NORD®
National Organization for Rare Disorders

USING REGISTRY DATA TO EMPOWER YOUR ORGANIZATION AND ENGAGE WITH RESEARCHERS

NORD®
National Organization for Rare Disorders

ACD
ASSOCIATION FOR CREATINE DEFICIENCIES

HOW TO LAUNCH A REGISTRY IN SIX MONTHS:

A case study from the
Association for Creatine Deficiencies

CONSIDERATIONS

To launch the registry in six months, the Association for Creatine Deficiences (ACD) had the following priorities:

- Start with initial, short surveys and ensure that relevant information is shared with the research and patient community from the beginning
  - Drive consistent engagement from the community: make it easy for parents
- Work with a dedicated person to coordinate the project who can engage with multiple researchers and stakeholders including our parent community
- Be the hub for patient-reported data and ensure as much research as possible happens
  - The community owns the registry; the ACD Board has full authority to share the data, and values open science

CREATINEINFO TIMELINE OVERVIEW

Funding, Platform

CreatineINFO™

Outreach, Consistent Engagement, Robust Data

October

Onboarding

Research

Project outline

November

1-1 calls with researchers

Defining the Initial Surveys

December

Data Dictionary

FamilyAdvisory Board onboarding

January

General Advisory Review

Beta-Testing Demo site

IRB folder prep

February

IRB folder submission

March

Registry Launch

Some steps that helped us to reach our goal:

Start with WHY

Build a Strategic Team

Select your tools

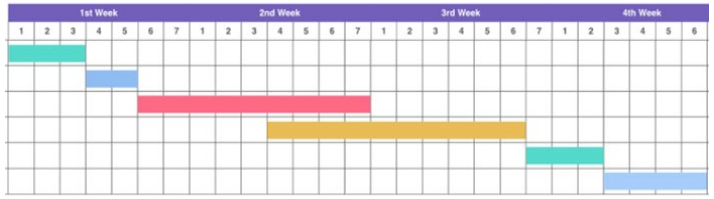
Partnerships and Networking groups

SELECT YOUR TOOLS

Before diving into the day-to-day tasks:
Look for **friendly project management tools** that can help you organize better:

Timelines

- Unify **your knowledge, platform guidelines and other guidelines** to create an initial timeline with all the "Known Tasks."
- It will change many times.
- Helps to visualize the project and adjust time if needed.



Canva Notion

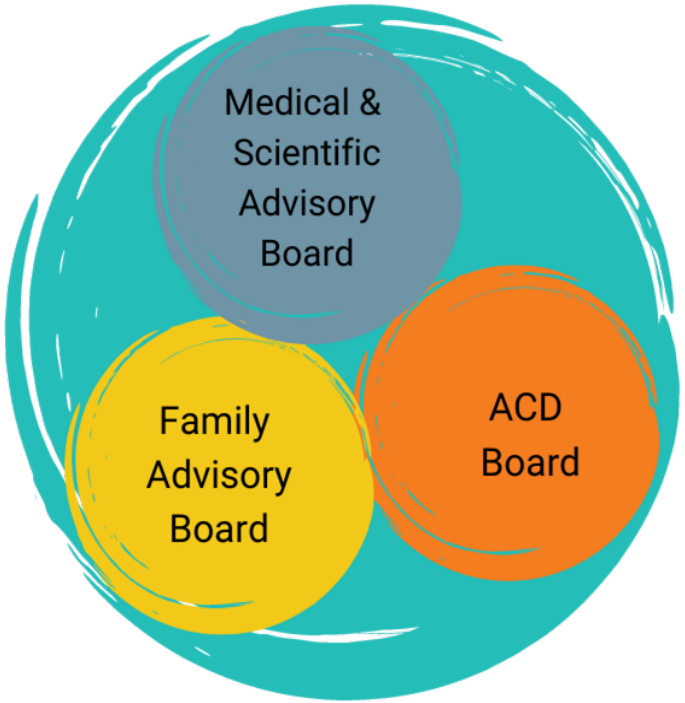
Checklists

- No matter how small the tasks are or how much you trust your memory, **writing everything down** can help you move forward easier and faster.
- Don't forget to **celebrate your progress.**

asana Google Keep

BUILD A STRATEGIC TEAM

CreatineINFO™



CreatineInfo Registry Team

Medical & Scientific Advisory Board:
Ensures all questions are relevant for research.
Provides expertise and research guidance.

