Welcome!

Please be seated by 8:55 AM ET

The webinar will go live at 9:00 AM ET

Advisory Panel on Rare Disease: In-Person Meeting

October 27, 2016

9:00 AM – 4:00 PM

Welcome, Introductions, and Setting the Stage

Parag Aggarwal, PhD
Senior Program Officer, *Addressing Disparities*

Danielle Whicher, PhD, MHS
Program Officer, *Clinical Effectiveness Research*

Vincent Del Gaizo
Acting Chair, Rare Disease Advisory Panel

Housekeeping

- Today's meeting is open to the public and is being recorded
  - Members of the public are invited to listen to the teleconference and view the webinar
  - Meeting materials can be found on the PCORI website
  - Anyone may submit a comment through the webinar chat function, although no public comment period is scheduled
- Visit www.pcori.org/events for more information

Housekeeping (cont.)

- We ask that panelists stand up their tent cards when they would like to speak and use the microphones
- Please remember to state your name when you speak

Agenda

Agenda Item	Time
Welcome, Introductions, and Setting the Stage	9:00-9:15 AM
Background on the RDAP	9:15-10:00 AM
Overview of PCORI Rare Disease Research Portfolio	10:00-11:00 AM
Break	11:00-11:15 AM
Overview of Eugene Washington Engagement Awards and Pipeline to Proposal Awards Focused on Rare Diseases and Presentation of Results	11:15-12:15 PM
COI Forms & Lunch	12:15-1:00 PM
PCORnet Rare Disease Patient Powered Research Networks (PPRNs)	1:00-1:45 PM
FDA Presentation: Rare Disease Activities at the FDA	1:45-2:30 PM
Rare Disease Methodology Paper	2:30-3:00 PM
Recognition of Panelists Rolling off	3:00-3:30 PM
Wrap Up and Next Steps	3:30-3:45 PM

Introductions

- Please quickly state the following:
  - Name
  - Stakeholder group you represent
  - Position title and organization

Introductions: Current Panelists

Introductions (cont.)

Jacqueline Alikhaani, BA, MBA Candidate

Volunteer Heart Survivor Spokesperson, *American Heart Association*

Representing: Patients, Caregivers, and Patient Advocates

Introductions (cont.)

Naomi Aronson, PhD

Executive Director, *Clinical Evaluation, Innovation, and Policy, Blue Cross and Blue Shield Association (BCBSA)*

Ex-Officio Member from PCORI's Methodology Committee

Introductions (cont.)

Marilyn Bull, MD, FAAP

Morris Green Professor of Pediatrics, *Indiana University School of Medicine*

Representing: Clinicians

Introductions (cont.)

Vincent Del Gaizo (*Acting Chair*)

Owner, *Plaza Dry Cleaners*

Representing: Patients, Caregivers and Patient Advocates

Introductions (cont.)

Uday U. Deshmukh, MD, MPH

Chief Medical Officer, *HealthHelp*

Representing: Payers

Introductions (cont.)

Sindy N. Escobar-Alvarez, PhD

Senior Program Officer for Medical Research, *Doris Duke Charitable Foundation*

Representing: Researchers

Introductions (cont.)

Patricia Furlong

Founder, President and CEO, *Parent Project Muscular Dystrophy*

Representing: Patients, Caregivers, and Patient Advocates

Introductions (cont.)

Mardi Gomberg-Maitland, MD, MSc

Associate Professor of Medicine and Director, *Pulmonary Hypertension Program, The University of Chicago*

Representing: Clinicians

Introductions (cont.)

Lisa Heral, RNBA, CCRC*

Registered Nurse, *Pacific Quest and Bay Clinic - Hawaii*

Representing: Patients, Caregivers, and Patient Advocates

* Attending via teleconference

Introductions (cont.)

Kate Lorig, MS, MPH, DrPH

Professor Emerita and Director of the Stanford Patient Education Research Center, *Stanford University*

Representing: Patients, Caregivers, and Patient Advocates

Introductions (cont.)

