Rare Diseases Resources and Activities

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NCATS ORDR NIH

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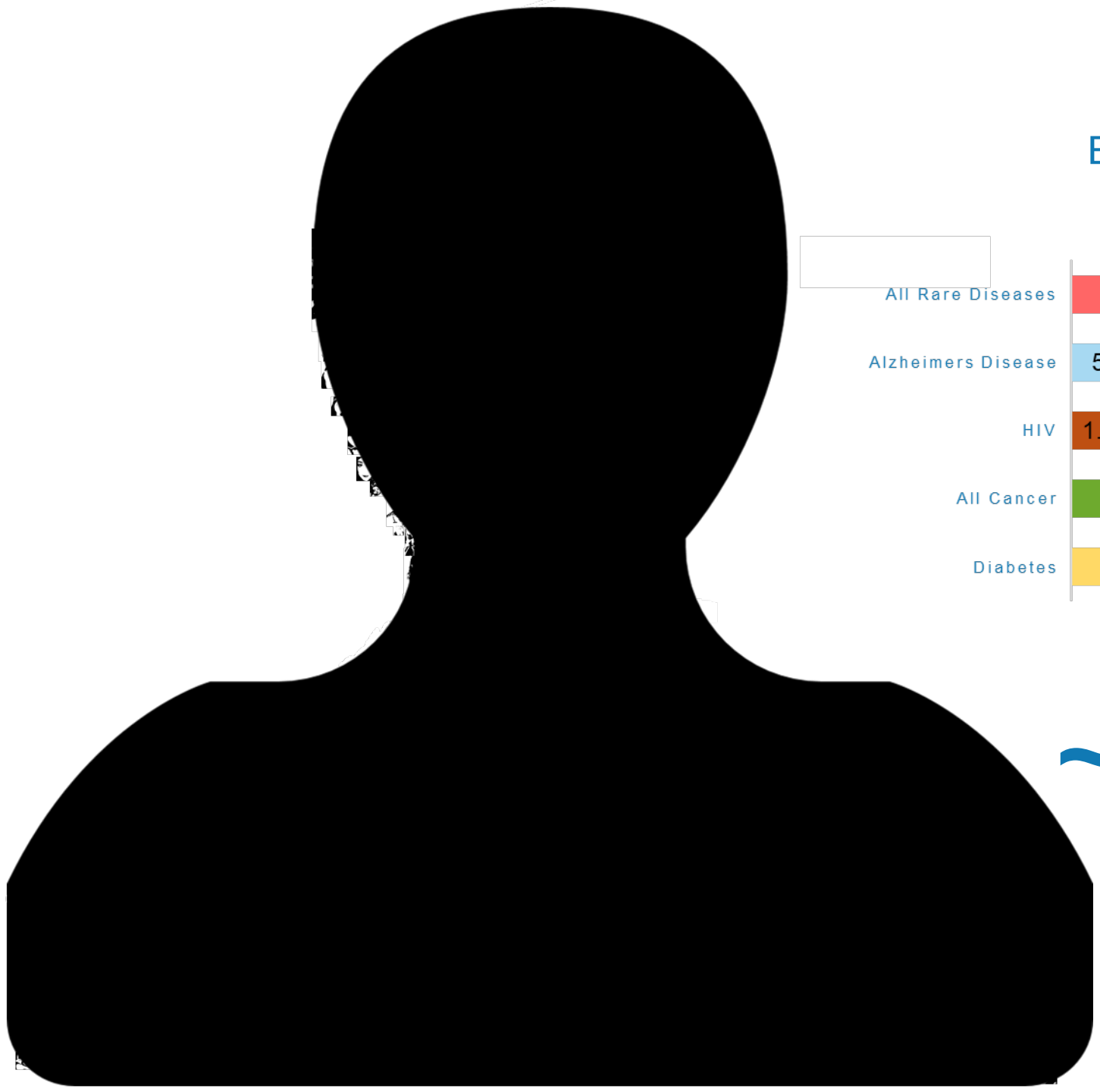
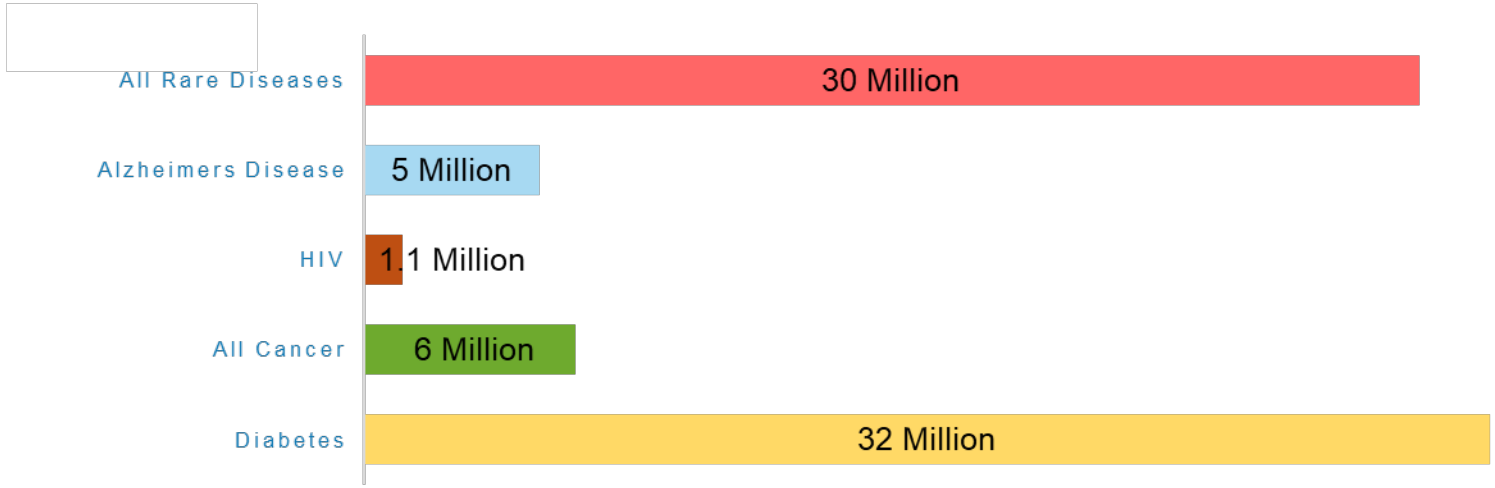
Disclaimer/Disclosure

- Disclaimer/Disclosure This presentation reflects the views of the presenter and does not represent NIH’s views or policies.
- no conflicts to disclose.