Family Advisory Board:
Ensures patients' and caregivers' voices are represented.
Provides input and help with testing.

ACD Board:
Oversees the development of the registry and pursues all collaborations that can lead to treatments for Creatine Deficiencies.

PARTNERSHIPS & NETWORKING GROUPS

Partnerships:

ClinGen Data Sharing Program

- Helps in the understanding of genetic variants for Creatine Deficiencies.
- Provides **curated de-identified genotype data** that we can share directly with researchers.

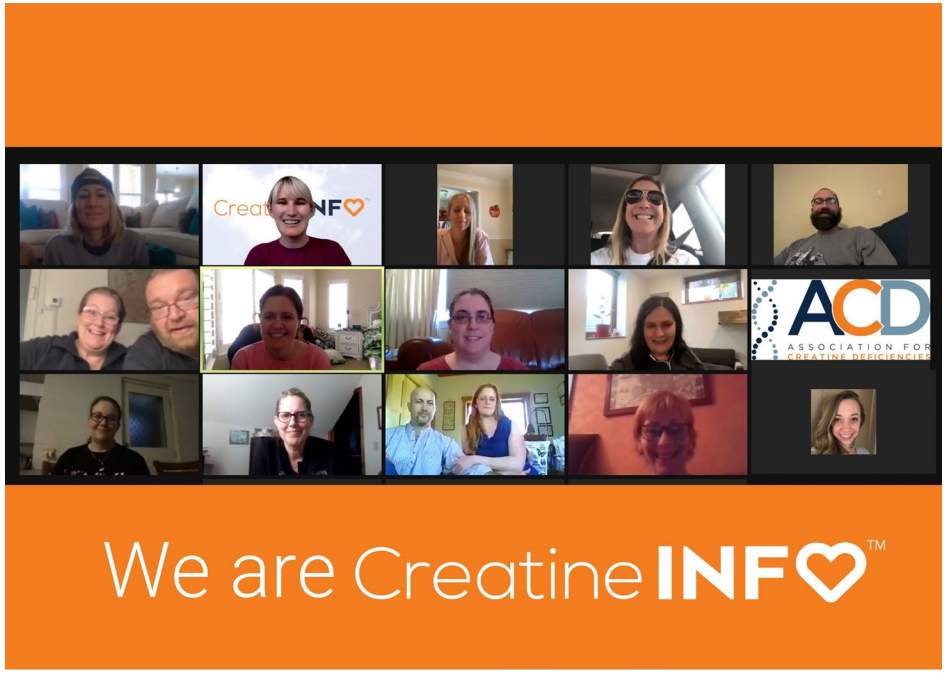
NORD®

- Platform, ongoing support, training, community

Networking groups:

- CZI Rare as One Workstreams
- NORD community forum

THANK YOU!
LET'S CONNECT



If you have any questions or would like to connect:

registry@creatineinfo.org

Social Media: @creatineinfo

Website: creatineinfo.org

Registry: creatineinfo.iamrare.org

NORD®
National Organization for Rare Disorders

A RARELAUNCH® FOR IMMUNE THROMBOCYTOPENIA (ITP)

Caroline Kruse, President and CEO
Platelet Disorder Support Association (PDSA)

Platelet Disorder Support Association
Empowering ITP Patients

Platelet Disorder Support Association
Empowering ITP Patients

RESEARCH READY:
The ITP Natural History Study Registry

Launch Date:
02/28/17

Total Registry Participants:
1,413

Total Countries Represented:
23

TRANSFORMING THE FUTURE WITH PATIENT-POWERED DATA

RESEARCH FOCUS ON QUALITY OF LIFE:

including fatigue, anxiety, mental health and patient experience

POSTERS

- American Society of Hematology (ASH) - **3**
- European Hematology Association (EHA) - **3**
- National Organization for Rare Disorders - **2** (NORD)

