The NIH/NCATS GRDR℠ 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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The NIH/NCATS GRDR℠ Program
Global Rare Diseases Patient Registry
Data Repository

GRDR℠ - a data repository by the patients for the patients, to improve the quality of life of millions suffering from rare diseases

Users requiring approval
No Pending Actions

The NIH/NCATS GRDR℠ Program
The GRDR program aims to build a rapid-learning health program, incorporating data from electronic health records (EHRs), and to develop large global data sets of patients with rare diseases. It will accelerate development and uses of new knowledge to improve the health and quality of life for millions of persons.

Rare Diseases have no borders!
They don’t affect individuals, they affect entire families.

- Background
- History
- Lessons learned
- Current state
- GRDR scientific & clinical value
- Future plans
NIH/NCATS GRDRSM Program

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

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

De-identified patient data is shared with GRDRSM 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

Researchers conduct various biomedical studies within & across diseases

Researchers, Clinicians, Industry, Pharma

Other RD Databases

Linking to other databases
GRDR\textsuperscript{SM} Data Repository
https://grdr.ncats.nih.gov/

- Template Patient Informed Consent for participating in Patient Registries
- Global Unique Identifier-GRDR-GUID

- 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
Integrating Electronic Health Record

Common Data Elements

- Identifiers
- Socio-Demographics
- Rare Disease Diagnosis
- Family History
- Birth & Reproductive History
- Medications & Dietary Supplements
- Utilization

Ascertain whether a hybrid between the EHR and the organization's registry can be used to populate the GRDR repository.
NIH/NCATS GRDR℠ Program
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
From a pilot project

Lessons learned

to a GRDRSM 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
GRDR Program Collaboration

- Through its GRDR\textsuperscript{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 NIH/NCATS/GRDR\textsuperscript{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.
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.
GRDRSM Program Governance
Organizational Chart

NIH/NCATS GRDRSM Program Leadership

GRDR DMCC

GRDR IRB

GRDR SC

GRDR Support
GRDR℠ 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.
GRDR\textsuperscript{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.)
Mapping Process

- Registry provides data dictionary
- DMCC team works with each registry on mapping their 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
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)
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

GUID (Global Unique Identifier)

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)
The GRDR Data Repository supports data query through two levels:

1. For population level – open access (statistics and aggregated data)
2. For observational research – detailed data – controlled access.

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 Trials through the home registry. Recruitments of patients will be available only through the registry that provided the data.
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
Current Distribution of GRDR Patients:
75 diseases, 51 countries and 7600 patients
Resources Developed/Provided through the NIH/NCATS GRDR℠ 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
NIH/NCATS GRDR℠ 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
RD-HUB
Rare Diseases Human Biospecimens-Biorepositories Database/Website
http://biospecimens.orcdr.info.nih.gov/
Future Plans
Acknowledgements

NCATS team
Pamela McInnes (NCATS Deputy Director)
Yaffa Rubinstein (GRDR Program Director)

- Charlie Jones
- Josephine Kennedy
- Mark Backus
- Erica Helzner
- 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