National Center for Advancing Translational Sciences

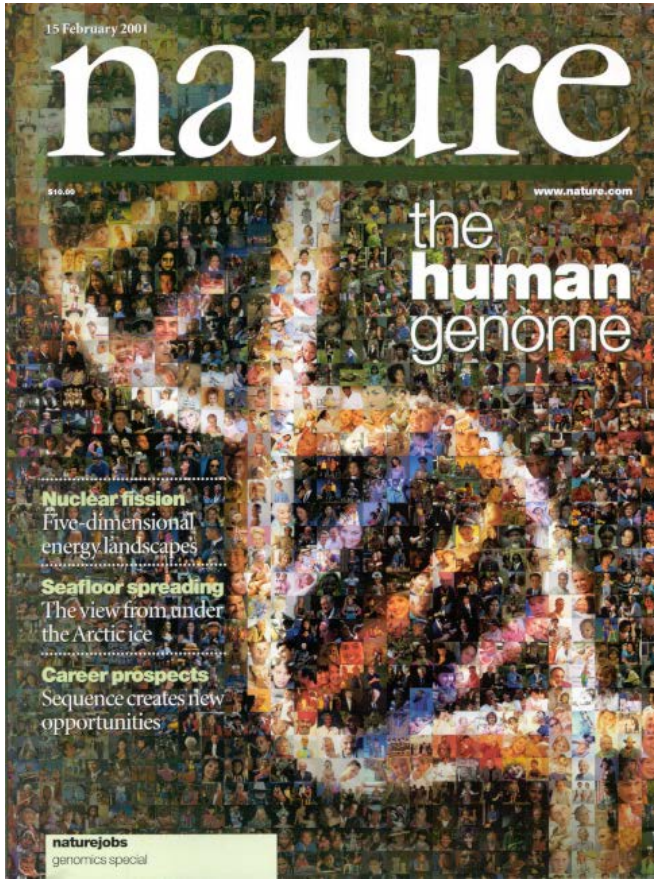


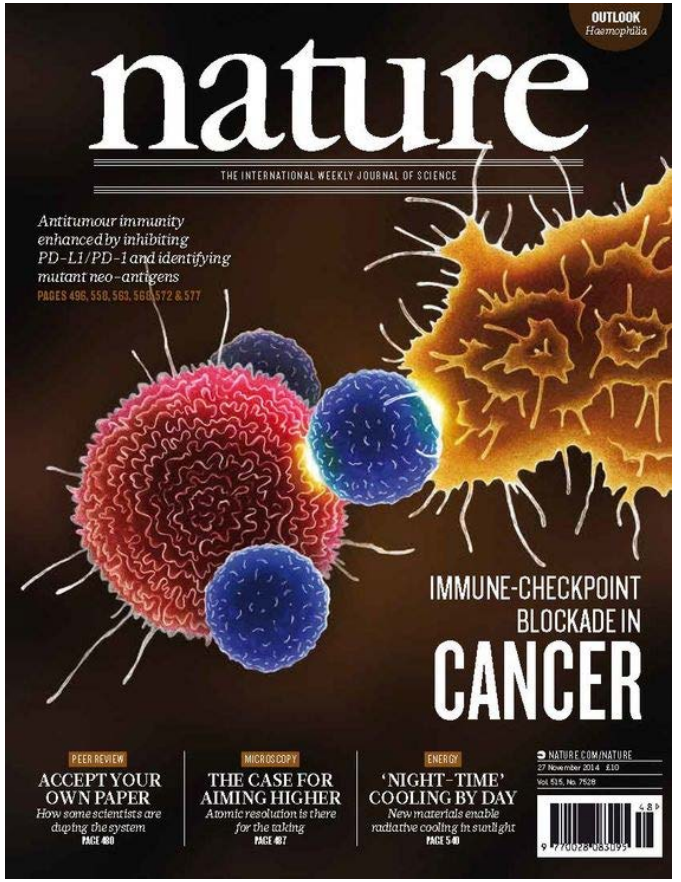
Estimated Prevalence Disease



~7,000 **+230 per year**

Science is advancing at breakneck speed…there are enormous opportunities

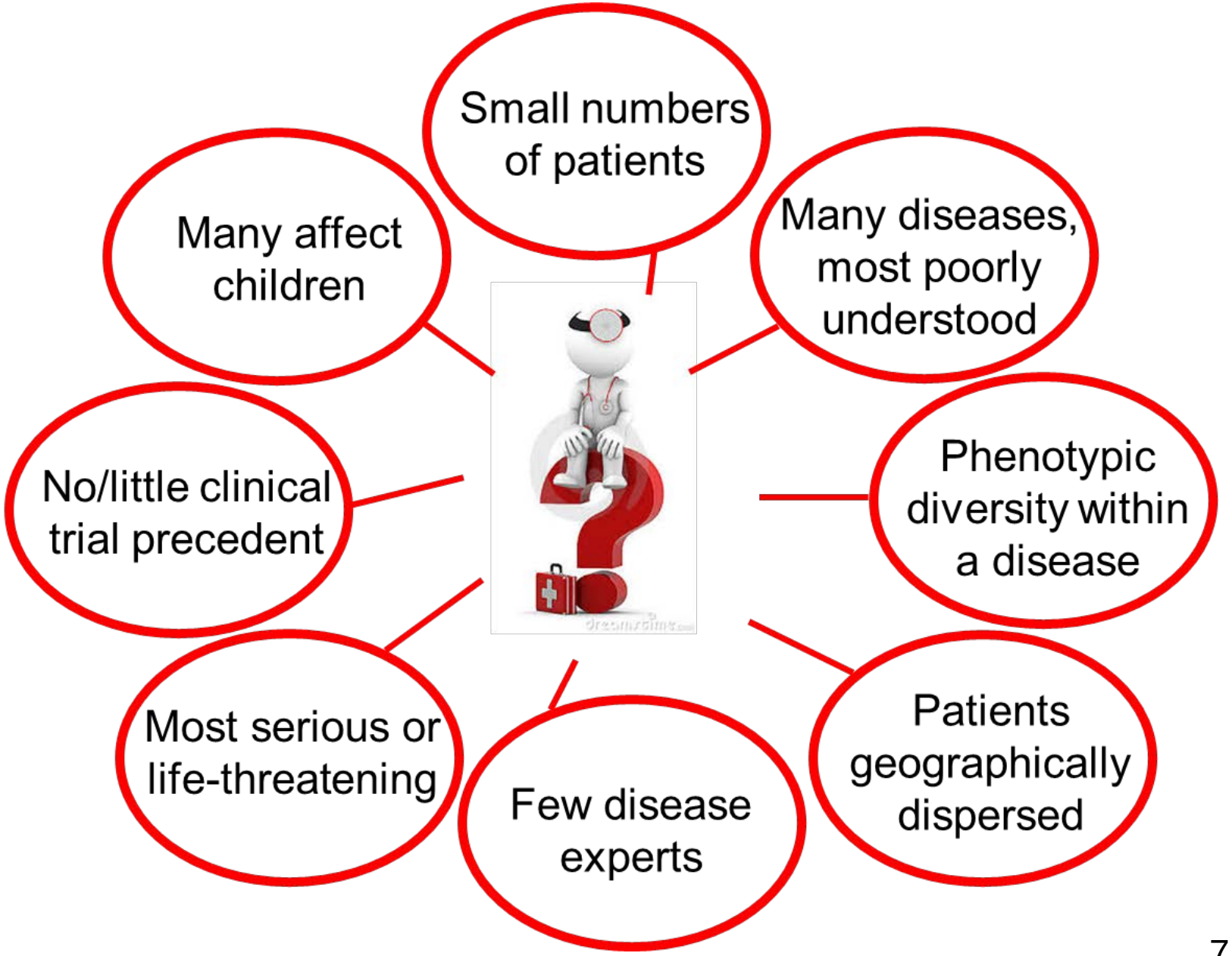




With these opportunities come the need to deliver on the promise of science for patients