PUBLICATIONS

- *Blood* - **2**

PRESENTATIONS

- ASH Friday Morning ITP Breakfast – **1**

Platelet Disorder Support Association
Empowering ITP Patients

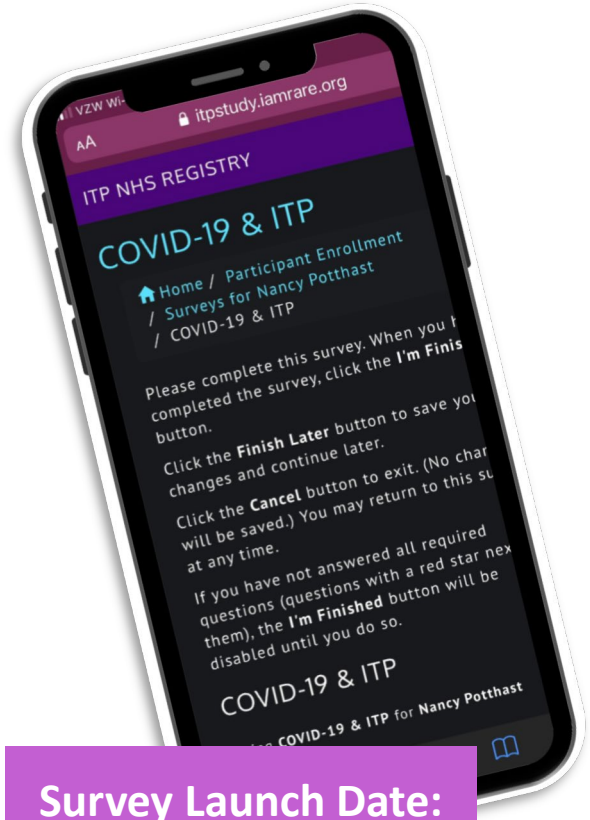
IMMUNE THROMBOCYTOPENIA (ITP):
the most common autoimmune bleeding disorder

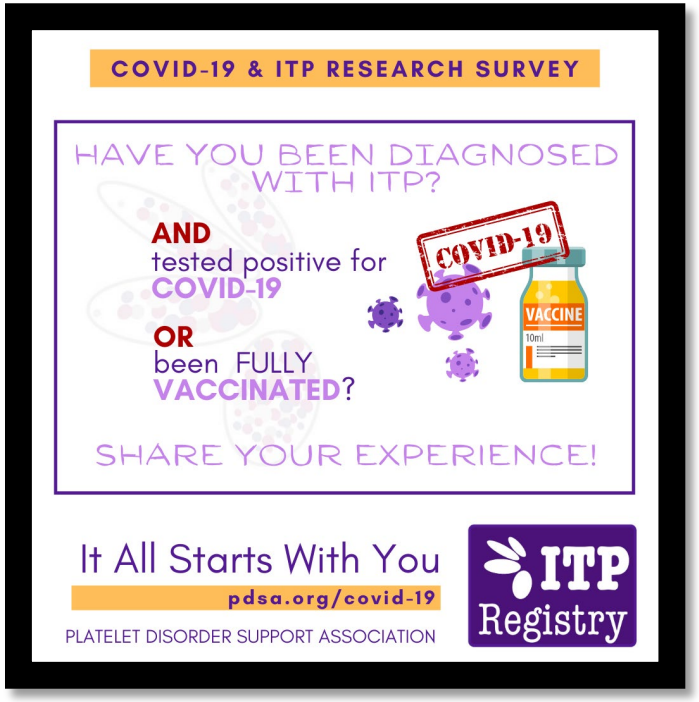
ITP does not discriminate. It affects both sexes, all ages, races and ethnicities.

Total U.S. Cases:
60,000 - 100,000

New Cases:
5,000 - 6,000 yearly

PANDEMIC PREPARED: COVID-19 & ITP





Survey Launch Date: 02/13/21

Communications Plan: social, e-comm, print, & web

Total COVID Surveys Completed: 155

Thank you.

U.S. FOOD & DRUG ADMINISTRATION

Role of the Patient and Patient Registries: Rare Disease Cures Accelerator

Michelle Campbell, PhD

Office of Neuroscience

CDER

Key Activities Presenting Areas of Challenge

Discovery / Translational / Preclinical **Clinical Development**

Characterization of Disease

- What is known about the disease?
- Are there well-defined lab tests to diagnose the disease?
- What is the natural history of the disease?
- What causes the disease (pathogenesis)?

Getting Patient Perspectives on their Disease and Treatment

- What disease impacts matter most to patients?
- What is the landscape of currently available treatments?