Philip W. Ruff, PhD, BSc, CChem MRSC, Csci

Director, *Global Market Access, Shire Pharmaceuticals*

Representing: Industry

Introductions (cont.)

Mark W. Skinner, JD

President/CEO, *Institute for Policy Advancement, Ltd.*

Representing: Patients, Caregivers, and Patient Advocates

Introductions (cont.)

James J. Wu, MSc, MPH

Senior Manager, *Global Health Economics, Amgen Inc.*

Representing: Industry

Introductions: New Panelists

Introductions (cont.)

Matt Cheung, PhD, RPh

Adjunct Professor, *Pharmacy Practice, University of the Pacific*

Representing: Payers

Introductions (cont.)

Kathleen Gondek, MS, PhD

Vice President, *Global Health Economics Outcomes Research and Epidemiology, Shire PLC*

Representing: Industry

Introductions (cont.)

Maureen Smith, MEd

Board Member, *Canadian Organization for Rare Disorders (CORD)*
Patient Member, *Ontario Ministry of Health and Long Term Care*

Representing: Patients, Caregivers, and Patient Advocates

RDAP Panelists (cont.)

Michael Kruer, MD**

Representing: Researchers

Yaffa Rubinstein, MS, PhD**

Representing: Researchers

Marshall Summar, MD**

Representing: Clinicians

Rare Disease Advisory Panel – PCORI Staff

Parag Aggarwal, PhD
Senior Program Officer,
Addressing Disparities

Dionna Attinson
Program Assistant,
Addressing Disparities

Sarah Philbin, MPH
Program Associate,
Clinical Effectiveness Research

Danielle Whicher, PhD, MS
Program Officer,
Clinical Effectiveness Research

Rare Disease Advisory Panel: Background

Parag Aggarwal, PhD
Program Officer, Addressing Disparities

Evelyn Whitlock, MD, MPH
Chief Science Officer, PCORI

Danielle Whicher, PhD, MHS
Program Officer, Clinical Effectiveness Research

PCORI's Mission and Vision

PCORI helps people make informed healthcare decisions, and improves healthcare delivery and outcomes, by producing and promoting high-integrity, evidence-based information that comes from **research guided by patients, caregivers, and the broader healthcare community.**

Our Strategic Goals:

Increase quantity, quality, and timeliness of useful, trustworthy research information available to support health decisions

Speed the implementation and use of patient-centered outcomes research evidence

Influence research funded by others to be more patient-centered

RDAP's Mission and Vision – Charter

Purpose:

The Advisory Panel on Rare Disease (RDAP) will advise and provide recommendations to PCORI's Board of Governors, Methodology Committee, and staff on the conduct of patient-centered comparative clinical effectiveness research in rare diseases. It will provide recommendations on:

- How to improve the quality of rare disease applications received by PCORI
- Appropriate methods for designing studies and analyzing data from comparative clinical effectiveness research in rare diseases
- Approaches for recruiting and engaging patients
- Coordination and engagement with the rare disease research community, including other funders of research on rare diseases and existing infrastructure dedicated to research on rare diseases

RDAP's Mission and Vision – Charter (cont.)

Function and Scope of Work:

- Provide input to PCORI on research needs of the rare diseases community and on specific issues and concerns in conducting research on rare diseases
- Identify existing infrastructure (data sources, tools, etc.) that can be a resource for conducting research
- Serve on or assist in identifying experts to serve on ad hoc panels to assist in evaluating, designing and conducting PCORI-funded research specific to a rare disease
- Provide ongoing feedback and advice on evaluating and disseminating PCORI's research portfolio on rare diseases
- Consider study findings and advise on appropriate audiences and strategies for PCORI dissemination efforts
- Identify opportunities for collaboration with existing international, federal, public and private entities doing similar work in the rare disease space
- Advise other PCORI committees and panels to ensure the unique considerations of rare disease are addressed

