The NIH/NCATS GRDR^{SM} Program

Global Rare Diseases Patient Registry Data Registry

Collaborative Program for the Patients by the Patients

Second International Rare Diseases Research Consortium (IRDiRC)
Shenzhen, China
November 6-9, 2014

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ORDR/NCATS/NIH

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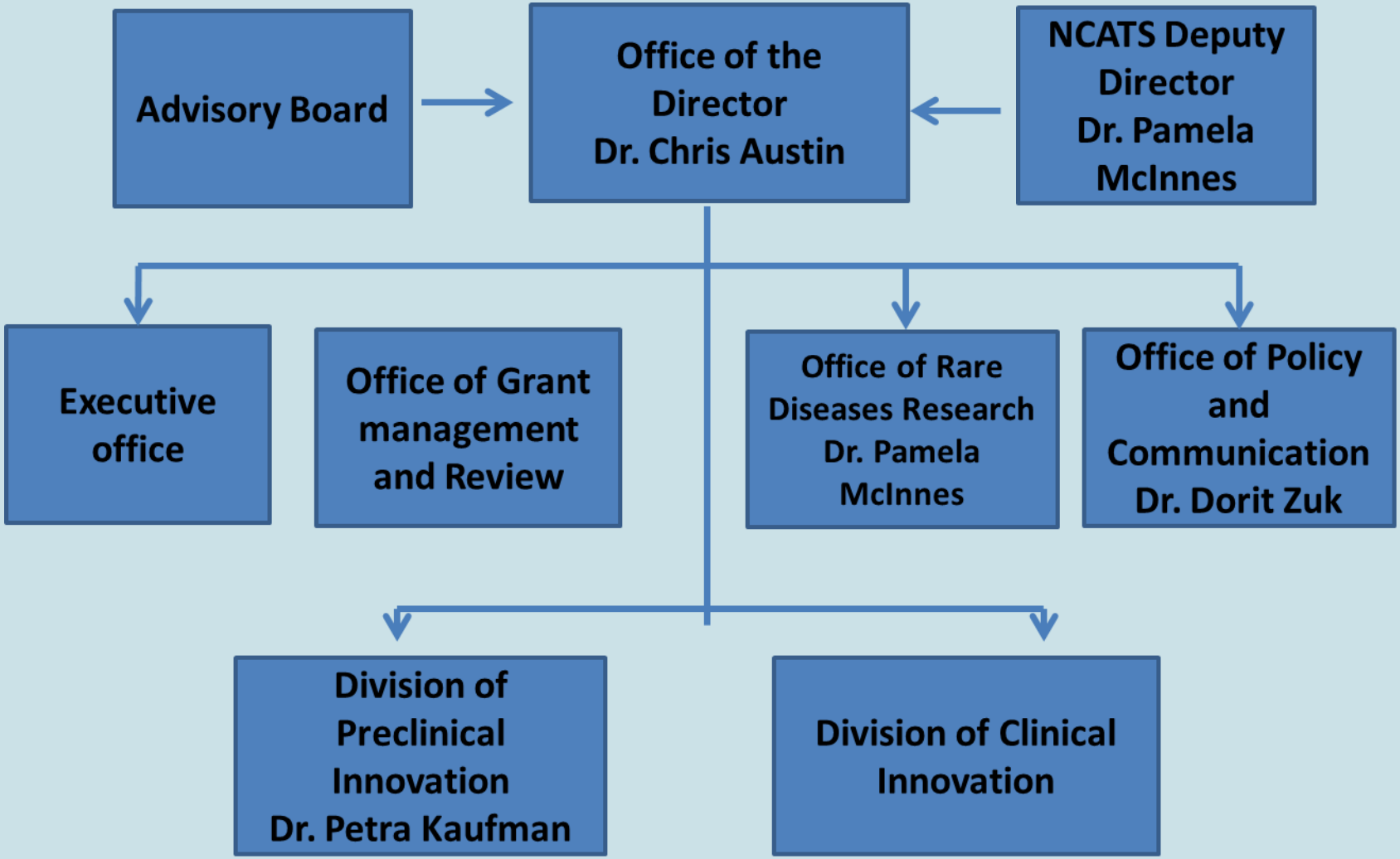