Clinical Study of New Treatments

- Is the investigational drug available in a form that can be administered?
- Pre-clinical safety testing done to inform assessment of safety in humans?
- A study design specified?
- A study protocol?
- IRB review and approval?
- IND submitted for FDA review?
- Plan for patient enrollment?
- Patient access to the trial site?
- Plan for study data collection?

Background: 5+ Years of Listening to Patients' Perspectives in PFDD Meetings

Patients are uniquely positioned to inform regulator understanding of the burden of disease and available treatment

Patients with chronic serious disease **are experts** on what it's like to live with their condition

Their **"chief complaints" may not be factored explicitly into drug development** plans

PFDD meetings elicit patient input to better inform clinical context of BR assessment. Patient stakeholders also asked: **What's next?**

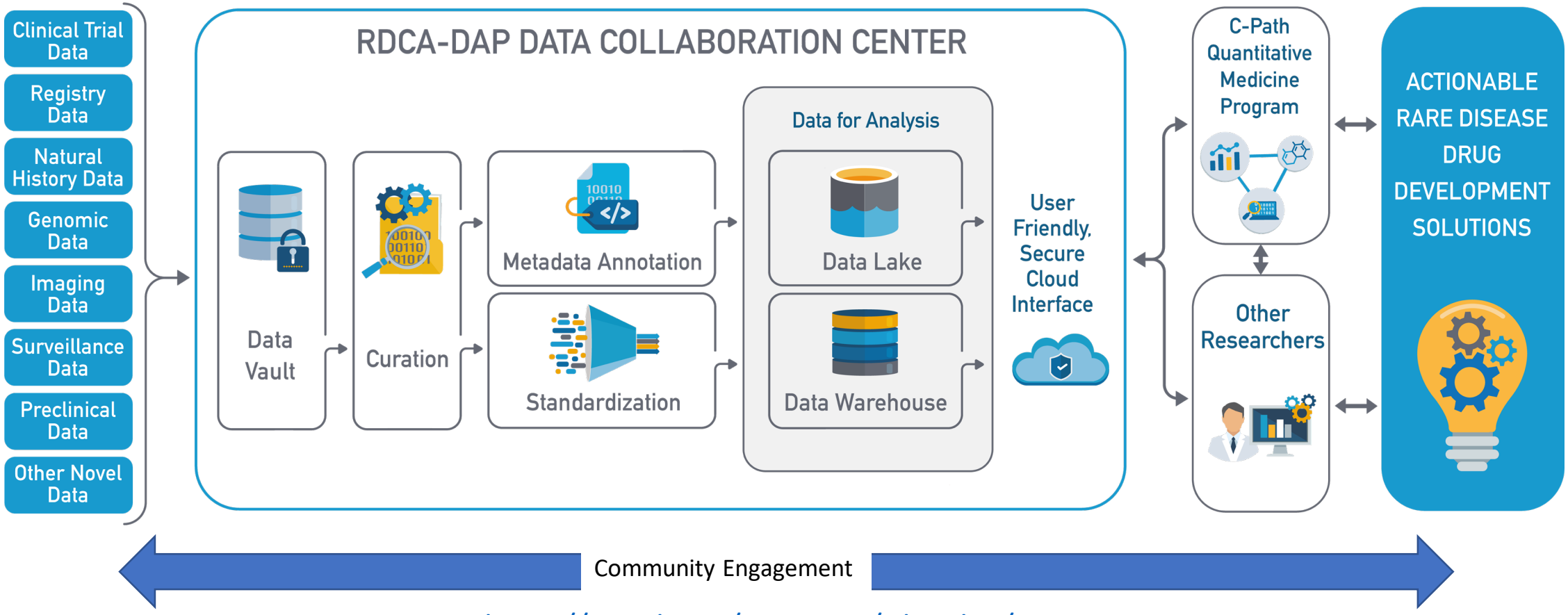
RDCA-DAP®

The primary objective:

- Establish a data management and data repository system
- Which will house data from existing and planned rare disease clinical studies and trials
- Data to be contributed from different organizations

RDCA-DAP®

- Critical Path Institute and NORD partnering on initiative



https://c-path.org/programs/rdca-dap/

Thank you and questions?

Thank you.