PCORI Advisory Panels

- Program-specific:
  - Advisory Panel on Assessment of Prevention, Diagnosis, and Treatment Options
  - Advisory Panel on Improving Healthcare Systems
  - Advisory Panel on Addressing Disparities
  - Advisory Panel on Communication and Dissemination Research

- Cross-PCORI:
  - Advisory Panel on Patient Engagement
  - Advisory Panel on Rare Disease*
  - Advisory Panel on Clinical Trials*

History of the RDAP

- RDAP established: November 2013
- Inaugural in-person meeting: April 2014
- Key Activities:
  - Commissioned a review of standards for research on rare diseases. The review was discussed in May 2015 and a list of evidence gaps were identified
    - This lead to additional work to develop guidance on three additional topics
  - Recommended that PCORI create a pool of experts to address staff questions related to monitoring funded rare disease projects
  - Drafted a list of special considerations for merit review of rare disease CER projects and helped develop a set of guidelines for research on rare diseases
  - Served as technical experts to the Advisory Panel on Assessment of Options on the following research topic: “Genetic Testing for Rare Diseases”

Rare Disease Advisory Panel Meeting
October 27, 2016

Evelyn P. Whitlock, MD, MPH

Chief Science Officer

CSO Vision

Vision

- "One Science"
  - Consistent approach and supportive response to applicants and awardees
  - Strategic thinking around portfolio
  - Excellence, collegiality, camaraderie across and beyond department

- 2016 Goals
  - Establish Evidence Synthesis Program
  - Enhance integration of scientific programs within department and across PCORI
  - Improve interface and relationships with the researcher community
  - Align mission of advisory panels to overall PCORI direction

PCORI RESEARCH FRAMEWORK

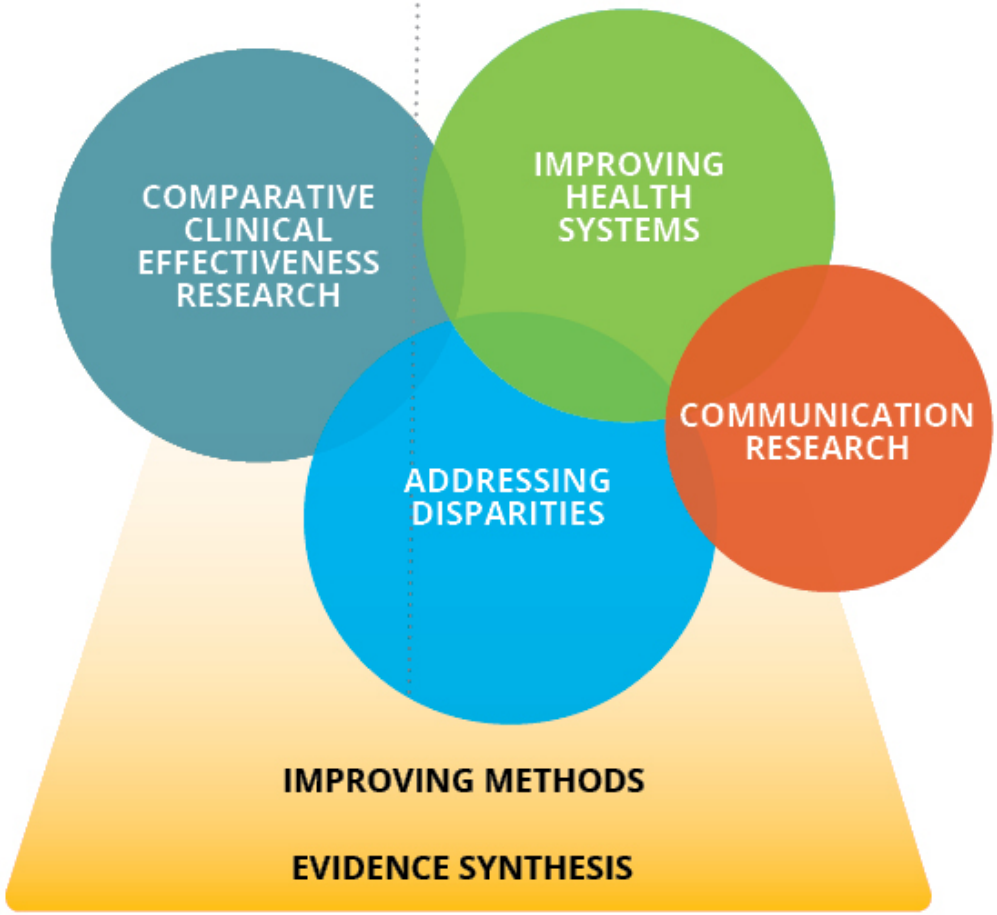
Producing the comparative clinical effectiveness research (CER) evidence to improve patient-centered outcomes and inform value considerations in healthcare decisions by patients, clinicians, payers, and policy makers.