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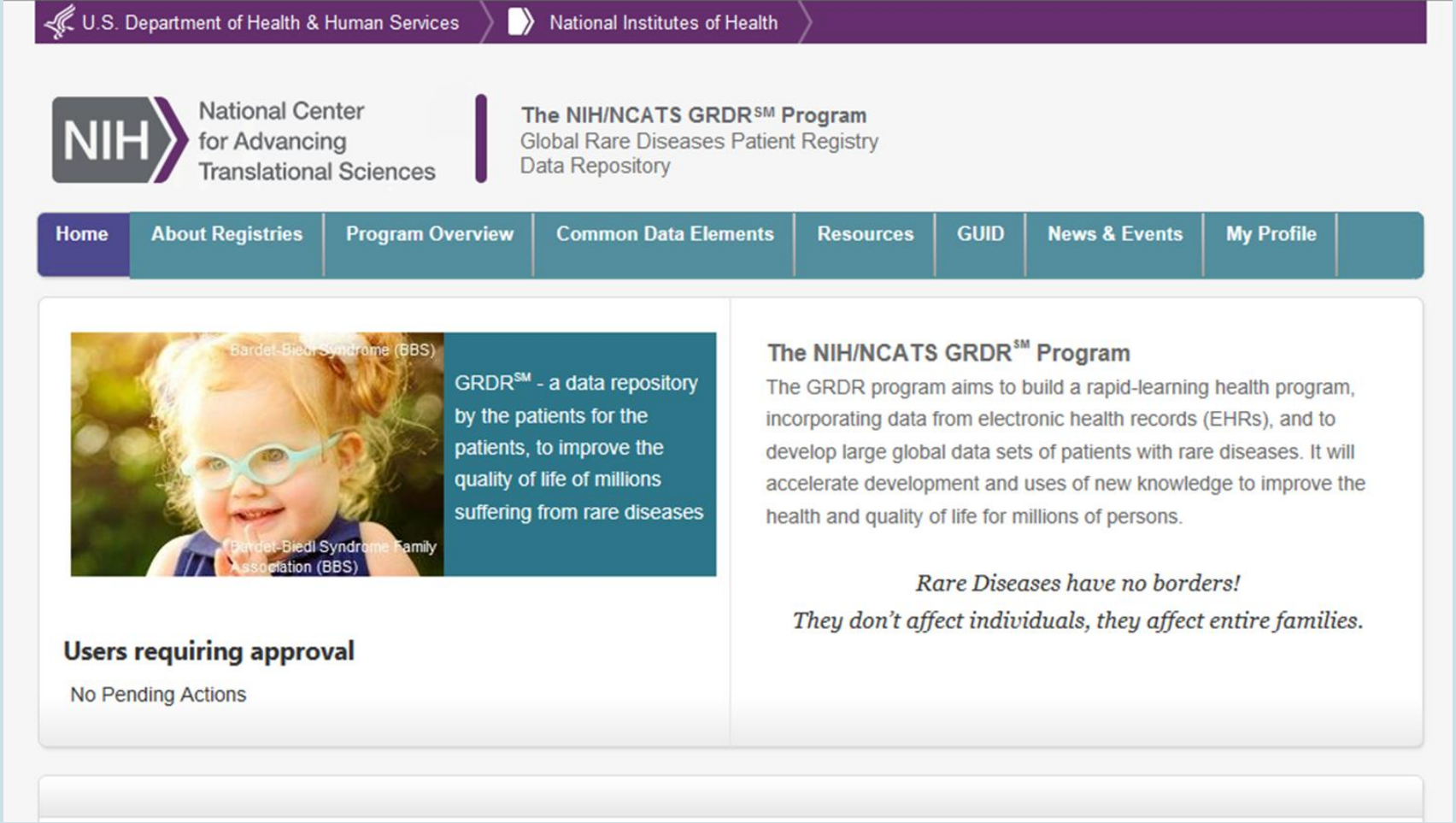

- **Background**
- **History**
- **Lessons learned**
- **Current state**
- **GRDR scientific & clinical value**
- **Future plans**

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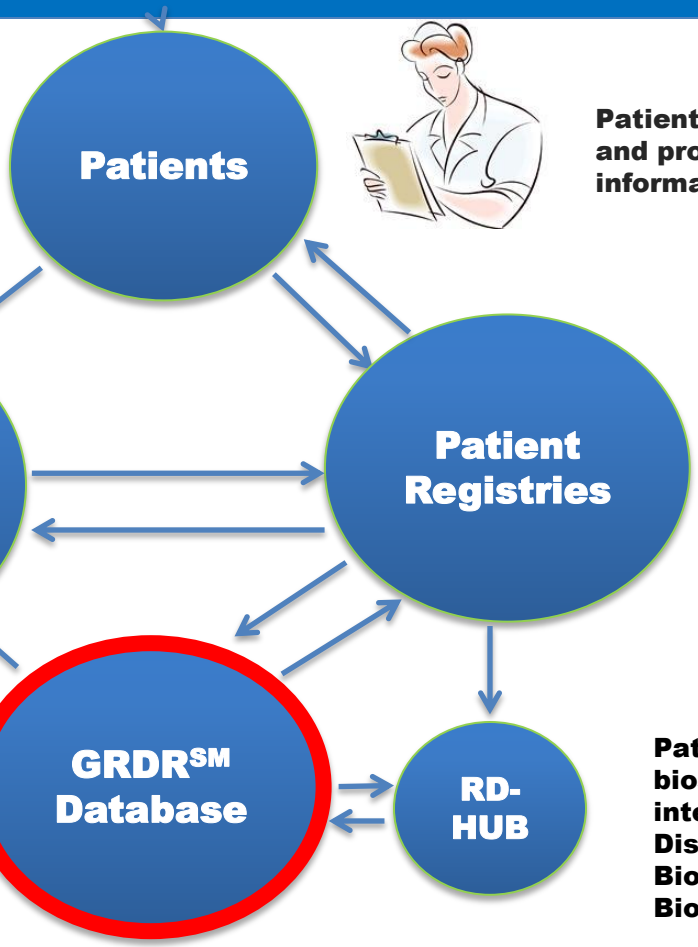
Global Rare Diseases Patient Registry Data Repository



