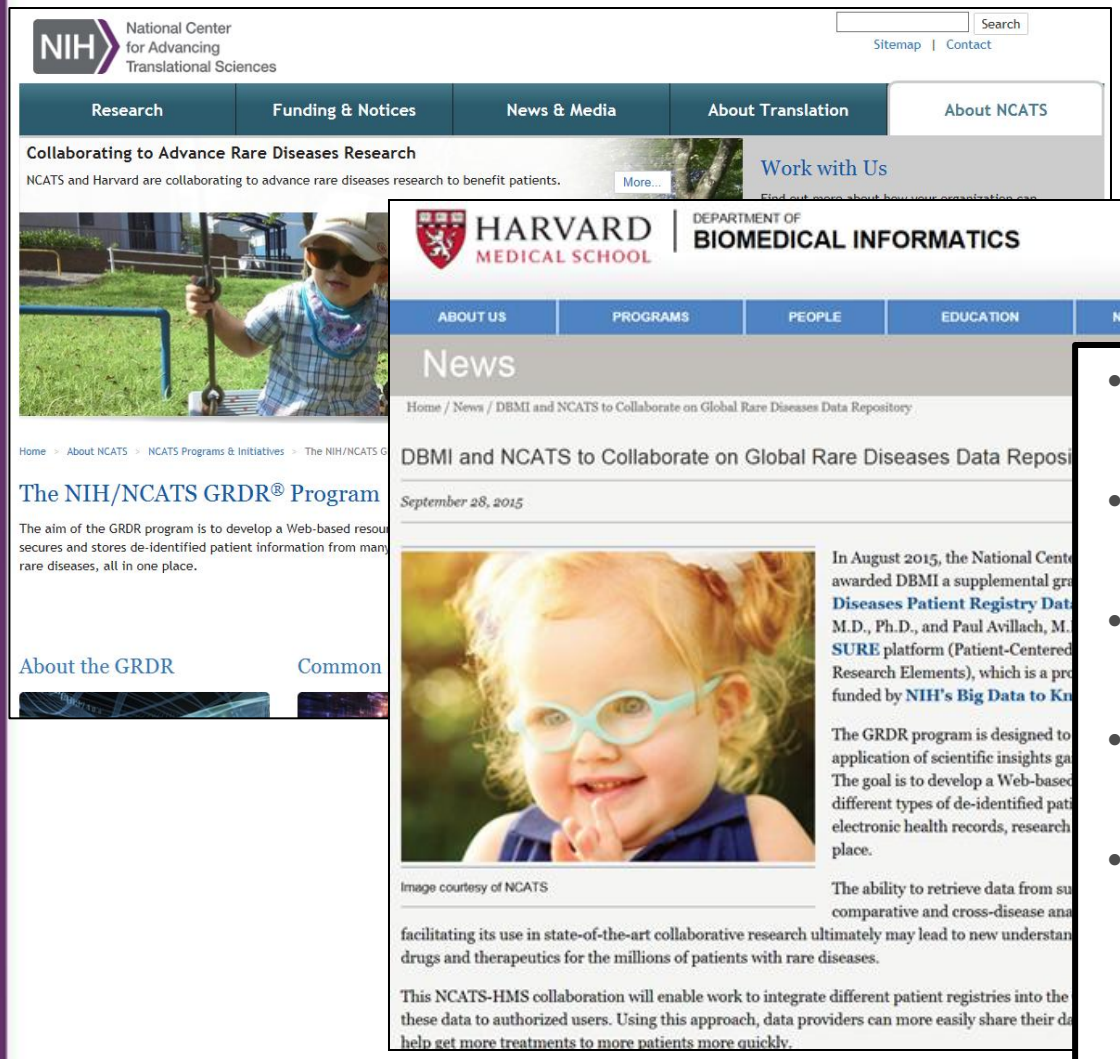
NIH/NCATS Global Rare Diseases Registry (GRDR®) Program



- 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

National Center for Advancing Translational Sciences

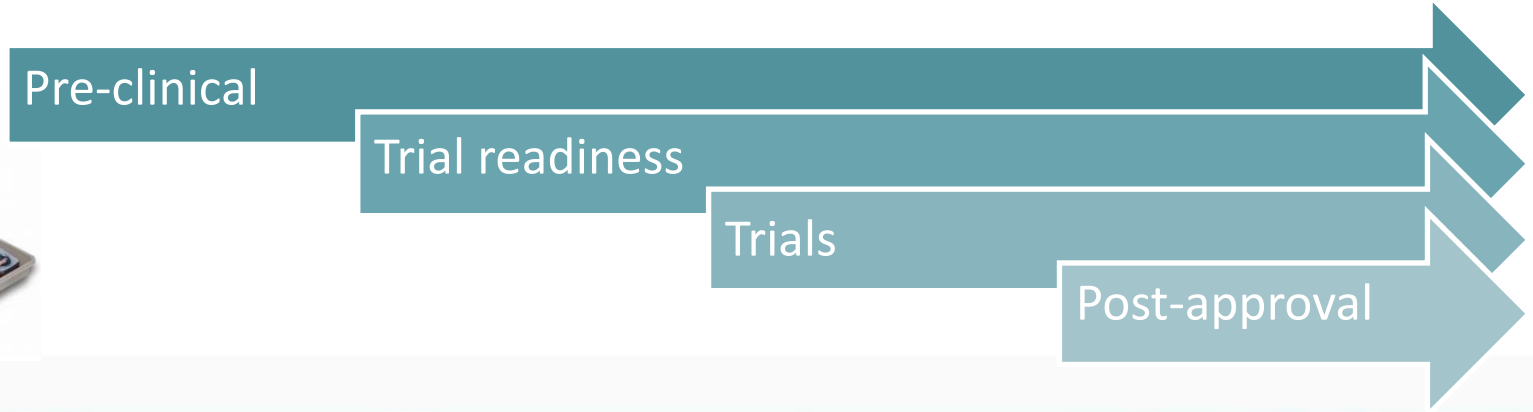
Rare Diseases Toolkit

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**.



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

Ascertain needs of patient groups

Survey landscape of available tools

Develop & Demonstrate
Sept. 20, 2016 workshop

- Identify gaps & opportunities
- Organize tools based on exemplary use cases

Disseminate
starting spring 2017, via larger meeting(s) & webinars

- Educate & inform rare disease community
- Develop programs that assist with use-case based strategies for patient groups at different stages

* 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.

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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

ORDR staff

- **Dr. Petra Kaufmann**, Petra.Kaufmann@nih.gov

Director, Office of Rare Diseases Research and Division of Clinical Innovation

- **Dr. David Eckstein**, David.Eckstein@nih.gov

Senior Health Science Administrator

- **Dr. Rashmi Gopal-Srivastava**, Rashmi.Gopal-Srivastava@nih.gov

Director of Extramural Research Program

- **Mr. Chris Griffin**, Chris.Griffin@nih.gov

Program Analyst

- **Ms. Henrietta Hyatt-Knorr**, Henrietta.Hyatt-Knorr@nih.gov

Senior Program and Policy Analyst

- **Ms. Susan Orr**, lowes@mail.nih.gov

Program Assistant

Connect With NCATS:
ncats.nih.gov/connect

Website: ncats.nih.gov

Facebook: facebook.com/ncats.nih.gov

Twitter: twitter.com/ncats_nih_gov

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