



NORD®
National Organization
for Rare Disorders

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



Research is...



...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



[The importance of NHS](#)



Challenges for Rare Patients and Caregivers



Average of 5-7 years
for diagnosis



Few medical experts



90% without an FDA
approved treatment
or therapy



Little research or
known about diseases



Extensive, life-long
medical needs



Social Isolation



High cost of care and
treatment



Small, scattered
patient populations



Patient Registries

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

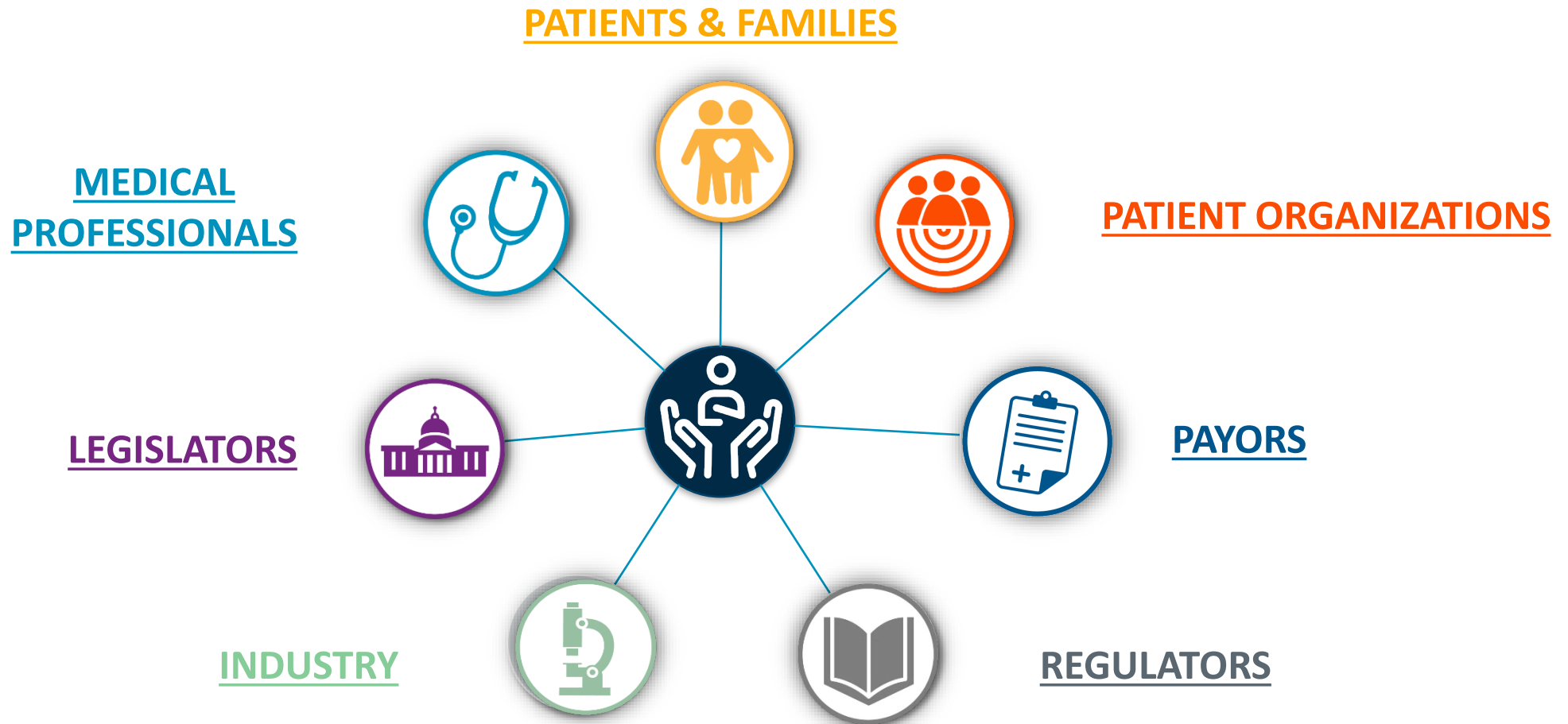
- Contact registries
- Patient registries
- Surveillance registries