Registry owners notify identified participants and direct them to study PI

Patients join a registry and provide health information

Researchers conduct various biomedical studies within & across diseases

Registry managers de-identify collected patient data and biospecimens, and assign Global Unique Identifier (GUID)

De-identified patient data is shared with GRDR[SM] program staff

Patient data linked to biospecimens via the GUID interfacing with Rare Diseases Human Biospecimens/ Biorepositories (RD-HUB)

GRDR aggregates, maps data to CDEs & national standards, integrates patient clinical information and provides access to approved researchers

Linking to other databases

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GRDR^{SM} Data Repository

https://grdr.ncats.nih.gov/

Common Data Elements →

- Current contact information
- Socio-demographic information
- Diagnosis
- Family history
- Birth and reproductive history
- Anthropometric information
- Patient-reported outcome
- Medications/devices/health services
- Clinical research and biospecimen
- Communication preferences

- ***Template Patient Informed Consent*** **for participating in Patient Registries**

- **Global Unique Identifier-GRDR-GUID**

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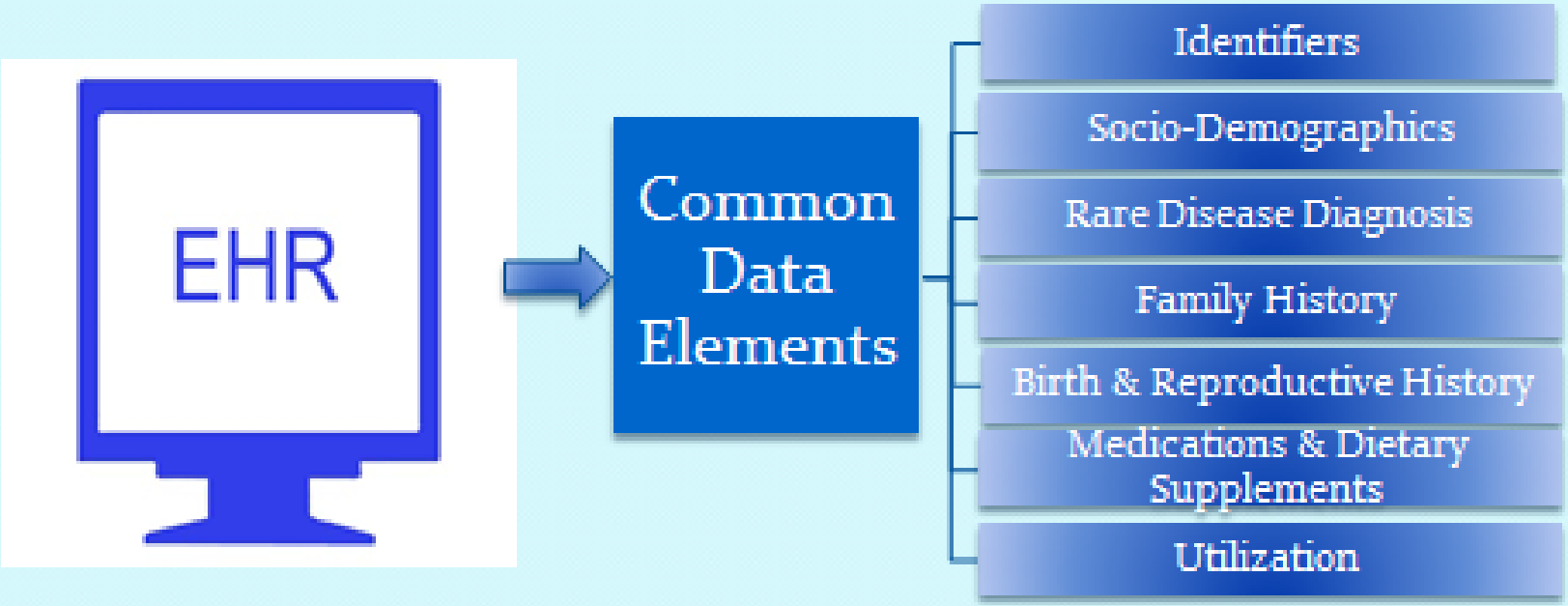
Integrating Electronic Health Record



Ascertain whether a hybrid between the EHR and the organization's registry can be used to populate the GRDR repository

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Keys for success

Things that we know we don't know and things that we don't know we don't know

- **Learn from your previous experiences**
- **Start small and go big**
- **Recognize that you are not everything and about everything**
- **Expertise resides all over- use them, don't ignore them**
- **Your way is not THE WAY. There are different models**
- **Follow the 3 big Cs: Communicate, Coordinate, & Collaborate (CCC)**
- **Don't give up; with persistence, together, we can make it**

- **Recognize that the patients are the centerpiece**

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https://grdr.ncats.nih.gov/

From a pilot project

Lessons learned

to a GRDR^{SM} Program

The repository is being developed with an open-science principle that supports clinical research, population health, and improvements in health care for patients with rare diseases.

- Map data of existing registries
- Develop tools for the RD community

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GRDR Program Collaboration

- Through its GRDR^{SM} program, NCATS staff currently are working in collaboration with a team from the Children's Hospital of Philadelphia, and the participating groups to create a standardized and interoperable data repository.