CHALLENGE

Rare Diseases Research Challenges



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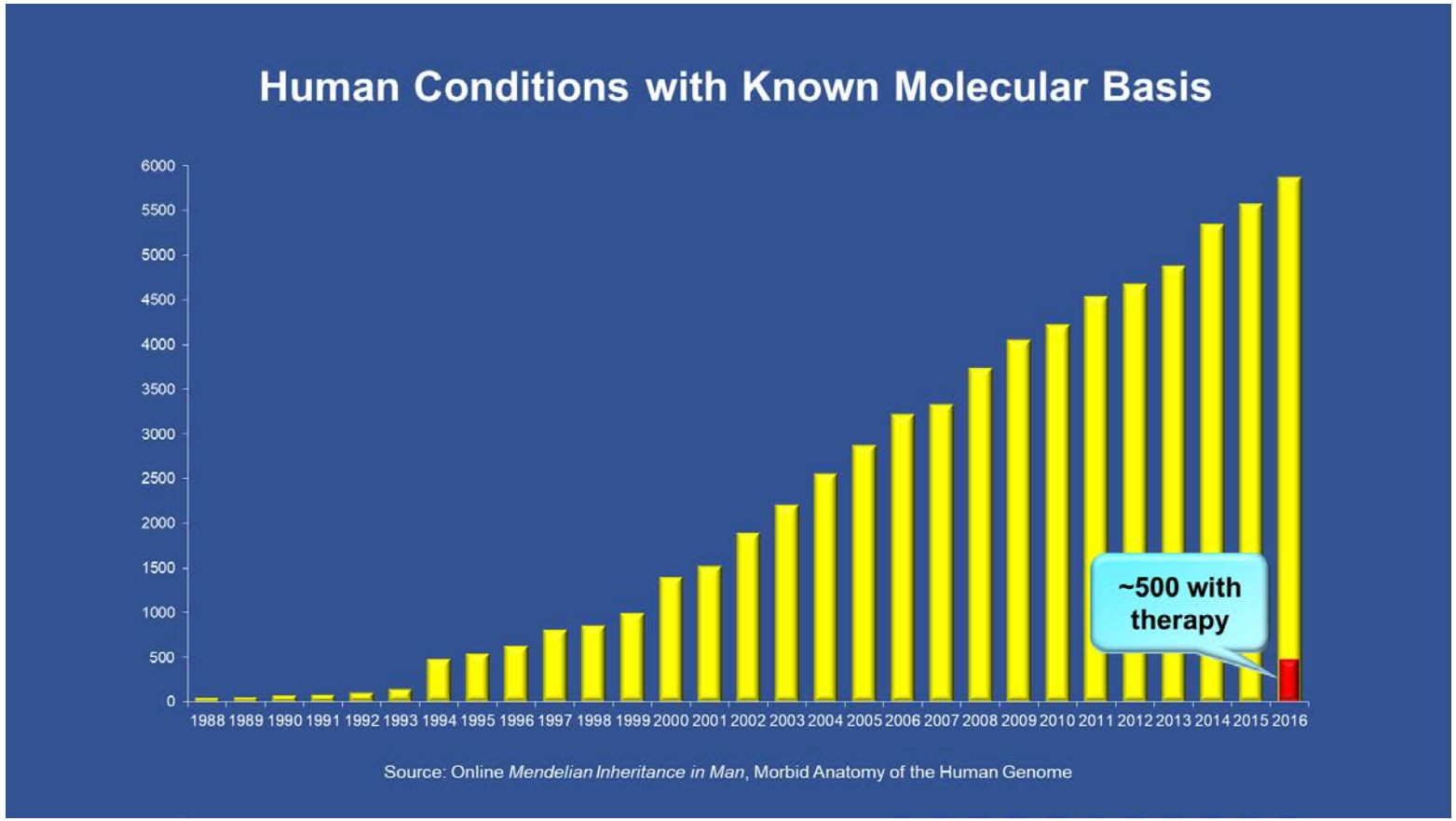
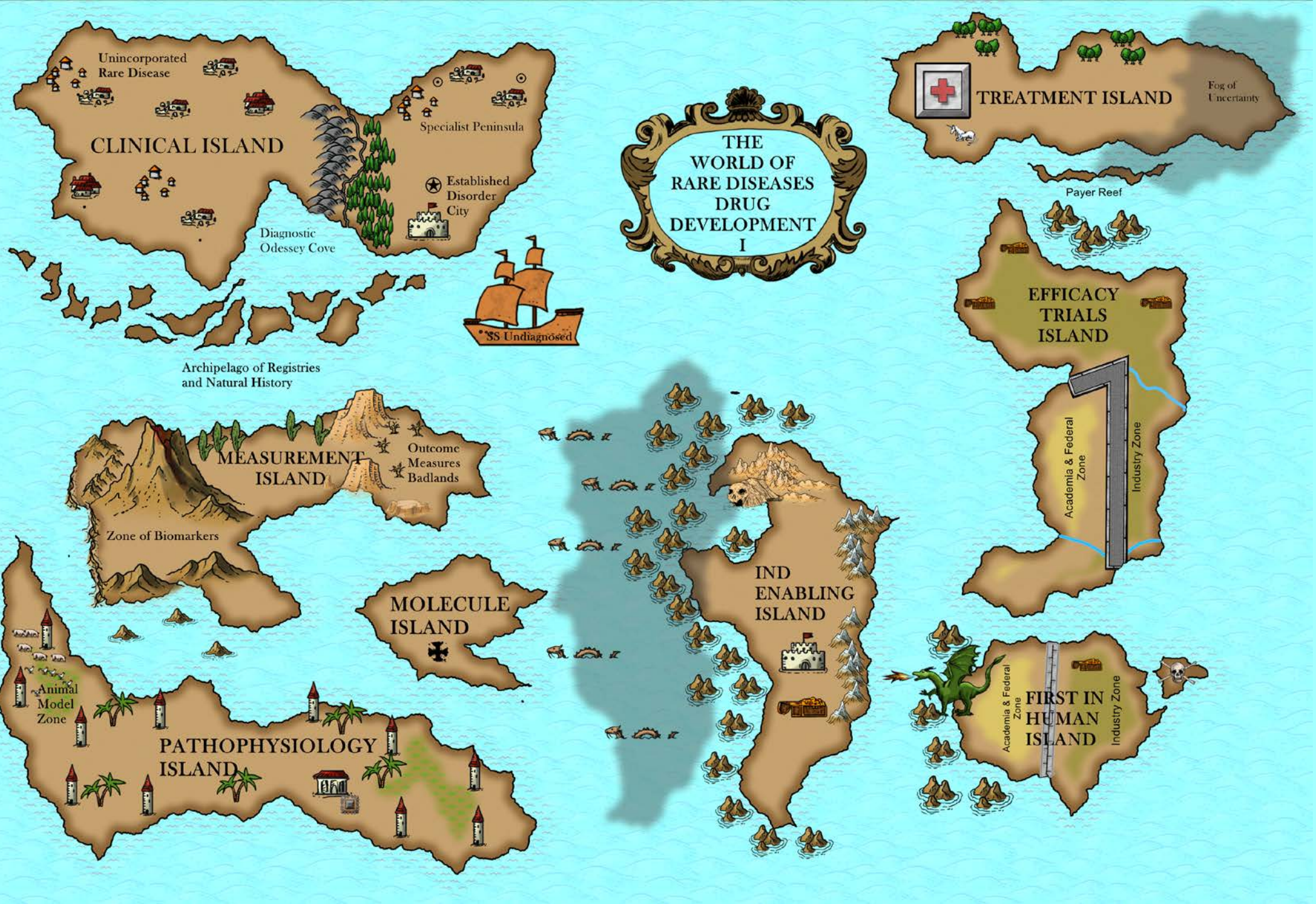


