CRITICAL PATH INSTITUTE

NORD®
National Organization for Rare Disorders

Rare Disease Cures Accelerator-Data and Analytics Platform
Virtual Workshop 2020

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

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National Organization for Rare Disorders

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

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Overview

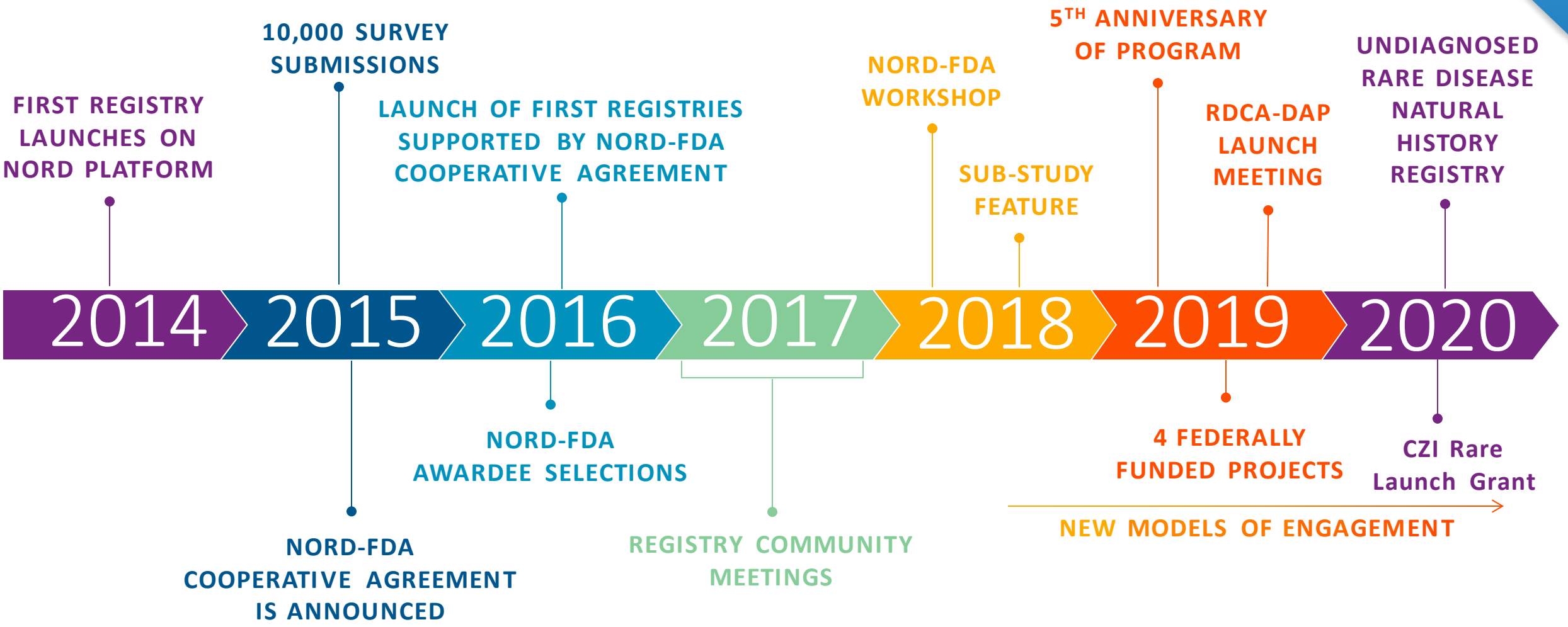
- Brief introduction to NORD
- The value of patient-reported data in rare disease drug development
- Case study on the process and examples of data-driven insights
- How RDCA-DAP supports rare disease drug development

National Organization for Rare Disorders (NORD)

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.

