Up Next: Case Study 2: Integration and application of phenylketonuria patient registry data in RDCA-DAP

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NORD
Case Study 2: 
Integration and application of phenylketonuria registry data in RDCA-DAP 

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Overview

• Brief introduction to NORD
• The value of patient-reported data in rare disease drug development
• Case study on the process and examples of data-driven insights
• How RDCA-DAP supports rare disease drug development
NORD, an independent nonprofit, is leading the fight to improve the lives of rare disease patients and families.

We do this by supporting patients and organizations, accelerating research, providing education, disseminating information and driving public policy.

rarediseases.org
IAMRARE™: History and Growth

- **2014**: First Registry launches on NORD platform
- **2015**: 10,000 survey submissions
- **2016**: Launch of first registries supported by NORD-FDA cooperative agreement
- **2017**: NORD-FDA Cooperative agreement is announced
- **2018**: Registry community meetings
- **2019**: NORD-FDA awardee selections
- **2020**: RDCA-DAP launch meeting, 4 federally funded projects, new models of engagement, CZI Rare Launch Grant

**5th Anniversary of Program**

**Undiagnosed Rare Disease Natural History Registry**
IAMRARE™: Foundational goals

- Empower and support patient advocacy organizations, patients and caregivers to be equal partners and participants

- Drive and support the pursuit of new, collaborative research models that challenge the status quo

- Catalyze and accelerate the development of treatments and cures
IAMRARE™: Foundational goals

- Empower and support patient advocacy organizations, patients and caregivers to be equal partners and participants
- Drive and support the pursuit of new, collaborative research models that challenge the status quo
- Catalyze and accelerate the development of treatments and cures
Data is key!

What is a Registry?

https://rarediseases.org/iamrare-registry-program/

RARE DISEASE PATIENT REGISTRIES UNLOCK CURES

Joining a registry opens the door for more and better research.

Adding your data can make clinical trials shorter and less expensive, delivering therapies to the community faster.

Your unique experience adds to what we know about a disease. The more patients share their experiences, the better we understand the full spectrum of the disease.

Researchers, health care providers and the FDA need help to understand different diseases. Your data is key for these decision makers!

Learn more about how you can contribute data at: rarediseases.org/RDCA-DAP

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https://rarediseases.org/rdca-dap/resources/
How do larger, standardized datasets help my disease state?

Bigger datasets, especially in rare diseases, increase our collective power.

That power can translate to greater, more efficient drug development.

Data belongs to patients – and they want it used as much as possible to help pave the way towards treatments!
RDCA-DAP and Phenylketonuria (PKU) Case Study
About: Phenylketonuria (known as PKU) is an inherited metabolic disease affecting the brain through increased levels of a substance called phenylalanine (Phe) in the blood. PKU infants in the United States are diagnosed in the first few days of life through the federally mandated Newborn Screening Program.

Organization: The National PKU Alliance, formed in 2008, with the goal of improving the lives of families and individuals associated with PKU through research, support, education and advocacy, while ultimately seeking a cure.

Registry:
- January 2017
- Open ended longitudinal study
- Open enrollment
- Currently ~1200 participants
Although newborn screening for phenylketonuria (PKU) identifies affected individuals pre-symptomatically allowing for timely initiation of therapeutic intervention, little is known about long-term health outcomes and natural history of PKU which informs clinical trial readiness for emerging therapies including cell and gene-based therapeutics.
Case summary - PKU Diets: Patient Experiences

- Most participants reported actively being on a PKU diet
- Some participants reported that they have used PKU diets before but have stopped at some point
- For patients who stopped, the most common reason to return to the diet was due to a pregnancy
- When returning to a diet, the options were frequently a Low Protein diet

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<th>Have you ever returned to a PKU Diet?</th>
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How RDCA-DAP impacts rare disease drug development

- **360° view** of disease characterization and natural history
- **Accelerate understanding** of conditions and commercial/research interest; inform the design of trials
- **Encourage greater representativeness** in study samples - steps toward more equitable and inclusive study designs
- **Opportunity** for cross-disease discovery
- **Efficient, effective** use of resources
Thank you to the National PKU Alliance for being a long time IAMRARE partner and an early data contributor to RDCA-DAP!

To learn more about NORD’s research portfolio and IAMRARE program visit: https://rarediseases.org/iamrare-registry-program/

Contact: research@rarediseases.org
THANK YOU!

Don't forget to answer survey questions.

For more information, email rdcadap@c-path.org