[What is a Registry?](#)



Engage with Stakeholders



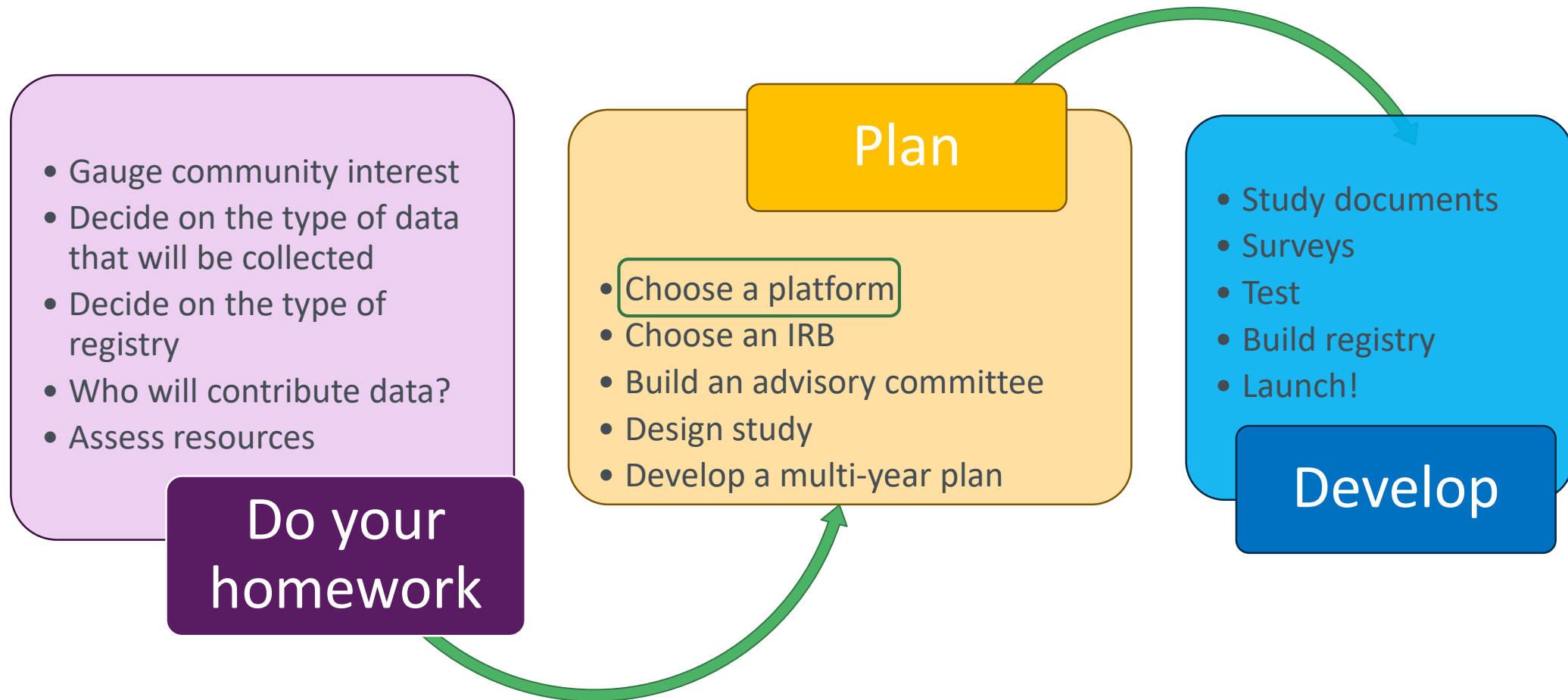


BUILDING A REGISTRY AND NHS





Registry Implementation Workflow





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

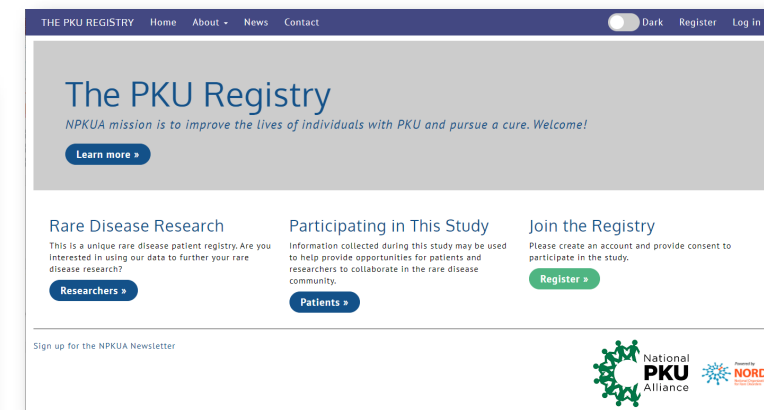
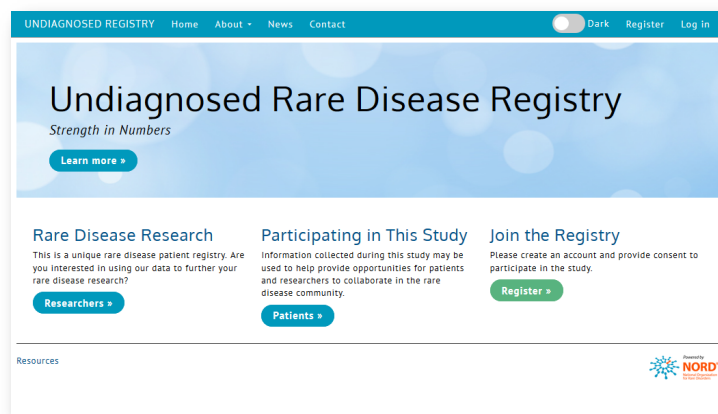
- Publications
 - Medical journal articles
 - Peer-reviewed papers
- Posters
- Community Reports
- Research collaborations
- Patient/family conferences
- Medical conferences
- Webinars
- Patient-Focused Drug Development meetings