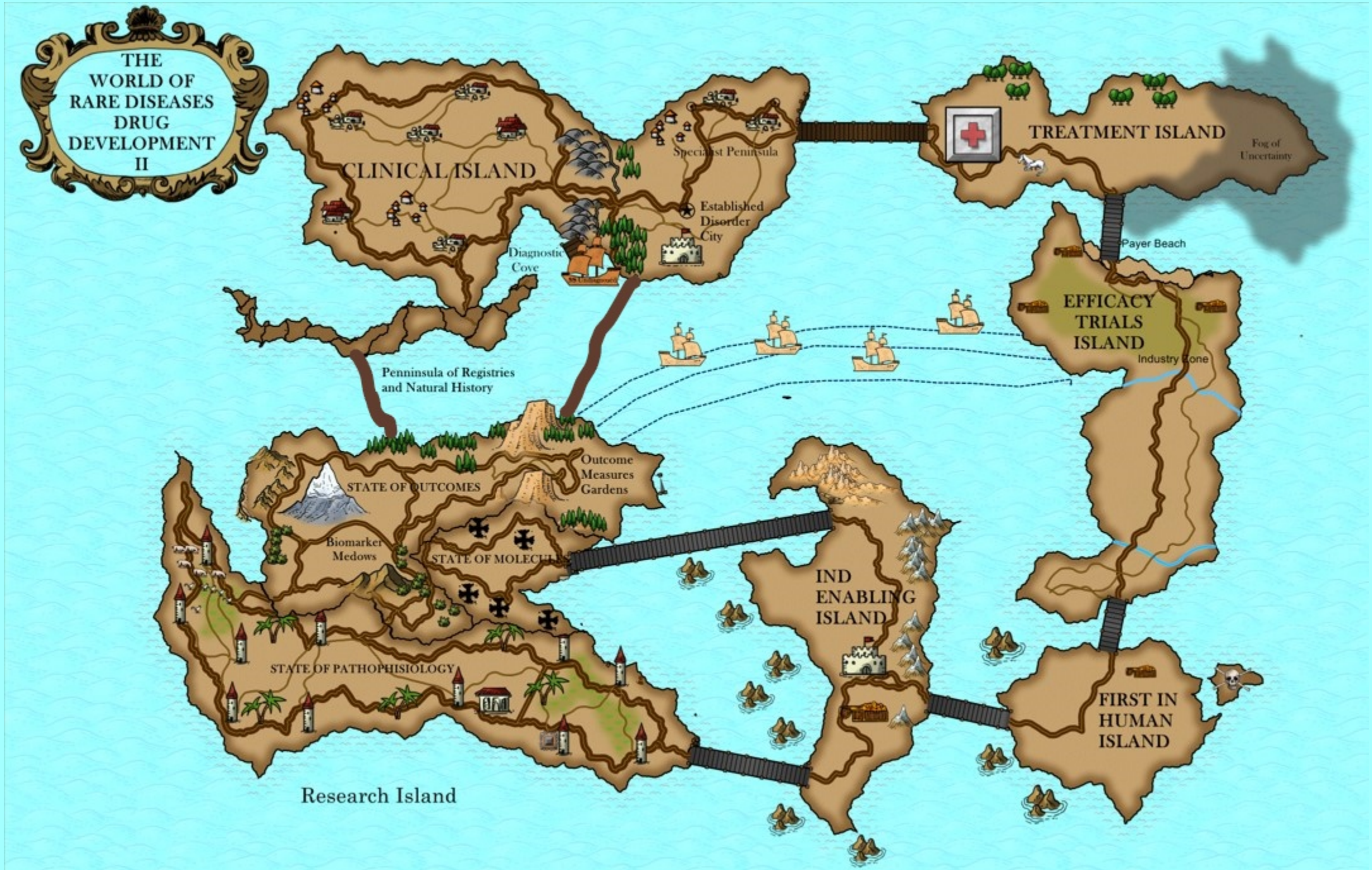
Treatments?

95% Have no treatment

5% Have a regulatorily approved Treatment

- At current rate 3-5 newly treatable diseases/yr…>1000 yrs to all


THE WORLD OF RARE DISEASES DRUG DEVELOPMENT I
Unincorporated Rare Disease
CLINICAL ISLAND
Specialist Peninsula
Established Disorder City
Diagnostic Odessey Cove
SS Undiagnosed
Archipelago of Registries and Natural History
TREATMENT ISLAND
Fog of Uncertainty
Payer Reef
EFFICACY TRIALS ISLAND
Academia & Federal Zone
Industry Zone
MEASUREMENT ISLAND
Outcome Measures Badlands
Zone of Biomarkers
MOLECULE ISLAND
IND ENABLING ISLAND
Animal Model Zone
PATHOPHYSIOLOGY ISLAND
FIRST IN HUMAN ISLAND
Academia & Federal Zone
Industry Zone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IND ENABLING ISLAND
STATE OF PATHOPHISIOLOGY
FIRST IN HUMAN ISLAND
Research Island


SIEMENS
5746
5746
5918
5918
5544
5544

What is being done to address these many challenges?

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ORDR (Office of Rare Diseases Research)

“Accelerating rare diseases research to benefit patients”

ORDR facilitates coordination between multiple stakeholders in the rare diseases community, including scientists, clinicians, patients, and patient groups

ORDR – Programs

-Websites
-Database

Knowledge & Information

Genetics **A**nd **R**are **D**iseases (**GARD**) Information Center

Toolkit for Patient-Focused Therapy Development

Rare Diseases Registry (RaDaR) Program

Research & Collaboration

Rare Diseases Clinical Research Network (RDCRN)

Clinical Trial Readiness and Bench-to-Bedside Grants

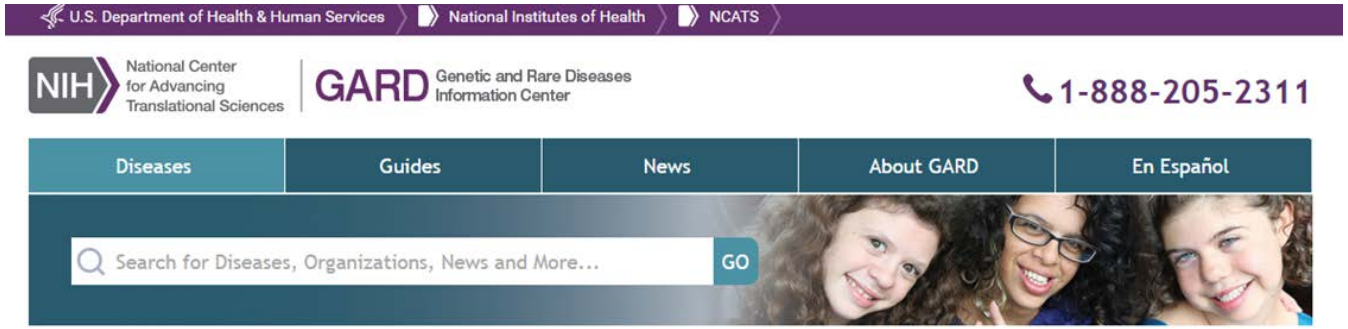
Scientific Conferences: Rare Disease Day at NIH, FDA/NCATS Gene Therapy Workshop, etc.