The Children's Hospital *of* Philadelphia

- The NIH/NCATS/GRDR^{SM} program is designed to advance research for rare diseases and, through application of scientific insights gained, to further research for common diseases, as well.

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Collaborate

pcori

An independent research institute authorized by Congress through the Patient Protection and Affordable Care Act.
Funds comparative clinical effectiveness research (CER) that engages patients and other stakeholders throughout the research process.
PCORI helps people make informed health care decisions, and improves health care delivery and outcomes, by producing and promoting high integrity, evidence-based information that comes from research guided by patients, caregivers and the broader health care community.

Patient-Centered Outcomes Research Institute

NORD Registry Platform Overview

- Empowering Patient Organizations to manage the collection of disease natural history information in order to accelerate research within a disease and across diseases
- Support for standards, including GRDR, PROMIS, and other NIH instruments
- Developed and hosted by NORD
- Features to support research-grade longitudinal data collection
- Web-based reporting, visualization, and management tools
- Collaborative development planning with members
- Support provided by NORD through the full lifecycle of conducting a successful Natural History Study

NORD National Organization for Rare Disorders

rarediseases.org

Genetic Alliance

CoRDS Registry

Coordination of Rare Diseases at Sanford

SANFORD RESEARCH

Marshfield Clinic®

Don't just live. **Shine.**

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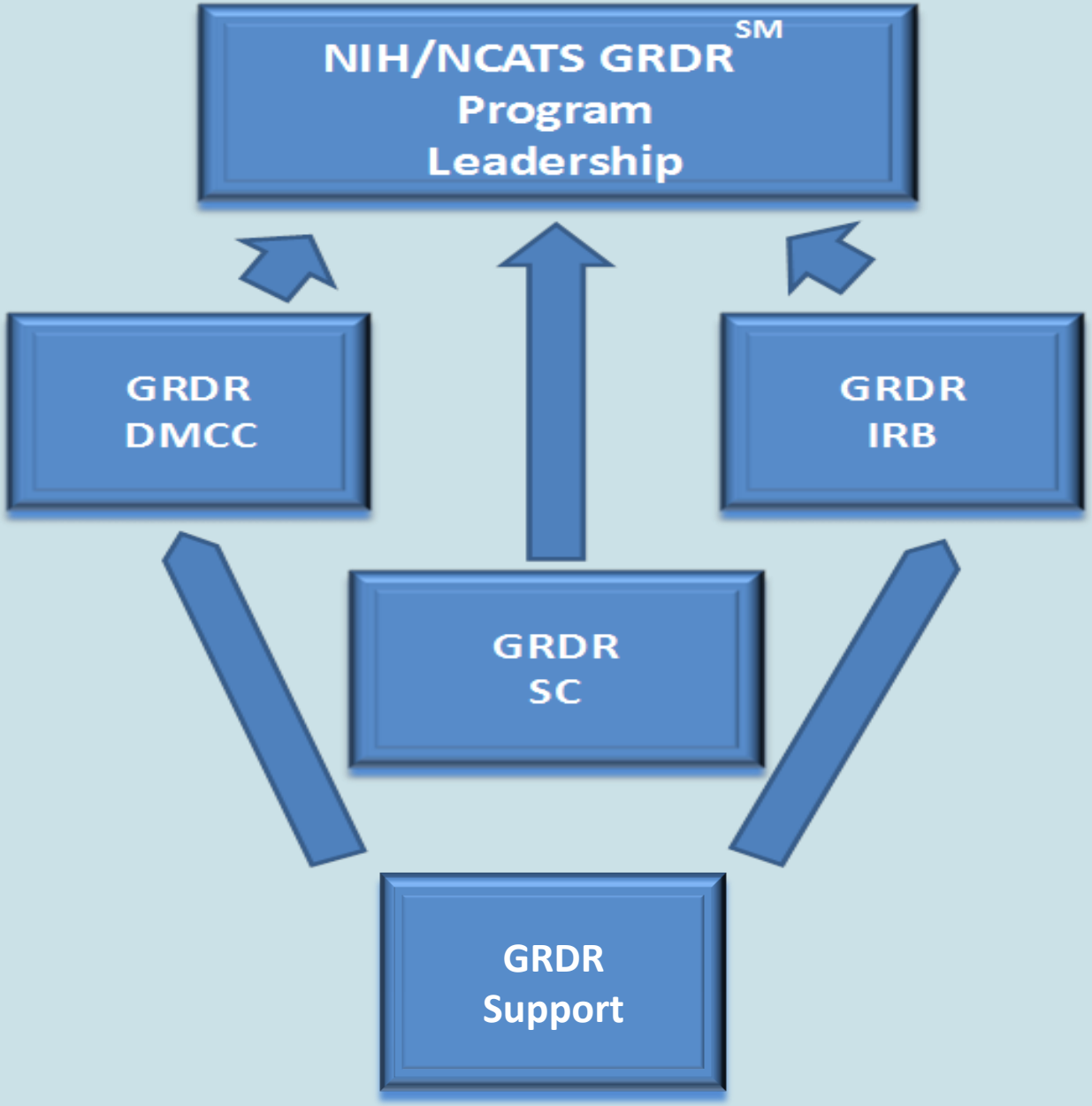
Collaborate

GRDR℠ Program Governance

Organizational Chart



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GRDR^{SM} Steering Committee

- **Charter**
- **Policy formation**
  - Data submission
  - Data access
  - Access to tools & information
  - Privacy
  - IRB
  - Membership
  - Others, as defined by SC and program leadership
- **Serves as a resource for consultation and recommendation for the GRDR leadership regarding network operational matters**
- **Provides input to the development and progress of the program's specific phases.**
- **Provides revisions and recommendations for existing and new operations, activities, collaborations and partnerships with other stakeholders**.

