Research Ready: Conducting Research Through Patient Registries

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Overview

• The importance of research
• Natural history studies
• Patient Registries
• Challenges in the rare community
• How to implement and manage a registry
• Models of Engagement
...a way to tell a story. It is the process of discovery.
Natural History Studies

• **Inform** patient care and best practices
• **Assess** patient and caregiver experiences and preferences
• **Contribute** to disease understanding
• **Identify** research priorities such as genetic, molecular and physical basis of rare diseases
• **Estimate** the number of affected patients and patients potentially available to participate in research
• **Evaluate** the individual and global economic burden of disease
• **Inform** drug development
• **Provide** an avenue for bio-specimen collection
Challenges for Rare Patients and Caregivers

- Average of 5-7 years for diagnosis
- 90% without an FDA approved treatment or therapy
- Extensive, life-long medical needs
- High cost of care and treatment
- Few medical experts
- Little research or known about diseases
- Social Isolation
- Small, scattered patient populations
Collection of information about individuals

- Specific disease or diagnosis
- General patient demographics
- Genetic Information
- Patient-reported outcomes
- Comprehensive medical data
- Clinical trial matching

Main types of registries

- Contact registries
- Patient registries
- Surveillance registries
Engage with Stakeholders

- PATIENTS & FAMILIES
- PATIENT ORGANIZATIONS
- MEDICAL PROFESSIONALS
- PAYORS
- LEGISLATORS
- INDUSTRY
- REGULATORS
BUILDING A REGISTRY AND NHS
Registry Implementation Workflow

**Do your homework**
- Gauge community interest
- Decide on the type of data that will be collected
- Decide on the type of registry
- Who will contribute data?
- Assess resources

**Plan**
- Choose a platform
- Choose an IRB
- Build an advisory committee
- Design study
- Develop a multi-year plan

**Develop**
- Study documents
- Surveys
- Test
- Build registry
- Launch!
Features and Functionalities in a Registry Platform

• Design and Usability – configurable, easy to navigate
• Governance – IRB, privacy, auditing
• Data - sharing, access and ownership
• Customer Support
• Security
MODELS OF ENGAGEMENT & USE CASES

- No patient organization
- Patient organizations as research partners
- Academic or industry researcher
- Bringing research partners together through central infrastructure
Using the Data

- Publications
  - Medical journal articles
  - Peer-reviewed papers
- Posters
- Community Reports
- Research collaborations

- Patient/family conferences
- Medical conferences
- Webinars
- Patient-Focused Drug Development meetings
Partnership Models

IAMRARE™ Platform
Building Collaboration Through Shared Infrastructure

REGISTRY PLATFORM

MAIN STUDY

SUB-STUDY A

SUB-STUDY B

SUB-STUDY C

SUB-STUDY

MAIN STUDY

SUB-STUDY

SUB-STUDY

rarediseases.org
The Power of the Data

**Patient Organizations**
- Research partners and leaders
- New models of engagement
  - New members/collaborations
  - Expand/strengthen existing partnerships
  - Unite divided communities

**Industry**
- Collaborating with patient groups and supporting meaningful patient centered product development
- Models of partnerships
- Pair registry data with clinical trial data
- Clinical trial recruitment

**Patients, Families & Caregivers**
- Access to data
- Insights into patient outcomes
- Symptom tracking
- Resources for conversations with clinicians, educators

**Clinicians**
- Data as a reference point for patient reported outcomes (PRO)
- Access to patient cohorts
- Registry data to supplement existing clinical trial data
- Document conditions (no data, progression, transitions in care)
- Inform new and revised standards of care
### The Power of the Data

<table>
<thead>
<tr>
<th><strong>Researchers</strong></th>
<th><strong>Regulatory Agencies</strong></th>
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<tr>
<td>• Compare and validate experimental findings with PRO data</td>
<td>• Registry data to accelerate and improve research</td>
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<td>• Use trends from preliminary data to explore new areas of a disease</td>
<td>• Outcomes from clinical trials</td>
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| • Provide real world evidence when applying for research grants | • Transitions in care  
  • Pediatric to adult  
  • Treatment protocols |
| • Inform product development | • Barriers to adherence and or compliance to treatment |
| • Regulatory approvals | • Contextualize clinical trial data |
| • Validate measures | • New models of engagement  
  • New members/collaborations  
  • Expand/strengthen existing partnerships  
  • Unite divided communities |
| • Establish new research protocols | |
The Power of Patients amplifies the experiences of individuals and families living with rare diseases and demonstrates the importance of registries and natural history data in helping to shape the rare disease landscape.


• NCATS Toolkit for Patient-Focused Therapy Development

• FDA Guidance
PKU Patient Registry

Lauren Youngborg, MS, CGC
PKU Patient Registry Director
National PKU Alliance
PKU Patient Registry: Development

NPKUA’S MISSION

Improve the lives of people impacted by PKU and pursue a cure

Inform & Accelerate Research

- Well-organized registries can attract the attention of the research community by reducing or eliminating common barriers.

Integrate the Patient Voice in Research

- Patient-reported data helps the research community better understand the patient journey and the issues that are important to PKU patients and families.

Facilitate Development of Guidelines

- Registries can improve the lives of patients and families by informing the development of best practices and management guidelines to optimize medical care.
PKU Patient Registry: Platform

Registry Platform Considerations

• Cost
• Data Ownership
• Input from FDA, NIH and Other Experts
• Community of Clients
• Participant Engagement
• Program Growth
PKU Patient Registry: Data

Current Enrollment
• 1,141 participants – 955 consented
• 7,000+ surveys submitted

Registry Demographics
• 59% female - 41% male
• 49/50 US states and 28 countries represented
• 52% < 18 years, 48% ≥ 18 years
  • Age range 10 months – 67 years
PKU Patient Registry: Stakeholders

Key Registry Stakeholders

- Patients and Families
- Healthcare Providers
- Research Community
- Regulatory Agencies
- Industry
**PKU Patient Registry: Impact**

**Patient Recruitment**
- Identification of patients who may be interested and eligible for participation in a specific study through targeted Registry communications

**Data Sharing**
- Data sharing collaborations with industry and academic researchers
- Rare Disease Cures Accelerator – Data and Analytics Platform

**#PKUinThisTogether**
- PKU community is united around a common goal
- A concrete and easy way that patients and families can partner with other stakeholders from the PKU community to make an impact