-Grants
-Meetings

Genetic **A**nd **R**are **D**iseases (**GARD**) Program

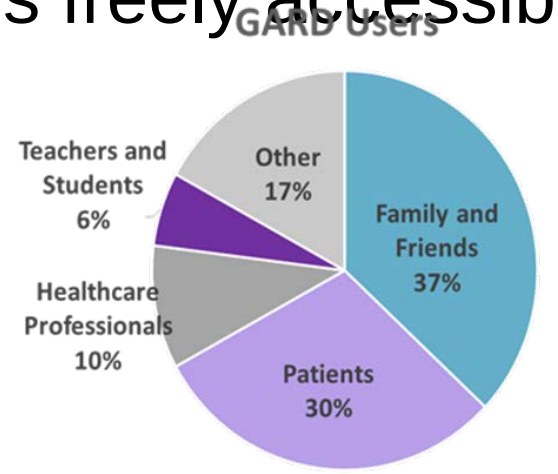
Established in 2002

GARD website:
https://rarediseases.info.nih.gov

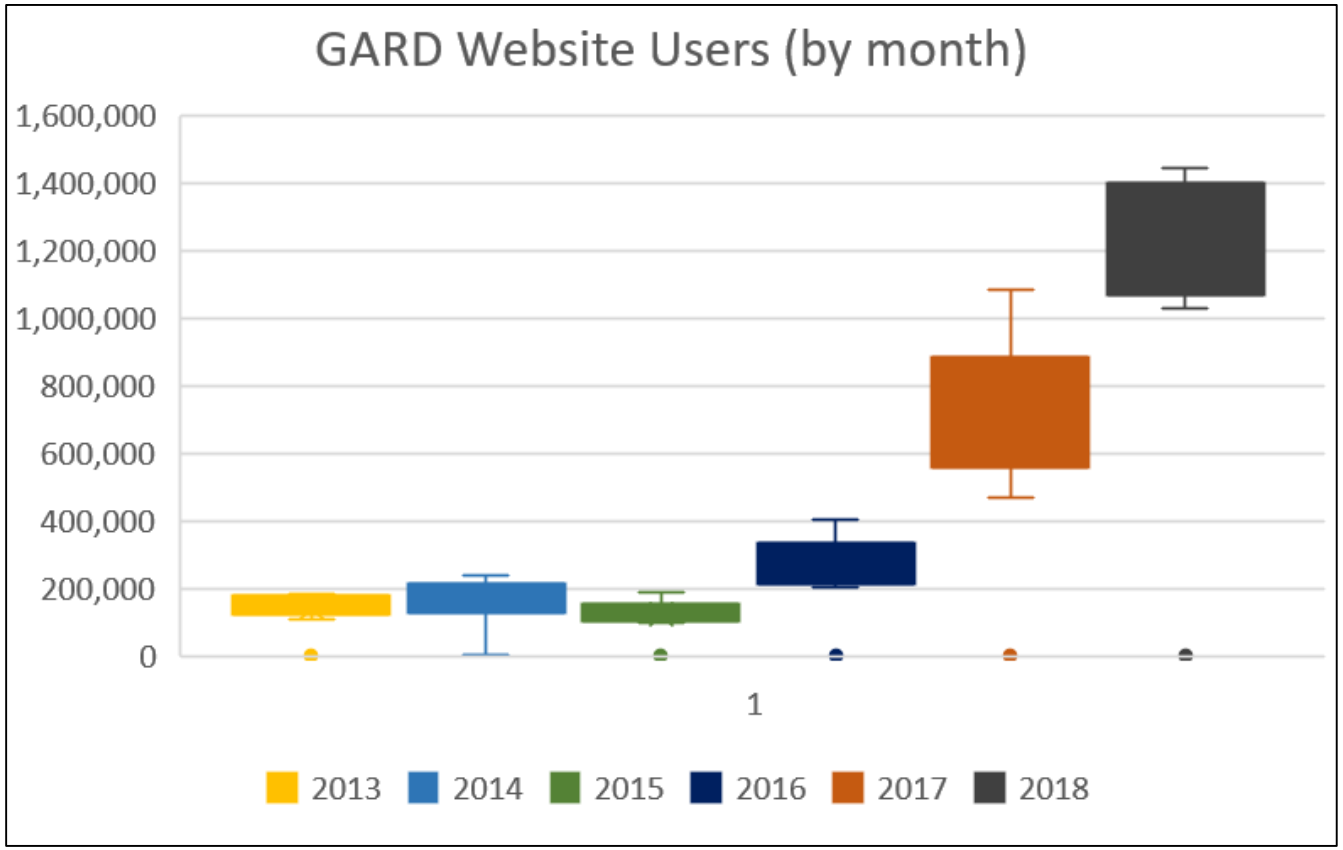


GARD's Mission: Provide comprehensive, plain-language information on rare diseases that is freely accessible to the public.

- **Website & Database**
- **Contact Center**

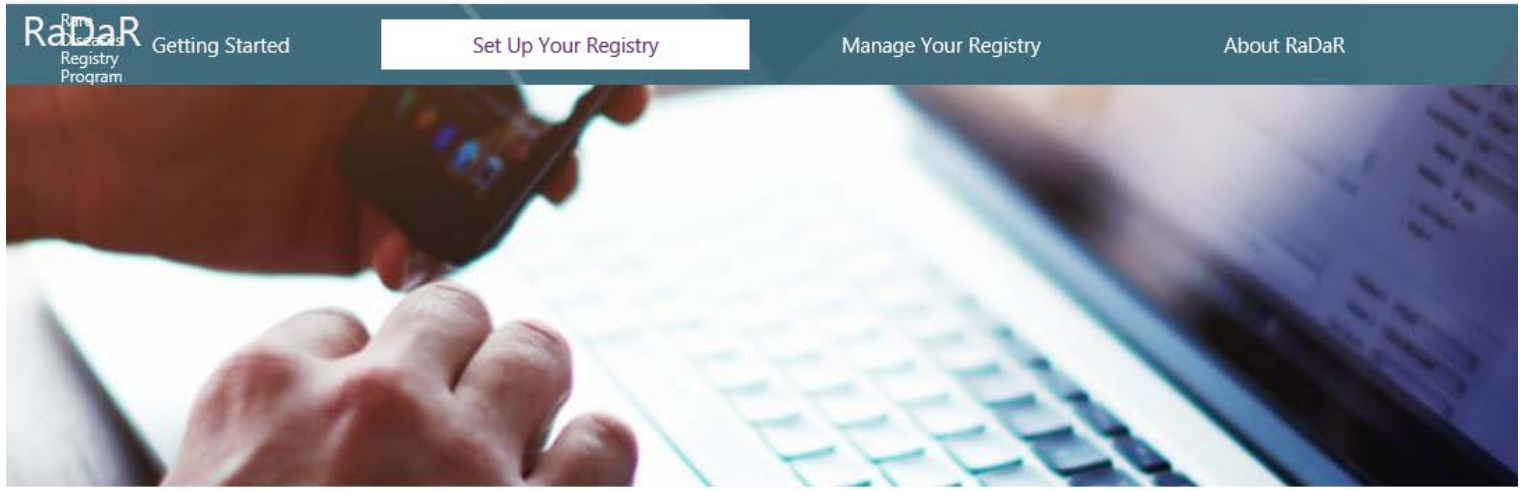


GARD – Utilization in recent years



What is RaDaR?

- RaDaR (Rare Disease Registry):
  - Mission: Provide an easy-to-use educational website that would enable new patient advocacy groups to adopt good quality practices earlier during registry development
    - Who: New and early-stage PAGs that are in the process of starting a registry
    - What: Promote standardization and data integration at the front-end (vs. cleaning up back-end data)
    - How: Develop an web platform that is easy-to-use and enables collaborative sharing of resources
  - Vision: “Registry-in-a-Box”
    - Provide stepwise instructions, best practices and examples, and templates/tools for registry building
    - Leverage existing knowledge, resources and assets from within the patient advocacy community
    - Focus on usability/UX for a patient organization audience



Set Up Your Registry

RaDaR guides you step-by-step help in building a registry for collecting participant contact and demographic information. This information will allow researchers to find people who are interested in participating in research studies.