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GRDR^{SM} Repository

Mapping

Mapping domains include:

- ❑ **Demographics and core data**
- ❑ **Disease specific data**
  - **Diagnosis**
  - **Diagnostic Testing (labs and images)**
  - **Therapeutic interventions (medicines, procedures)**
  - **Others (clinical observations, family history, etc**.)

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Mapping Process

- Registry provides data dictionary
- DMCC team works with each registry on mapping there data
  - Mapping to GRDR CDEs
  - Mapping to standard terminologies such as SNOMED CT, LOINC, RXNORM, ICD-9, PROMIS etc.
  - Cumulative record of registry-disease specific data- to develop future CDE
  - ETL process (Extract, Transform, Load)
  - (GUID vs. registry-specific)
  - Prepare Data for Query

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Global Unique Identifier – GUID

- Required PII
- In order to generate a GUID for the subject, the following PII is required (these elements are included in the ORDR/GRDR list of CDEs):
  - Complete legal given (first) name of subject at birth
  - Complete legal additional name of subject at birth (if the subject has a middle name)
  - Complete legal family (last) name of the subject at birth
  - Day of birth (1-31)
  - Month of birth (1-12)
  - Year of birth (####)
  - Name of city/municipality in which subject was born
  - County of birth
  - Physical sex of subject at birth (M/F)

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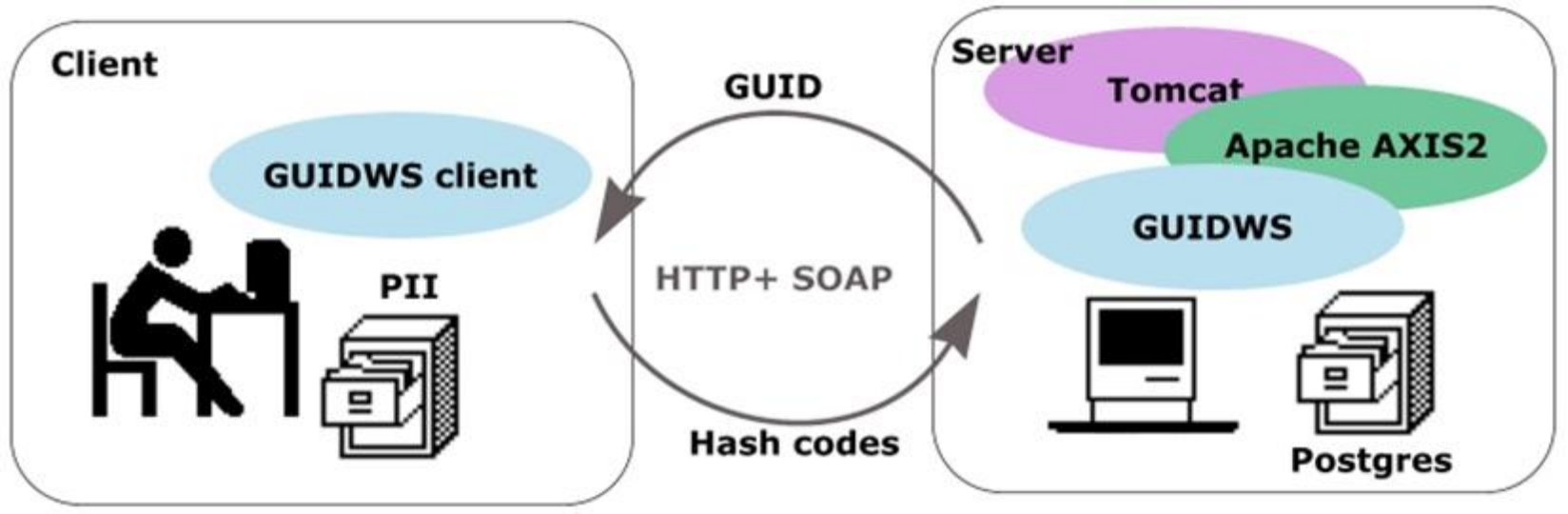
Global Unique Identifier – GUID



- subject PII will never be collected into GRDR

http://jamia.bmj.com/content

- Facilitate follow up of patient data over time, across studies, registries, clinical trials and across countries, while protecting patient privacy and linking patient clinical data to biospecimens data
- Currently, about 1000 patients participating in the GRDR were assigned the GUID

GRDR-GUID

Requesting an account and creating GUIDs

Account Management

Login

Please provide your username and password to access this content.

Username *

Password *

LOG IN

Need Assistance?

Request A New Account | Forgot your username? | Forgot your password?

