Lab Data (LAB and PGx)

- Study Data

Case Report Forms (CRF) (CDASH)
- Study Data

Protocol
- Study Design
- Eligibility
- Registration
- Schedule

Analysis Datasets (ADaM)

Tabulated CRF data (SDTM)
- Study Data
- Lab Data
- Study Design

eSubmissions (eCTD+data)
Analysis and Reporting

Adapted from: CDISC, 2011.
Patient Registries and Drug Development

Adapted from: Chris Austin, Director of NIH Chemical Genomics Center, National Human Genome Research Institute, NIH during the Research Daylong Symposium "GRADRx: Crafting a Plan for Rare Disease Drug Development" at the 2009 Genetic Alliance Annual Conference.

Identify populations for trials; understand their characteristics; develop measures for later trials

Identify trial participants; understand changing patient populations

Natural History Studies

Registries: Post-market safety; “comparative effectiveness research” in “real-world”
I AM 1 IN 10

One of NORD’s consistent messages is that rare diseases aren’t really rare. When considered together—and they should be, because there are significant challenges that all people with rare diseases have in common—they affect nearly 30 million Americans or almost 1 in 10 of us. NORD’s logo illustrates the 1 in 10 theme. So does our current annual report, which features photos of several people who have rare diseases. To read the stories behind those photos, go to page 4.

NORD TO HIGHLIGHT MEDICAL FOODS ISSUE

On February 10, 2011, NORD will host a conference in Washington, DC, to focus attention on the fact that many families are struggling to provide the medical foods and special formulas needed by children and adults with certain metabolic diseases. In many cases, without these special foods,
EURORDIS releases Q&A to help patients advocate for their right to cross border healthcare
SEARCH BY DISEASE/GENE

fabry

(*) mandatory field

Diseases & biobanks categories

Patient registries/Databases

Country

All countries

→ OK

:: 14 Result(s)
## Who is Developing Registries?

<table>
<thead>
<tr>
<th>Rare Disease Clinical Research Network</th>
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</thead>
<tbody>
<tr>
<td>Patient Advocacy Group Members</td>
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</table>

- Angelman, Rett, and Prader-Willi Syndromes Consortium
- Autonomic Rare Diseases Clinical Research Consortium
- Brain Vascular Malformation Consortium
- Consortium for Clinical Investigation of Neurological Channelopathies
- Spinocerebellar Ataxia - Clinical Research Consortium
- Chronic Graft Versus Host Disease Consortium (cGVHD)
- Dystonia Coalition
- Genetic Diseases of Mucociliary Clearance Consortium
- Inherited Neuropathies Consortium
- Lysosomal Disease Network
- NEPTUNE: Nephrotic Syndrome Rare Disease Clinical Research Network
- North American Mitochondrial Diseases Consortium
- Porphyrias Consortium
- Primary Immune Deficiency Treatment Consortium (PIDTC)
- Rare Kidney Stone Consortium
- Salivary Gland Carcinoma Consortium
- STAIR:Sterol and Isoprenoid Diseases Consortium
- Urea Cycle Disorders Consortium
- Vasculitis Clinical Research Consortium
Pharmaceutical Companies

Health Care Professionals
The Fabry Registry is an ongoing, observational database that tracks natural history and outcomes of patients with Fabry disease. Evaluate trends. Explore issues. Share observations. The Fabry Registry is your resource:

- Understand
- Participate
- Evaluate

Patients & Families
Are you looking for a way to contribute first-hand to a larger goal that may help you and other people living with Fabry disease? Ask your doctor about participating in the Fabry Registry:

- Learn More
- Go to Fabry Community

Physician Online Enrollment
Participation in the Fabry Registry is open to all physicians managing patients with Fabry disease. Physician enrollment in the Registry is simple. Complete the Physician Enrollment Form by clicking on the link below.

- Online Physician Enrollment

Related Links & Downloads
- Registry Materials
- Fabry Protocol (PDF)
and...
Who is developing registries?
Registry World

- Wild-west
  - Diverse diseases
  - Diverse sponsors
  - Diverse objectives
  - No recognized authority or regulator
Metadata: Dublin Core

Osteogenesis Imperfecta

Medical History Questions

Do other members of your family have Osteogenesis Imperfecta? Please include family members who are no longer living [+] / [-]

[Properties: Family Medical History, OI.0001, Brittle bone syndrome, Brittle bone disease, Fragilitas ossium, OI-Osteogenesis imperfecta, Osteogenesis imperfecta, Osteopatryhosis]

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</table>

○ No
○ Yes
“Do you have problems with your eyes?”
“Burning sensation on hands and feet?”
“History of major diseases: if yes, list below”
“…need for TV to fall asleep?”
“Spells of night screaming..”
“Fear/anxiety while sleeping…”
“Approximately how many fractures have you had in your lifetime?”
“Age at time of first fracture?”
“Have you had sinus surgery?”
## Neonatal

### Did your child have respiratory distress at birth? [+] / [-]

- **coverage**: yes/no indicator to ask if a patient has a medical history for respiratory distress at birth

  - **Options**:
    - Yes
    - No
    - Unknown

### Was your child full-term? [+] / [-]

- **coverage**: yes/no indicator to ask whether a child was carried to a full-term pregnancy, lasting approximately 40 weeks beyond the date of the mother's last menstrual period

  - **Options**:
    - Yes
    - No
    - Unknown

### How many days did your baby spend in the hospital after birth? [+] / [-]

- **coverage**: Numeric value to express the number of days the patient spent in hospital after delivery

### Did your baby have oxygen therapy while in the hospital after birth? [+] / [-]

- **coverage**: indicator of use of oxygen therapy on baby in the hospital after delivery (Inhalation of oxygen aimed at restoring toward normal any pathophysiologic alterations of gas exchange in the cardiopulmonary system, as by the use of a respirator, nasal catheter, tent, chamber, or mask. (From Dorland, 27th ed & Stedman, 25th ed)

  - **Options**:
    - Yes

### How many days did your baby spend on oxygen? [+] / [-]

- **coverage**: Numeric value to indicate the number of days patient spent on oxygen in the hospital after delivery (Inhalation of oxygen aimed at restoring toward normal any pathophysiologic alterations of gas exchange in the cardiopulmonary system, as by the use of a respirator, nasal catheter, tent, chamber, or mask. (From Dorland, 27th ed & Stedman, 25th ed)

  - **Options**:
    - No

### Did your child have meconium aspiration (a condition caused by inhalation of amniotic fluid and first passage of feces)? [+] / [-]

- **coverage**: yes/no indicator of diagnosis of condition caused by inhalation of meconium into the lung of a fetus or newborn (Inhalation of oxygen aimed at restoring toward normal any pathophysiologic alterations of gas exchange in the cardiopulmonary system, as by the use of a respirator, nasal catheter, tent, chamber, or mask. (From Dorland, 27th ed & Stedman, 25th ed)

  - **Options**:
    - Yes
Questions

PRISM

Registry Sponsors (Patient Advocacy Groups)

Various data standards & SDOs
Genomic research

Consent 2.0

A better way of signing up for studies of your genes

Apr 28th 2012 | NEW YORK | from the print edition
That’s My Data!

THE SATURDAY ESSAY  |  December 3, 2011

Citizen Scientists

Ordinary people are taking control of their health data, making their DNA public and running their own experiments. Their big question: Why should science be limited to professionals?