PCORI RESEARCH FRAMEWORK

APPLICABLE EVIDENCE ▶ ▶ ▶ ▶ ▶ ▶ ▶ ▶ **INFORMED DECISION MAKING**

WHAT CARE IS BETTER FOR INDIVIDUAL PATIENTS?

HOW CAN PATIENT-CENTERED CARE BE BEST DELIVERED?



OUR ULTIMATE GOAL

IMPROVING PATIENT-CENTERED OUTCOMES

RDAP & PCORI

- RDAP is one of two legislatively mandated advisory panels
- Rare diseases research is an important challenge and opportunity
- Aligning RDAP's activities with our programmatic initiatives going forward could be fruitful

RDAP Opportunities

- PCORnet: PPRNs and CDRNs
- Existing Topic Pathway—Sickle Cell Disease
- Core outcome sets in rare disease
- Other: (n-of-1?)

Q&A

PCORI's Rare Disease Portfolio

Heather Edwards, PhD, MPH, MBA
Program Officer, Evaluation and Analysis

PCORI's Rare Disease Portfolio

Strategic Portfolio Analysis, Evaluation & Analysis

October 27, 2016

pcori

PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

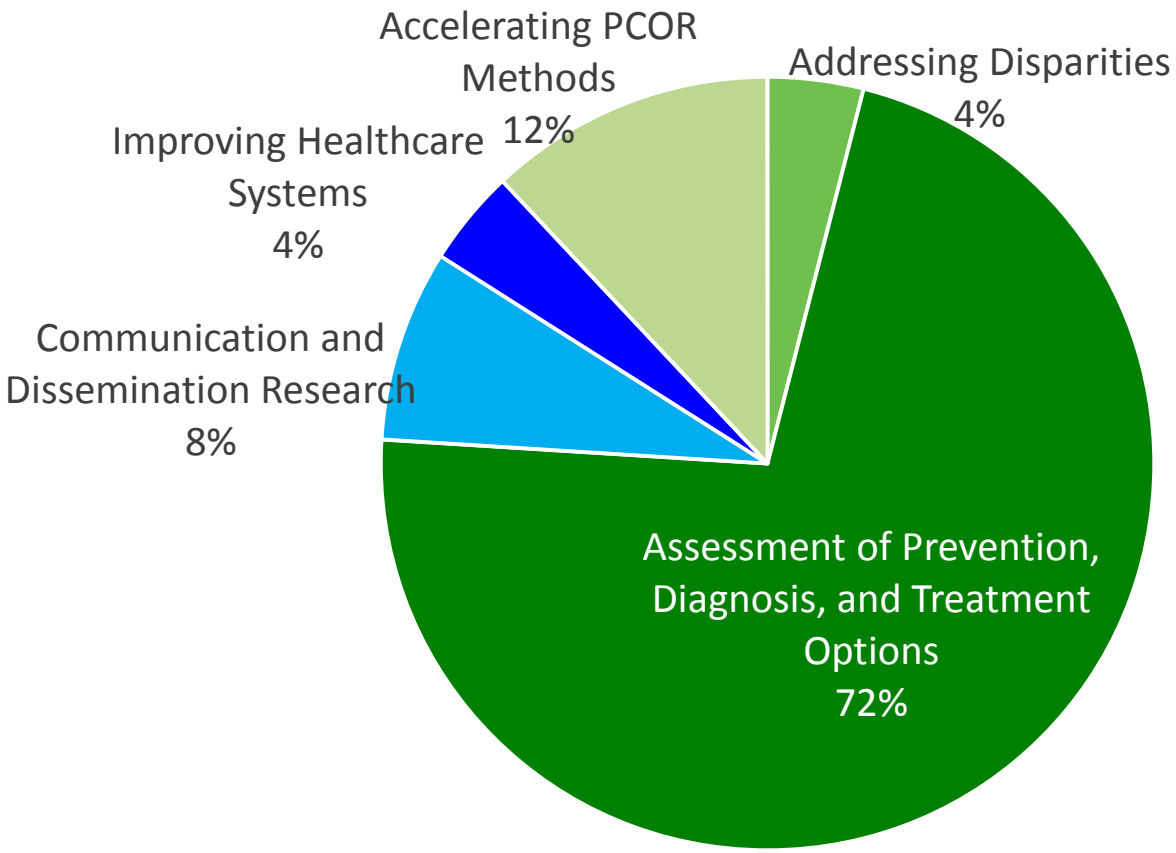
Rare Disease Portfolio

Overview

As of September 2016, PCORI has **22** active or completed comparative effectiveness research projects and **3** methods projects on rare disease, totaling **$57.3 million.**

Rare Disease Portfolio

Proportion of projects by PCORI priority area



N=25
Categories are mutually exclusive
Active and completed projects awarded through September 2016 (Cycle 3 2015)

Rare Disease Portfolio

Accelerating PCOR Methods Projects

Patient Centered Adaptive Treatment Strategies (PCATS) using Bayesian Causal Inference

Bin Huang, PhD
Cincinnati Children's Hospital Medical Center
Cincinnati, OH

Engaging Patients and Caregivers Managing Rare Diseases to Improve the Methods of Clinical Guideline Development

Dmitry Khodyakov, PhD, MA
RAND Health
Santa Monica, CA

Design and Methodological Improvements for Patient-Centered Small n Sequential Multiple Assignment Randomized Trials (snSMARTs) in the Setting of Rare Diseases

Kelley Kidwell, PhD,
University of Michigan
Ann Arbor, MI

Rare Disease Portfolio

Number of projects across care continuum

N=22
Categories are mutually exclusive
Active and completed projects awarded through September 2016 (Cycle 3 2015)
Excludes Methods

