USING REGISTRY DATA TO EMPOWER YOUR ORGANIZATION AND ENGAGE WITH RESEARCHERS
HOW TO LAUNCH A REGISTRY IN SIX MONTHS:

A case study from the Association for Creatine Deficiencies
To launch the registry in six months, the Association for Creatine Deficiencies (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

October
- Onboarding
- Research
- Project outline

November
- 1-1 calls with researchers
- Defining the Initial Surveys

December
- Data Dictionary
- Family Advisory Board onboarding

January
- General Advisory Review
- Beta-Testing Demo site
- IRB folder prep

February
- IRB folder submission
- IRB folder prep

March
- Registry Launch

Funding, Platform
Outreach, Consistent Engagement, Robust Data
Some steps that helped us to reach our goal:

- Start with WHY
- Select your tools
- Build a Strategic Team
- Partnerships and Networking groups
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.

**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.
BUILD A STRATEGIC TEAM

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
If you have any questions or would like to connect:

registry@creatineinfo.org

Social Media: @creatineinfo
Website: creatineinfo.org
Registry: creatineinfo.iamrare.org
A RARE LAUNCH® FOR IMMUNE THROMBOCYTOPENIA (ITP)

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

COVID-19 & ITP RESEARCH SURVEY

HAVE YOU BEEN DIAGNOSED WITH ITP?

AND tested positive for COVID-19

OR been FULLY VACCINATED?

SHARE YOUR EXPERIENCE!

It All Starts With You

Platelet Disorder Support Association
Thank you.
Role of the Patient and Patient Registries: Rare Disease Cures Accelerator

Michelle Campbell, PhD
Office of Neuroscience
CDER
### Key Activities Presenting Areas of Challenge

<table>
<thead>
<tr>
<th>Discovery / Translational / Preclinical</th>
<th>Clinical Development</th>
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</thead>
<tbody>
<tr>
<td><strong>Characterization of Disease</strong></td>
<td><strong>Clinical Study of New Treatments</strong></td>
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<tr>
<td>• What is known about the disease?</td>
<td>• Is the investigational drug available in a form that can be administered?</td>
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<tr>
<td>• Are there well-defined lab tests to diagnose the disease?</td>
<td>• Pre-clinical safety testing done to inform assessment of safety in humans?</td>
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<tr>
<td>• What is the natural history of the disease?</td>
<td>• A study design specified?</td>
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<tr>
<td>• What causes the disease (pathogenesis)?</td>
<td>• A study protocol?</td>
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<tr>
<td><strong>Getting Patient Perspectives on their Disease and Treatment</strong></td>
<td>• IRB review and approval?</td>
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<tr>
<td>• What disease impacts matter most to patients?</td>
<td>• IND submitted for FDA review?</td>
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<tr>
<td>• What is the landscape of currently available treatments?</td>
<td>• Plan for patient enrollment?</td>
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<tr>
<td></td>
<td>• Patient access to the trial site?</td>
</tr>
<tr>
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<td>• Plan for study data collection?</td>
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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?**
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
• Critical Path Institute and NORD partnering on initiative

RDCA-DAP®

RDCA-DAP DATA COLLABORATION CENTER

Data for Analysis

Data Lake

Data Warehouse

Metadata Annotation

Standardization

Curation

Data Vault

C-Path Quantitative Medicine Program

Other Researchers

User Friendly, Secure Cloud Interface

ACTIONABLE RARE DISEASE DRUG DEVELOPMENT SOLUTIONS

https://c-path.org/programs/rdca-dap/
Thank you and questions?
Thank you.