We recommend using one of the following supported browsers and versions for accessing this system: Internet Explorer 9+, Firefox 9+, Chrome

Workspace | GUID | Account Management

GUID (Global Unique Identifier)

GUID Tool

- GUID Overview
- Create GUIDs
- My GUIDs

GUID Admin

Create GUIDs

In order to protect the privacy of study participants, the GUID tool is run locally. By selecting the link below, the GUID tool will be downloaded and executed automatically from your computer. The GUID tool requires a version of Java to be installed on your computer.

- Launch GUID Tool

Helpful Documentation

- Getting started and need help? Download the GUID User Guide (pdf)
- Need to generate multiple GUIDs at one time? Download the GUID Batch Template (xls)

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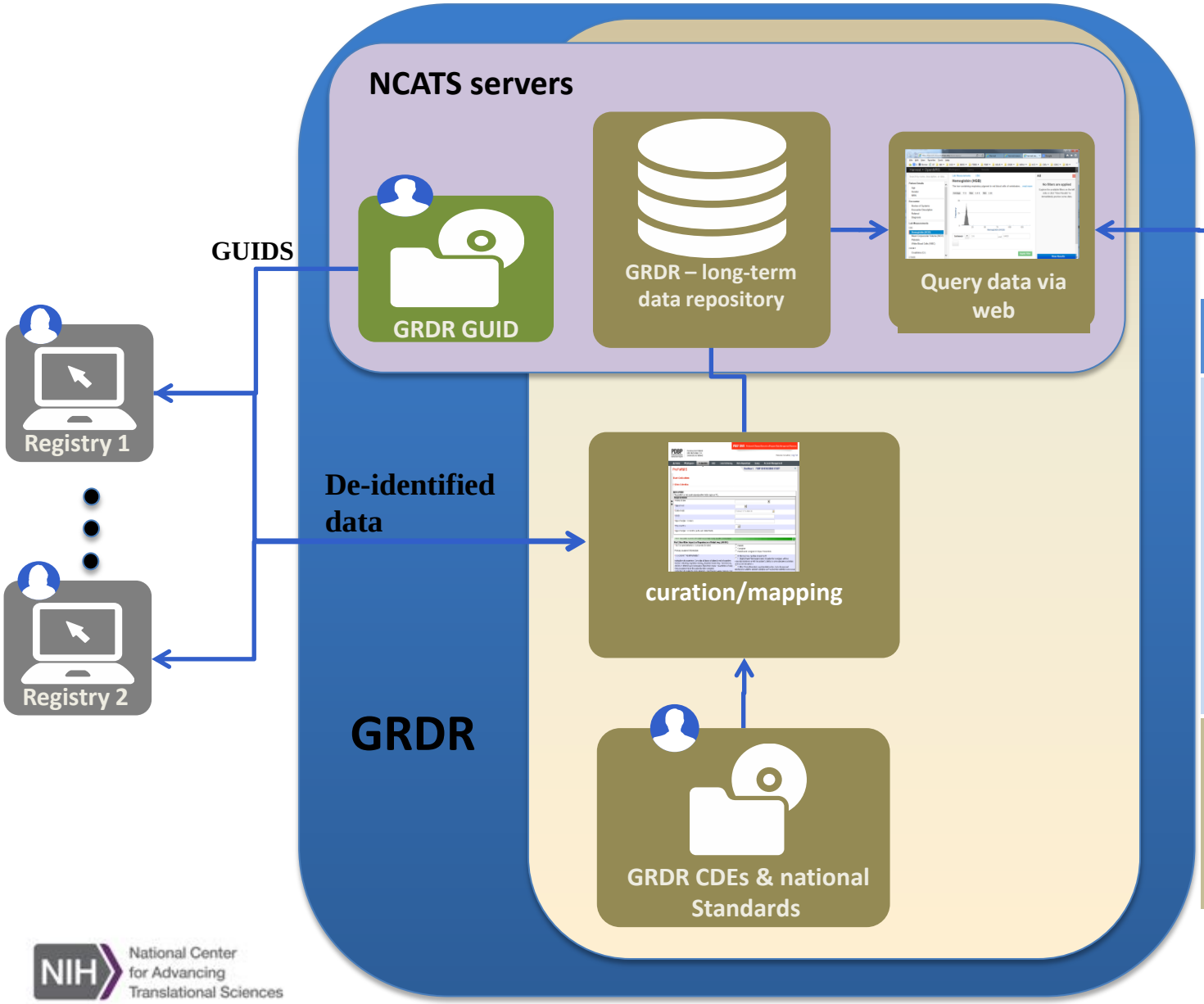
GRDR Data Repository



Query of data will be available in I two ways.

A. Data query through the GRDR repository in two levels.
1. For population level – open access (statistics and aggregated data)
2. For observational research – detailed data – controlled access.

B. Query for Clinical Trails through the home registry. Recruitments of patients will be available only through the registry that provided the data.