IAMRARE™: History and Growth



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

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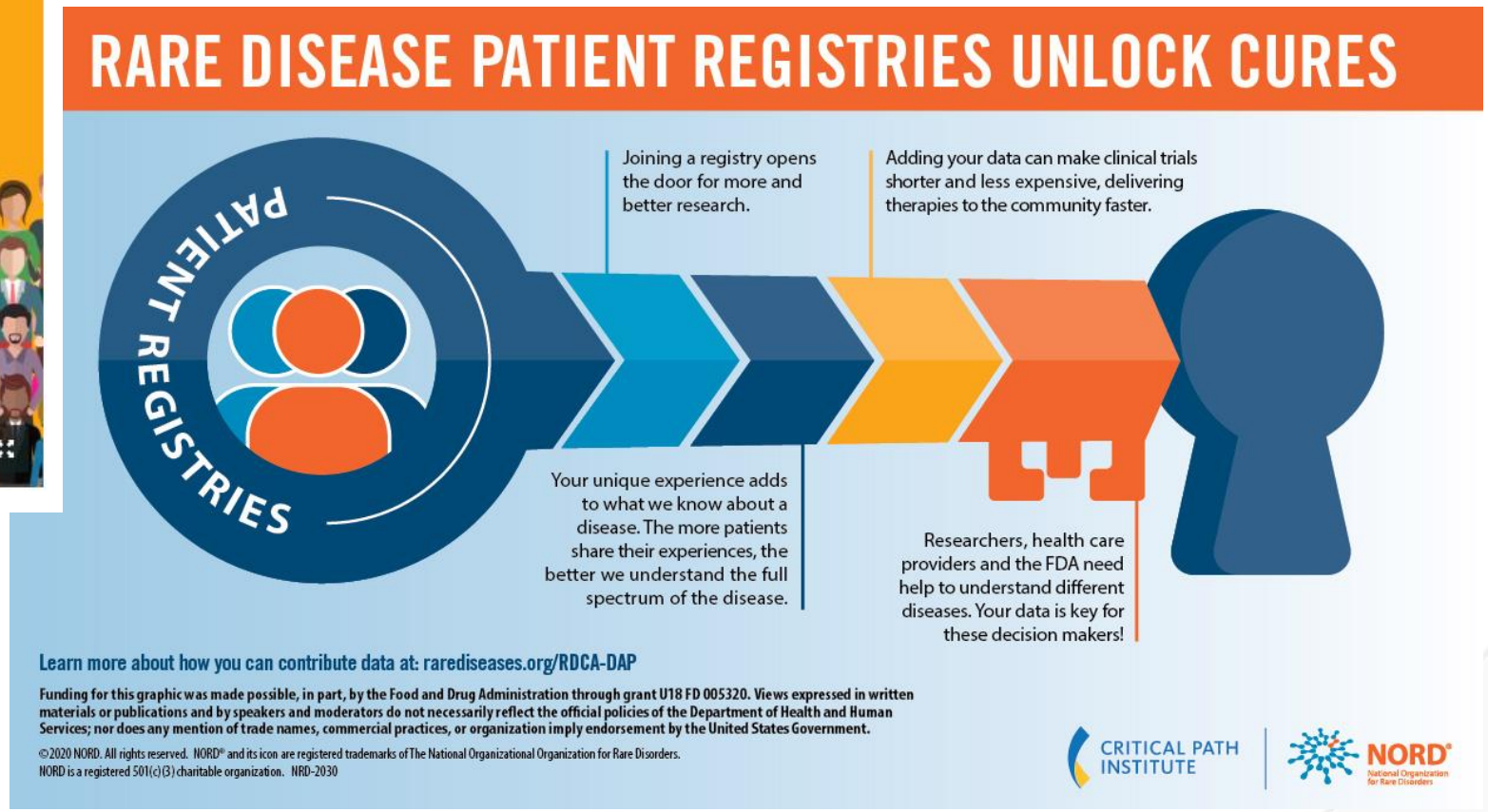
Data is key!

What is a Registry?



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

https://rarediseases.org/rdca-dap/resources/

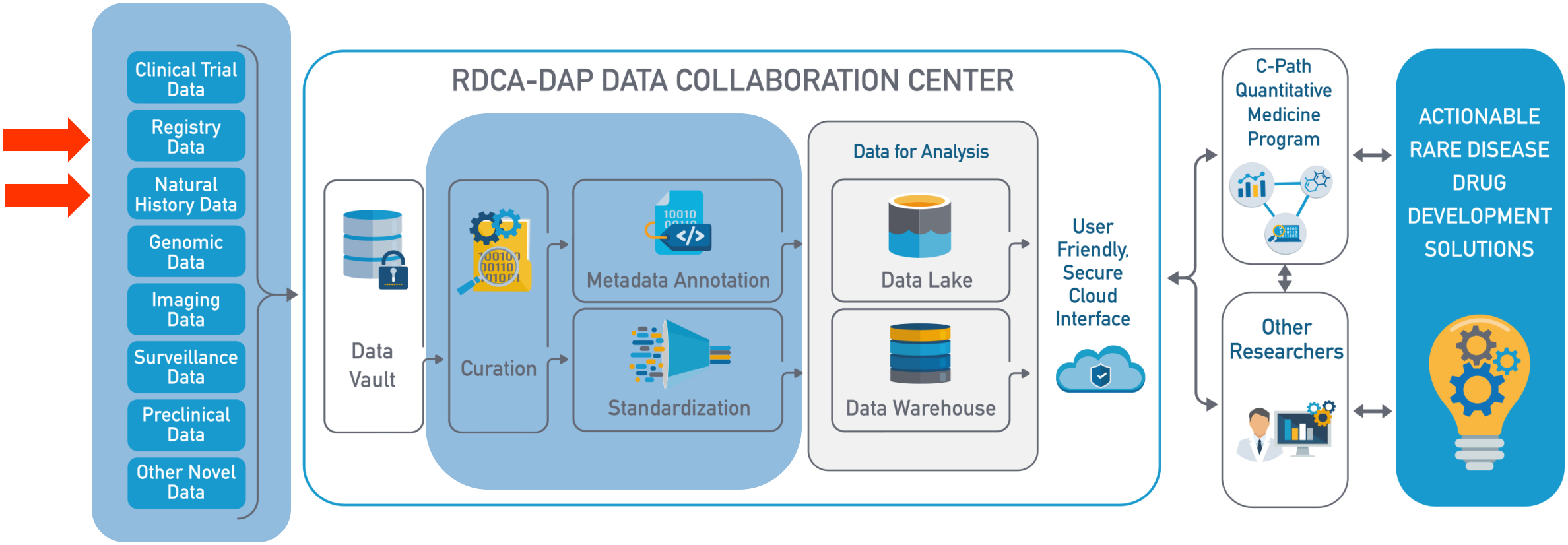
How do larger, standardized datasets help my disease state?