Create Your Registry Plan
Step 1

Determine Who Should Join
Step 2

Develop the RIGHT Questions
Step 3

Decide How to Collect & Store Data
Step 4

Step 1. Create Your Registry Plan

1.0 Overview

Planning ahead is essential to establish clear goals for your registry and a strategy for collecting quality data. In this step you will define specific, detailed, and attainable goals for your registry; define your constraints; plan for roadblocks and identify possible solutions to them; and establish milestones to track your success.

Continue to Step 2. Determine Who Should Join after you finish Step 1

1.1 Set Your Goals

Before creating a registry and setting your goals, determine whether a registry has already been created for your rare disease. Partnering with an existing registry allows for you to combine efforts, avoid "reinventing the wheel," and reduce redundancy. There are many ways to find out whether a registry currently exists for your rare disease.

- Search ClinicalTrials.gov using the term "registry" and the name of your rare disease
- Conduct a general internet search
- Contact patient advocacy groups for the disease
- Contact the Rare Disease Patient Registry Coordinators
- Search the Registry of Patient Registries (RoPR) of the Agency for Healthcare Research and Quality (AHRQ)
- Search the RD-Connect Registry & Biobank Finder
- Reach out to the Genetic and Rare Diseases Information Center (GARD)

After confirming that no other registry exists for your disease, you can start creating your registry.

Be specific, detailed, and realistic about the goals for your registry and clear about what you plan to do with the information you collect. Focus on how you can use your registry to organize your patient community and connect patients and researchers. Have a long-term vision for capturing detailed participant medical information to support the development of new treatments. Below are some goals to consider. Use the RaDar Tool: Registry Plan Template for help getting started.

Short-term registry goals:

- Identify patients who are interested in participating in research studies.
- Describe the personal characteristics of participants in your registry.
- Contact participants to inform them about new studies.

Long-term registry goals:

- Document patient medical history.
- Discover trends and common needs of participants.
- Improve scientific understanding of the disease.

Visit the Resources section of this step for resources that provide additional information for setting up your registry.

NIH National Center for Advancing Translational Sciences | Toolkit For Patient-Focused Therapy Development

Search for tools Search

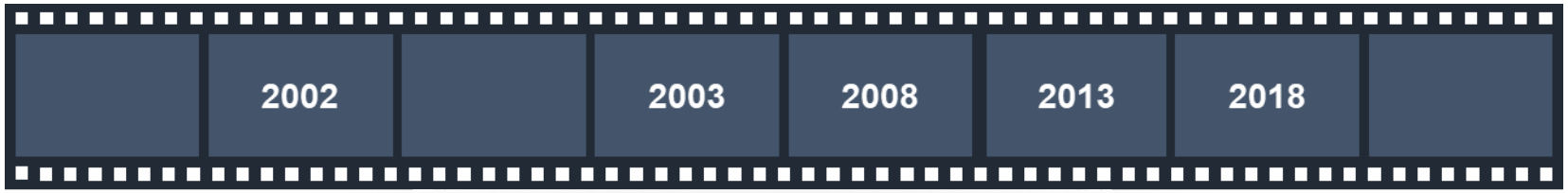
Contact Us

Home | Getting Started | Discovery | Preparing for Clinical Trials | Clinical Trials and FDA Review | After FDA Approval | About

Working Together to Advance Rare Diseases Research

This Toolkit was developed to provide your patient group with the tools needed to advance medical research. Our goal is to ensure that patients are engaged as essential partners from beginning to end of the research and development process. This is a living site where you will find tools being developed for and by patient groups in concert with their academic, government, industry and advocacy partners. Read more about why NCATS developed this Toolkit.

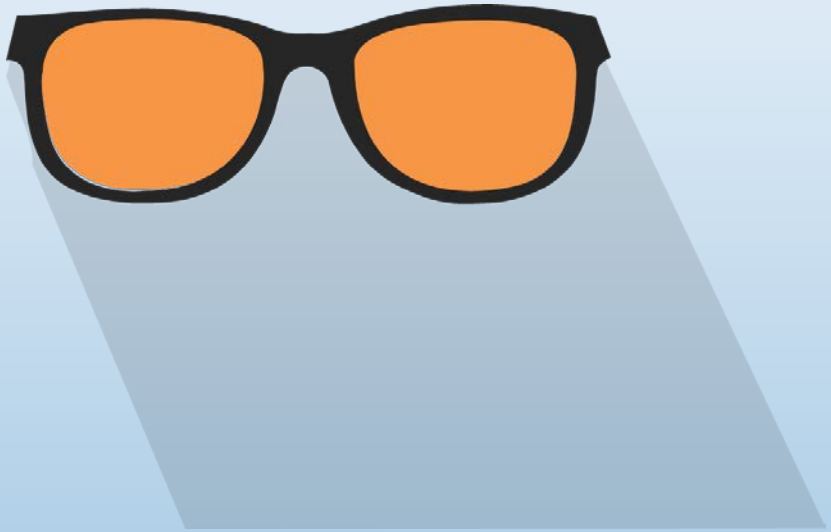
Rare Diseases Clinical Research Network



Beginning	Early Years	Development	Future
Rare Diseases Act of 2002 (Public Law 107-280) Established “RDCRC’s of Excellence”	• First RFA released • 7 consortia funded	• 2008 – 19 • 2013 – 22 • 31 Individual consortia • 238 Disorders • >40,000 Participants	RFA-TR-18-020 RFA-TR-18-021