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Participating Groups (Since May 2014)

- Intracranial Hypertension Research Foundation
- Pachyonychia Congenita Project
- Rare Cancer Genetics Registry
- The North American Malignant Hyperthermia Registry of the Malignant Hyperthermia Association of the United States
- Bardet-Biedl Syndrome Family Association
- National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Condition
- Cornelia de Lange syndrome

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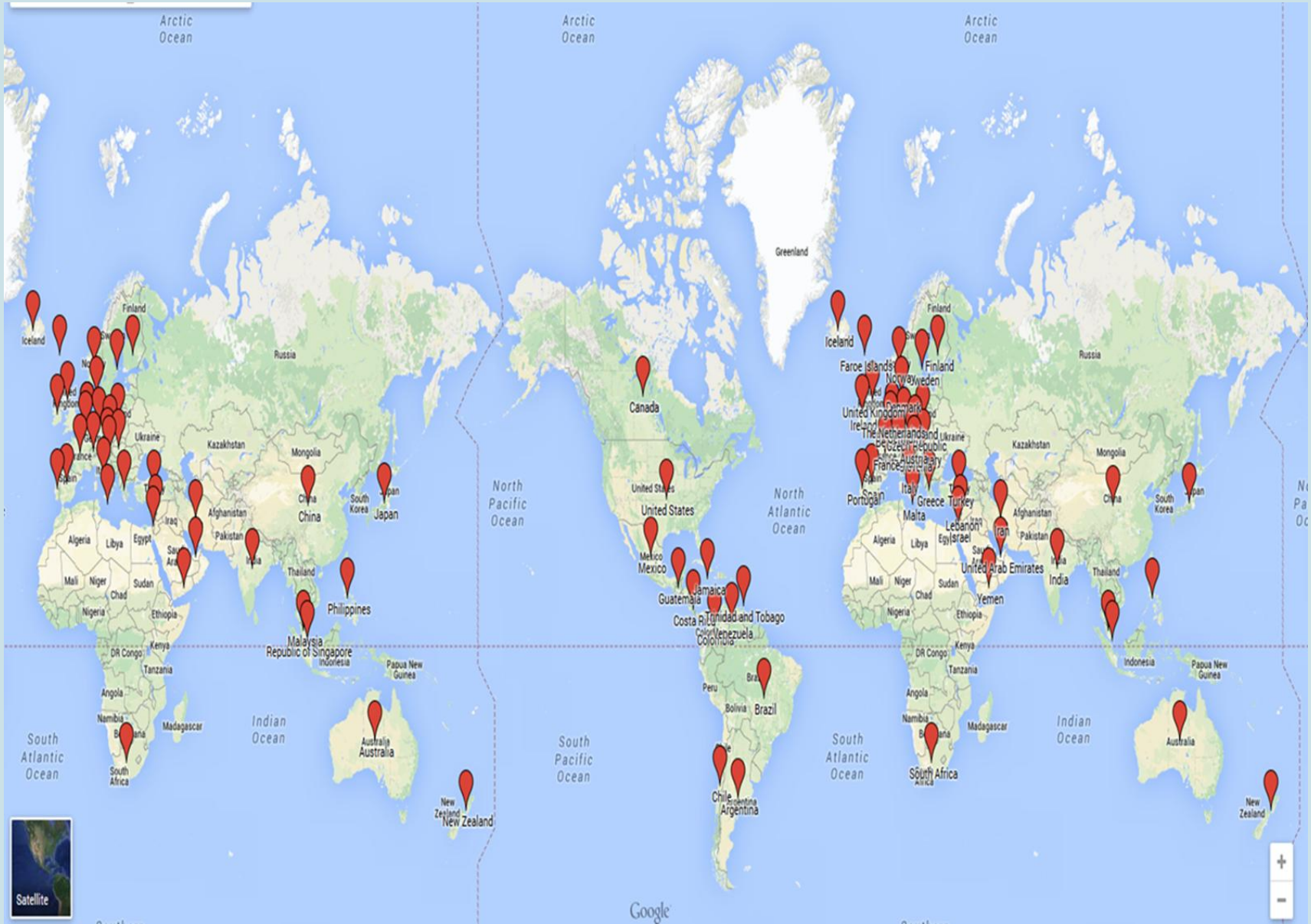
Current Distribution of GRDR Patients: 75 diseases, 51 countries and 7600 patients



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Resources Developed/Provided through the NIH/NCATS GRDR^{SM} Program

- Common Data Elements (CDEs)
- Template Informed Consent
- Template Patient registry
- Central IRB services
- Access to Global Unique Identifier (GUID)
- Mapping patients' data to GRDR CDEs and national standards
- Ability to link patient data to their biospecimens through the database for Biospecimens/Biorepositories (RD-HUB)
- Policies for submitting and accessing data
- Website with information for rare disease community and investigators with a link to other resources

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NIH/NCATS GRDR^{SM} Program Value

- **For patients and their families: Increase awareness of their specific rare disease and facilitating accelerated therapeutic development**

- **For rare disease organizations: Providing resources and registry tools. Map data from each registry to standards facilitating interoperability among them and between other databases**

- **For investigators and industry: Facilitate research collaboration and cross-disease analyses by lowering barriers to data access**