Rare Disease Portfolio

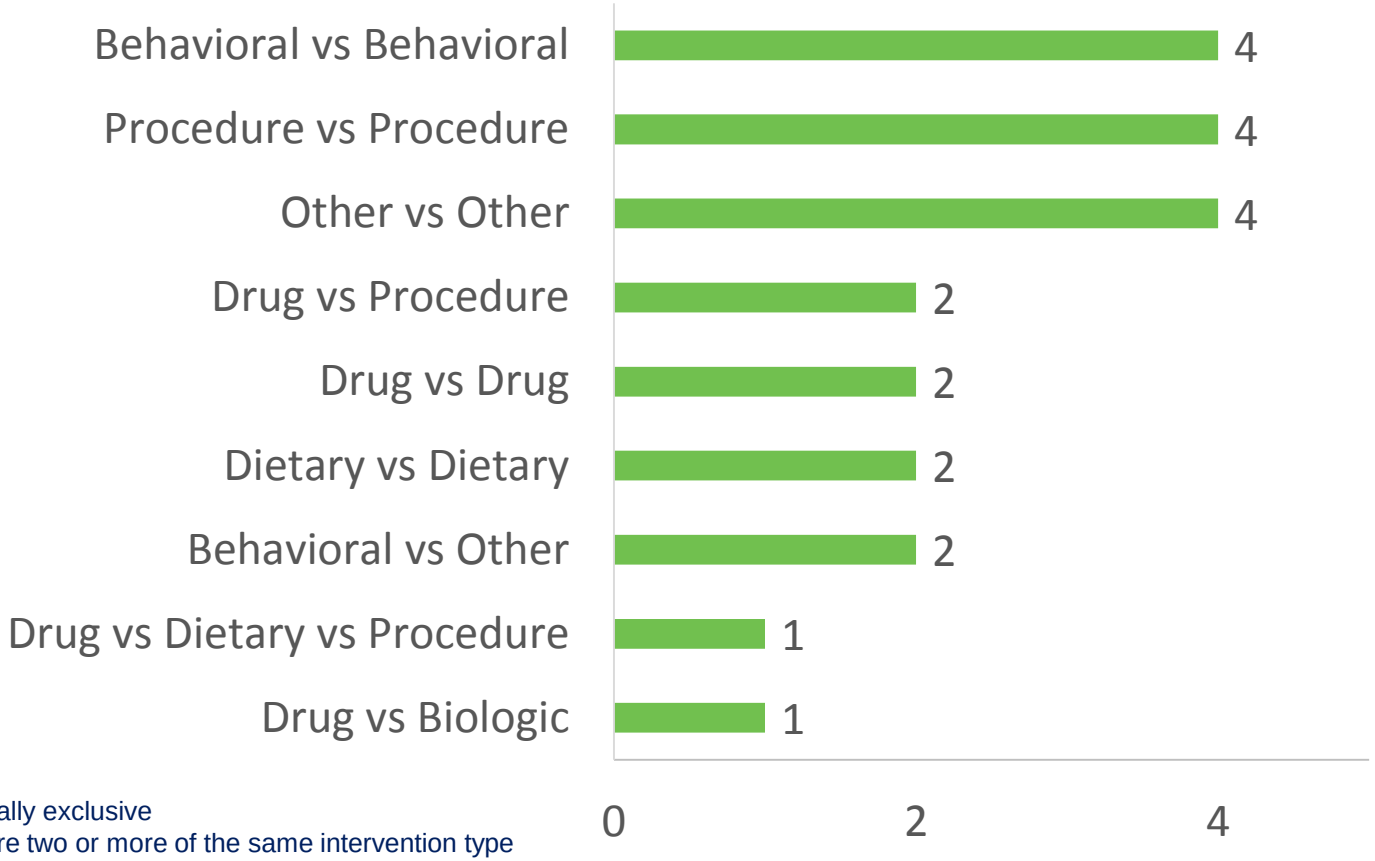
Specific conditions

Acute Myeloid Leukemia
Cerebral palsy
Chiari type I malformation (CM) & syringomyelia (SM)
Disorders of Sex Development
Duarte galactosemia
Eosinophilic Esophagitis
Hydrocephalus
Idiopathic Subglottic Stenosis
Lupus nephritis
Non-CF bronchiectasis
Pediatric Crohn's Disease
Pediatric Transverse Myelitis
Polyarticular Juvenile Idiopathic Arthritis
Sickle Cell Disease
Spinal Cord Injury and Spina Bifida
Systemic Scleroderma
Urea cycle disorders