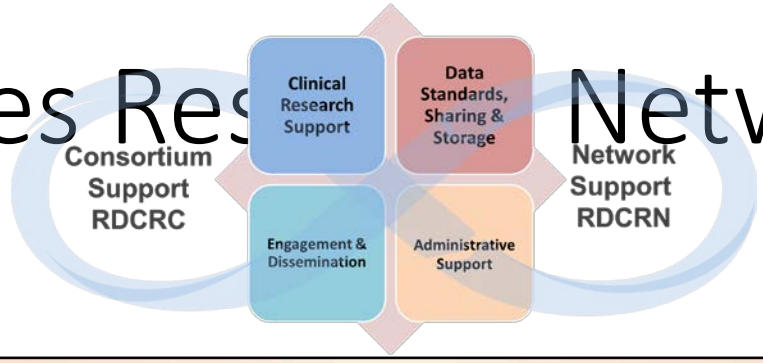
RDCRN

2019

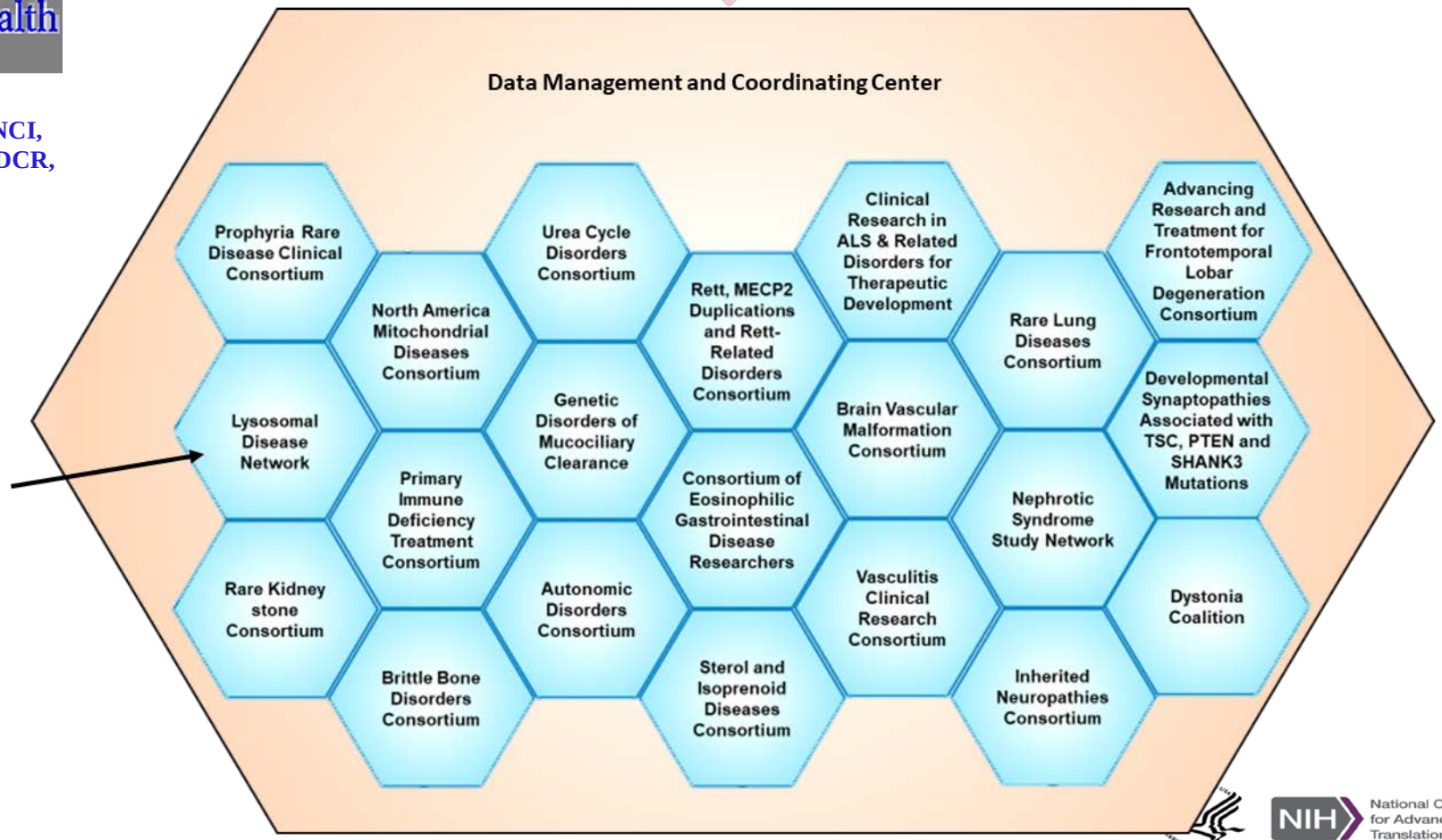


The RDCRCs are intended to advance the diagnosis, management, and treatment of rare diseases with a **focus on clinical trial readiness**. Each RDCRC will promote highly collaborative, multi-site, patient-centric, translational and clinical research with the intent of addressing unmet clinical trial readiness needs.

F Diseases Res Network



NIH Partners
NCATS, NINDS, NICHD, NCI, NHLBI, NIMH, NIDDK, NIDCR, NIAMS,NIAID, ODS



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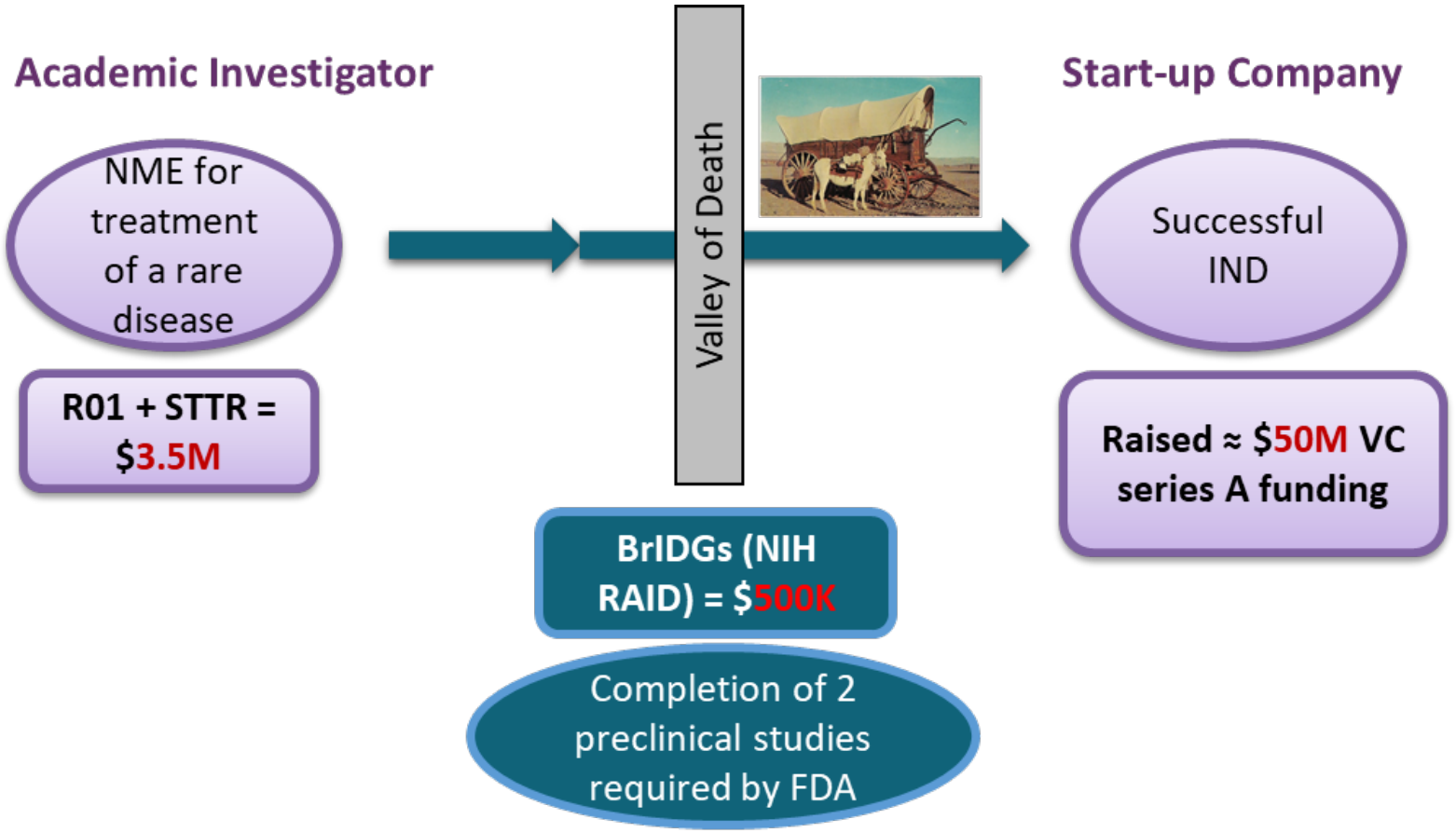
STTR/TRND/BrIDGs Project De-Risking