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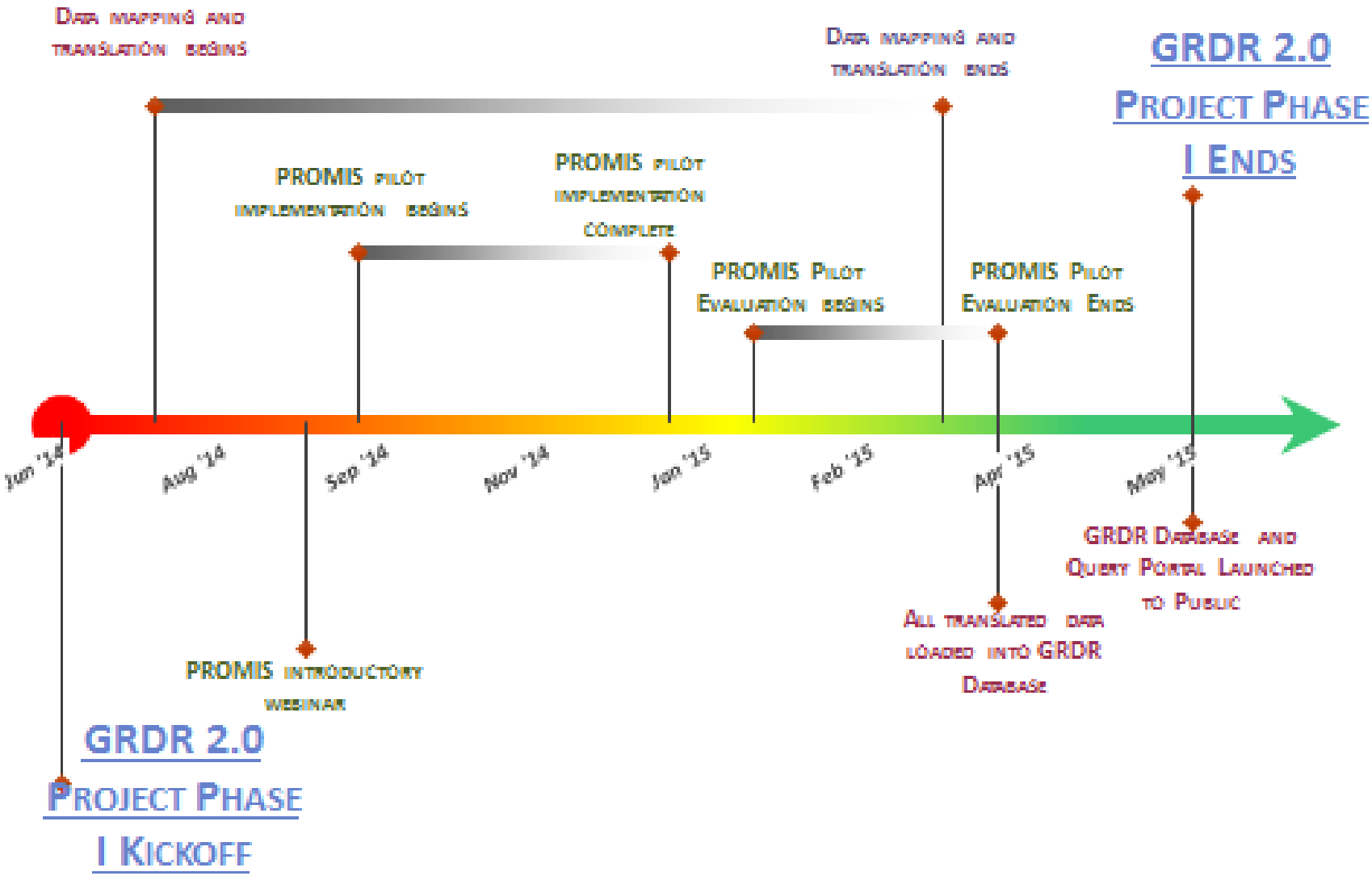
GRDR 2.0 Phase I Timeline



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RD-HUB
Rare Diseases Human Biospecimens-Biorepositories Database/Website
http://biospecimens.ordr.info.nih.gov/

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RD-HUB

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Search

RARE DISEASES HUMAN BIOSPECIMENS/BIOREPOSITORIES (RD-HUB)

- Home
- Enter Biorepository Data
- Search the Database
- Biorepositories
- Collaborations
- Other Related Resources
- News and Publications
- Events
- Frequently Asked Questions
- User Manual

Search the Human Biospecimens/Biorepositories Database

Select items from each tab below to refine your search. For detailed instructions about searching the database, please refer to the User Manual-Database Search.

Please note: Rare diseases are labeled with an asterisk (*) and are linked to disease-specific information Web pages created by the Genetic and Rare Diseases Information Center (GARD). Non-contributing biorepositories and biospecimens not yet available are grayed out or unselectable.

Quick Search: GO

Disease Name | Anatomic Source | OMIM Number | Repository | GUID

Advanced Search:

Disease | Specimen Type | Anatomic Source | Processing Method | Storage Method | Imaging | Repository

SEARCH | CLEAR ALL

All Diseases | Normal

#ABCDEFGHIJKLMNOPQRSTUVWXYZ

Diseases with specimens

- Search All
- 18 trisomy
- 7-Dehydrocholesterol reductase deficiency

Your Search Selection

Disease- Search All

Normal- Search All

Specimen Type- Search All

Anatomic Source- Search All

Processing Method- Search All

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Future Plans

Acknowledgements

NCATS team

Pamela McInnes (NCATS Deputy Director)

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- Matt McAuliffe
- Barbara Karp

CHOP team (DMCC)

Chris Forrest, Lead DMCC

- Charlie Bailey
- Ryan Vass
- Mark Padola

GRDR Steering members and the GRDR Leadership Committee

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