Projects awarded through September 2016 (Cycle 3 2015)
Excludes Methods

Rare Disease Portfolio

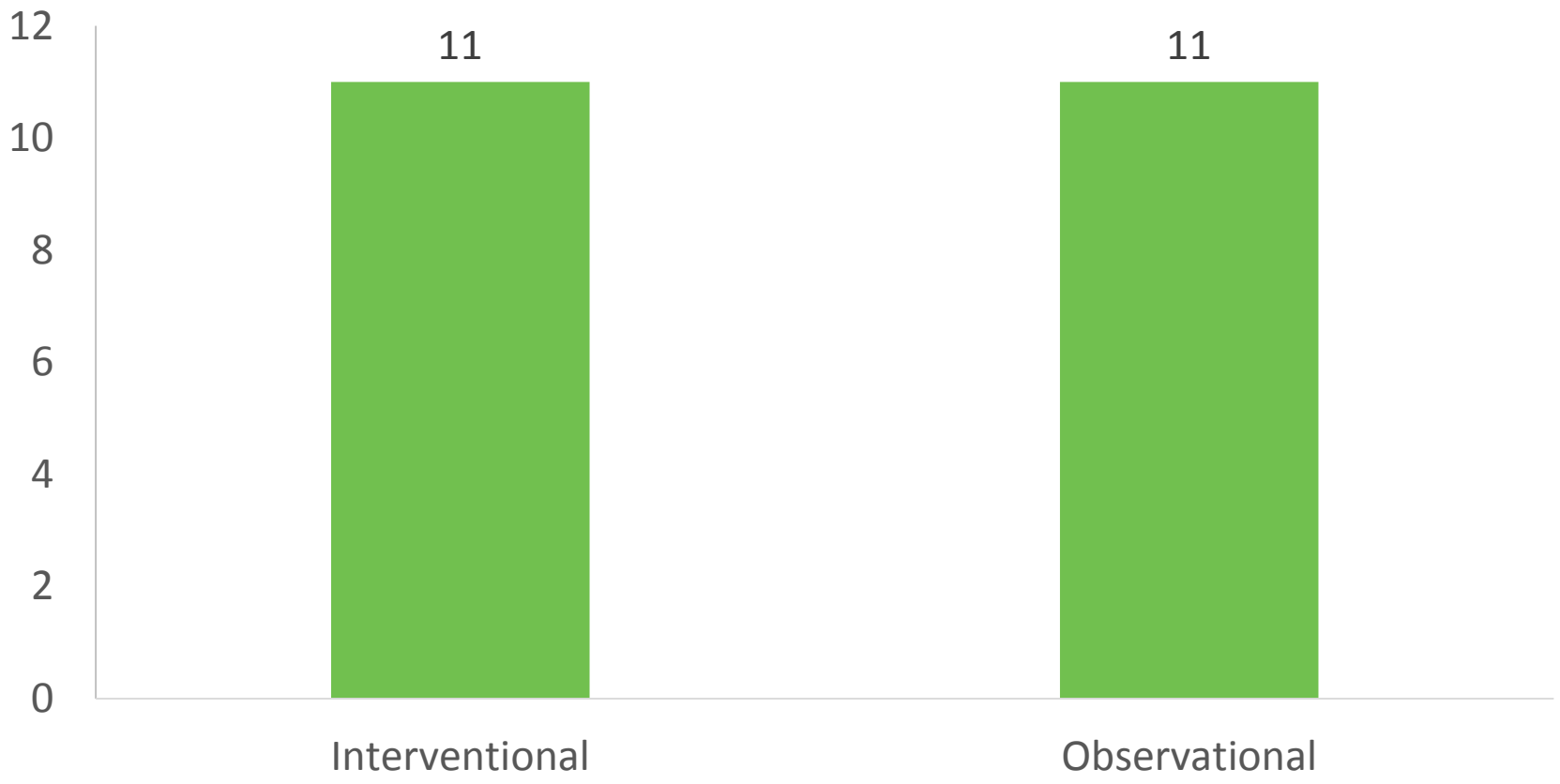
Number of projects by intervention type



N=22
Categories are mutually exclusive
Projects may compare two or more of the same intervention type
Active and completed projects awarded through September 2016 (Cycle 3 2015)
Excludes Methods