Minimum Time and Funding; Maximum Impact



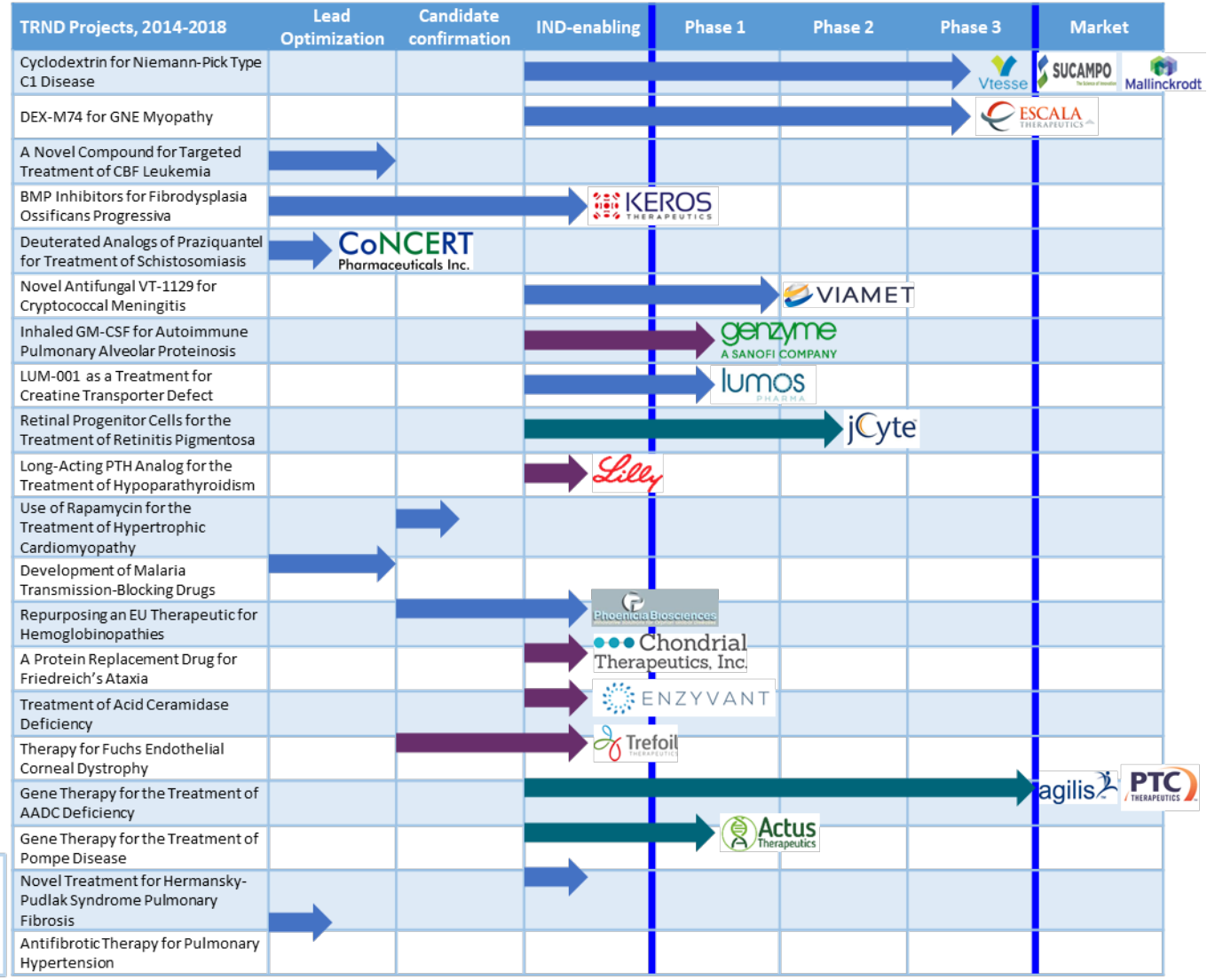
Therapeutics for Rare and Neglected Diseases (TRND) Program

- Model: Comprehensive drug development collaboration between the Division of Pre-Clinical Innovation (DPI) and extramural labs with disease-area/target expertise
- Projects
  - May enter at various stages of preclinical development
  - Disease must meet FDA orphan or WHO neglected tropical disease criteria
  - Taken to stage needed to attract external organization to adopt to complete clinical development/registration, max Phase 2a
  - Milestone driven
  - Therapeutic modalities: small molecules, proteins, peptides, oligonucleotides, gene therapy, antibodies, recombinant proteins
  - Aims to de-risk technology and develop new generally applicable platform technologies and paradigms
- Eligible Applicants
  - Academic, Nonprofit, Government Lab, Biotech/Pharma
  - Ex-U.S. applicants accepted

Bridging Interventional Development Gaps (BrIDGs) Program

- Model: Collaboration between DPI and extramural labs (Formerly NIH-RAID Program)
- Projects
  - Enter with clinical candidate identified
  - Any disease eligible
  - Gap analysis followed by data generation using DPI resources and expertise to generate data necessary for IND filing
  - Exit at or before IND
  - Milestone driven
  - Therapeutic modalities: small molecules, peptides, oligonucleotides, gene therapy, antibodies, recombinant proteins
- Eligible Applicants
  - Academic (U.S. and Ex-U.S.), Non-Profit, SBIR eligible businesses

TRND Projects



Small Molecule
Biologic
Gene and Cell Therapy

National Center for Advancing Translational Sciences

Thank you!

Rare Disease Programs Points of Contact

NIH National Center for Advancing Translational Sciences

Assay Development and Screening Technology (ADST)

ADST is designed to advance therapeutic drug development through research and development of innovative assay (test) designs and chemical library screening methods.
ADST webpage: ncats.nih.gov/adst

Contact: Nicole Spears, BS, Scientific Program Analyst
Email: Nichole.spears@nih.gov

CATS Chemical Genomics Center NCG

NCGC researchers advance small molecule therapeutic development through assay (test) design, high-throughput screening and medicinal chemistry.
NCGC webpage: ncats.nih.gov/ncgc

Contact: Matthew Hall PhD, Group Leader
Email: hallma@mail.nih.gov

Therapeutics for Rare and Neglected Diseases (TRND)

The TRND pogrom supports pre-clinical development of therapeutic candidates intended to treat rare/neglected disorders with the goal of enabling an Investigational New Drug application.
TRND webpage: ncats.nih.gov/trnd

Contact: Donald ID PhD, Director, Therapeutic Development Branch
Email: askTDB@nih.gov

Tissue Chips for Disease Modeling and Efficacy Testing

The Tissue Chips for Disease Modeling initiative supports further development of tissue chip models of human disease that mimic the pathology in major human organs and tissues.
Tissue Chips for Disease Modeling and Efficacy Testing webpage:
ncats.nih.gov/tissuechip/projects/modeling

Contact: Danklo Tagle, PhD, Director for Special Initiatives
Email: danilo.tangle@nih.gov

Bridging Interventional Development Gaps (BrIDGs)

The BrIDGs program assists researchers in advancing promising therapeutic agents through late-stage pre-clinical development toward an Investigational New Drug application and clinical testing.
BrIDGs webpage: ncats.nih.gov/bridgs

Contact: Donald Lo PhD, Director, Therapeutic Development Branch
Email: askTDB@nih.gov

Discovering New Therapeutic Uses for Existing Molecules (New Therapeutic Uses)

The New Therapeutic Uses program aims to improve the process of developing new treatments and cures for disease by finding new uses for assets that already have cleared several key steps along the development path.
New Therapeutic Uses webpage: ncats.nih.gov/ntu

Contact: Bobbie Ann Mount, PhD, Program Officer
Email: bobbieann.mount@nih.gov

Srnall Business Innovation Research (SBIR) and Small Business Technology Transfer (STTR)

These support NCATS' mission to transform the translational science process by helping small businesses develop and commercialize new technologies.
STTR webpage: ncats.nih.gov/smallbusiness/about

Contact: Lili Portilla, MPA, Director for Strategc Alliances
Email: portilll@mail.nih.gov

RDCRN — **Contact:** Tiina Urv

Director Office of Rare Diseases Research

Anne Pariser – Email: anne.pariser@nih.gov