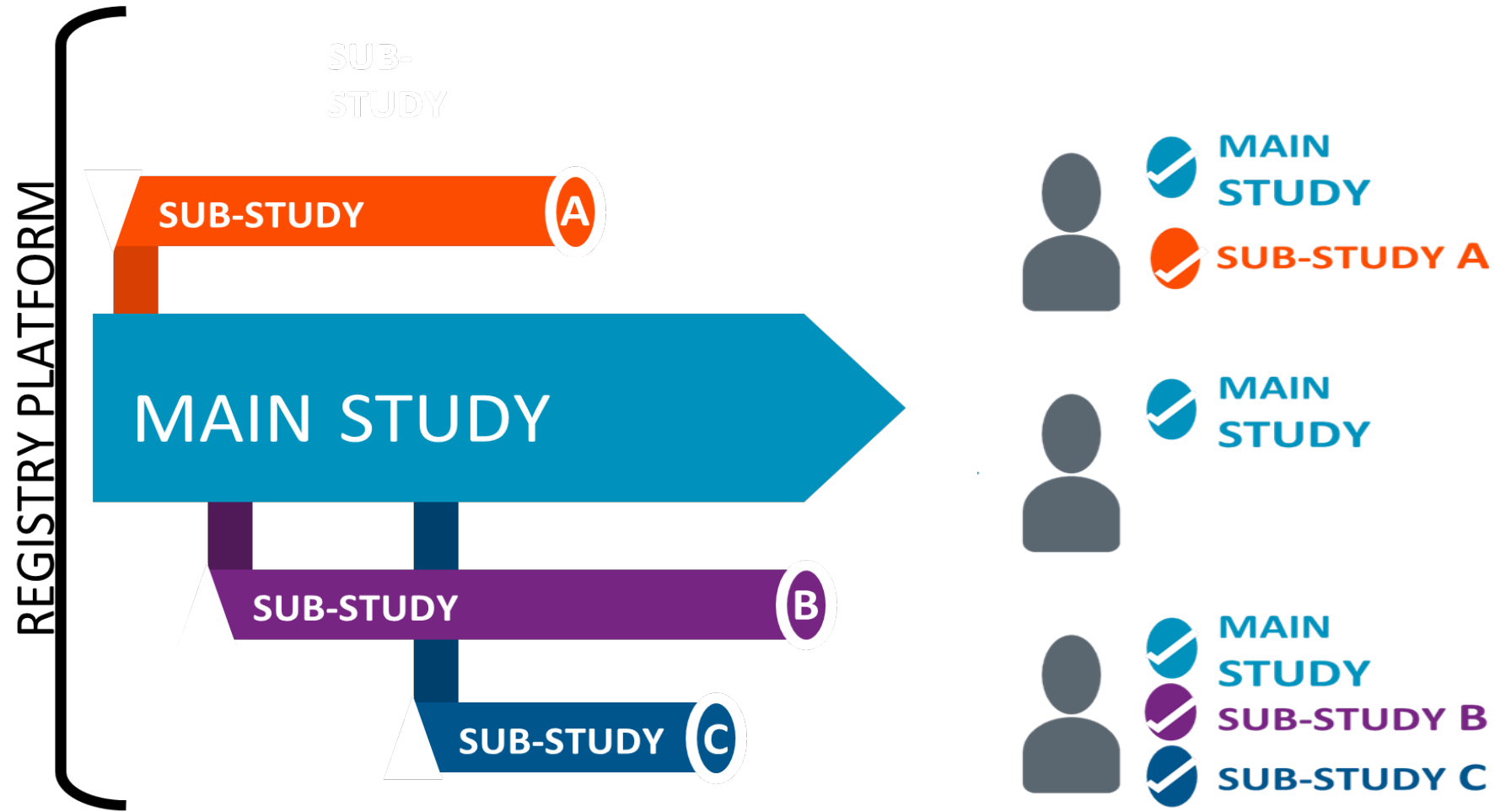
Partnership Models

IAMRARE™ Platform





Building Collaboration Through Shared Infrastructure





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



Researchers

- Compare and validate experimental findings with PRO data
- Use trends from preliminary data to explore new areas of a disease
- Provide real world evidence when applying for research grants
- Inform product development
- Regulatory approvals
- Validate measures
- Establish new research protocols

Regulatory Agencies

- Registry data to accelerate and improve research
- Outcomes from clinical trials
- Transitions in care
 - Pediatric to adult
 - Treatment protocols
- Barriers to adherence and or compliance to treatment
- Contextualize clinical trial data
- New models of engagement
 - New members/collaborations
 - Expand/strengthen existing partnerships
 - Unite divided communities



The Power of Patients

THE POWER OF PATIENTS