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



PKU Case Study: Background

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

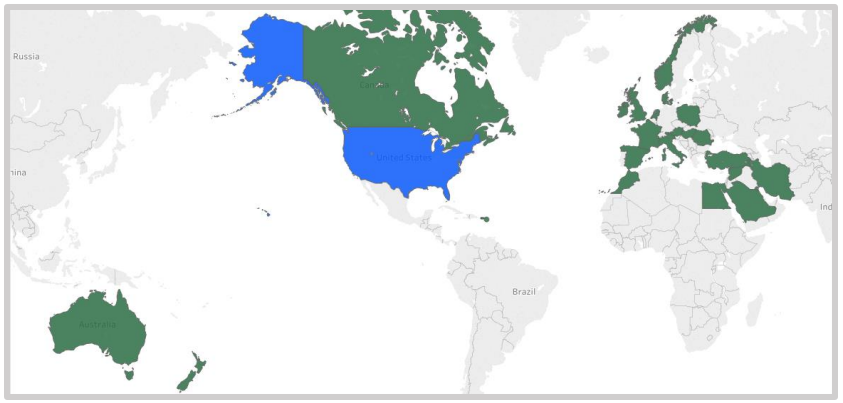
Organization: The National PKU Alliance, formed in 2008, with the goal of improving the lives of families and individuals associated with PKU through research, support, education and advocacy, while ultimately seeking a cure.

Registry:

- January 2017
- Open ended longitudinal study
- Open enrollment
- Currently ~1200 participants

National PKU Alliance

Data driven insights



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.

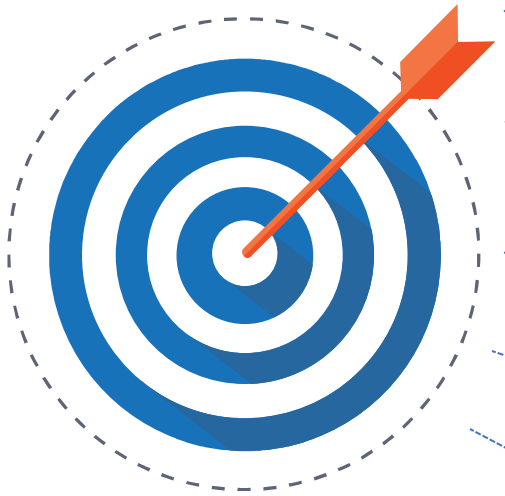
- Distribution of Reported Blood Phenylalanine Levels by Participants
- Types of Medical Formula used by Participants
- Mean Blood Phenylalanine Levels by Types of Medical Formula Used by Participant
- Low Protein Foods Used by Participants
- Mean Blood Phenylalanine Levels by Types of Low Protein Foods Used by Participant
- Other PKU Specific Treatments Used by Participants
- Mean Blood Phenylalanine Levels by PKU Specific Treatments

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

Have you ever been on a PKU Diet?	Are you currently on a PKU Diet?	Have you ever returned to a PKU Diet?	Number of Participants
No	No	No	<5
Yes	No	Don't know	<5
Yes	No	No	25
Yes	No	Yes	13
Yes	Yes	NA	507

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

Acknowledgements

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

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THANK YOU!

Don't forget to answer survey questions.

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

#RDCADAP