*Informing Our Understanding
of Rare Diseases*



Trio Health

***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.*





Resources

- Registries for Evaluating Patient Outcomes: A User's Guide.
[AHRQ 4th ed.](#)
- Registries for Evaluating Patient Outcomes: A User's Guide.
[AHRQ 3rd ed.](#)
- [NCATS Toolkit for Patient-Focused Therapy Development](#)
- [FDA Guidance](#)



NORD®
National Organization
for Rare Disorders



National
PKU
Alliance

PKU Patient Registry

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



PKU Patient Registry: Development

NPKUA'S MISSION

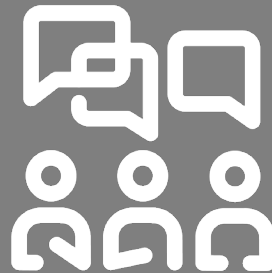
Improve the lives of people impacted by PKU and pursue a cure

Inform & Accelerate Research



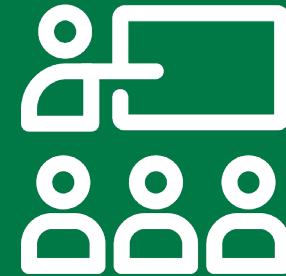
- Well-organized registries can attract the attention of the research community by reducing or eliminating common barriers.

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