Rare Disease Portfolio

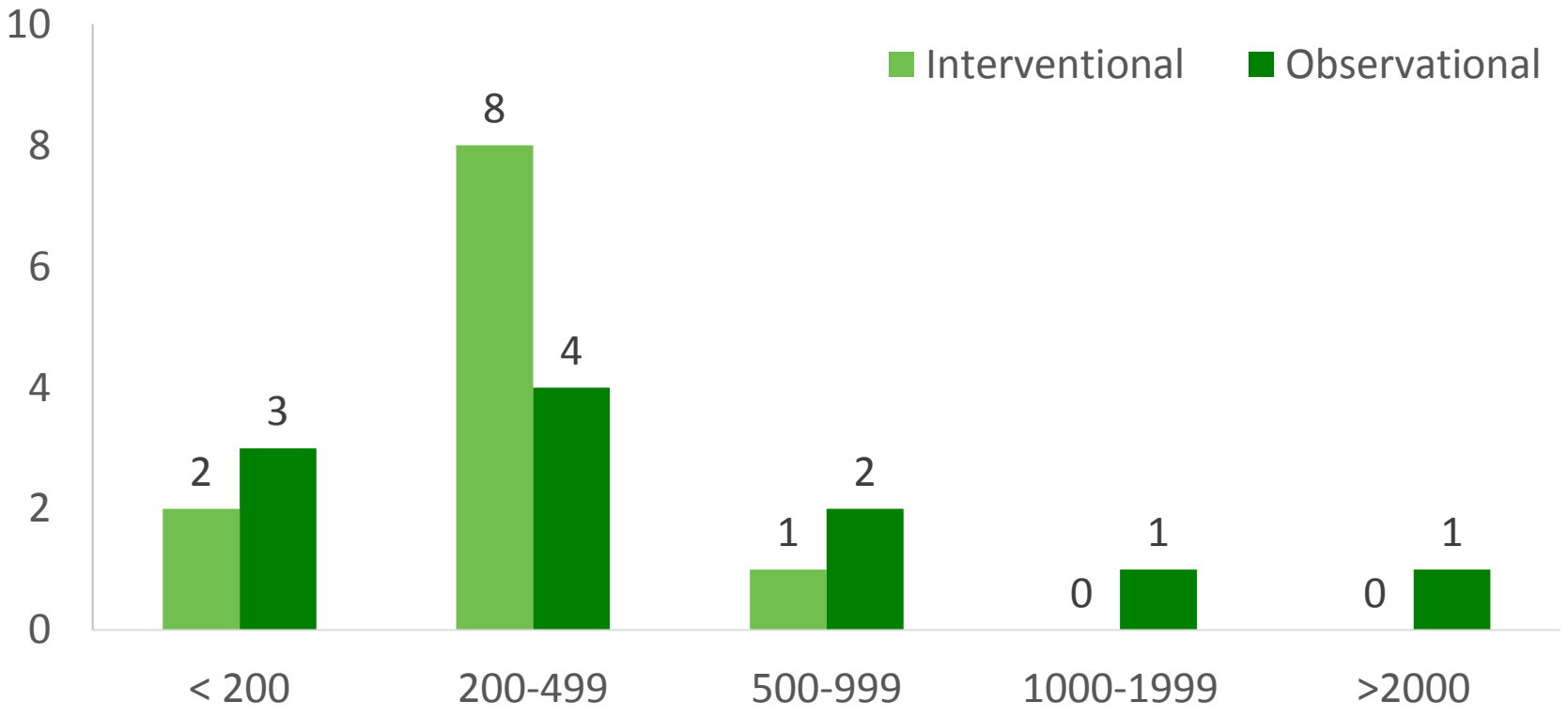
Number of projects by study design



N=22
Active and completed projects through September 2016 (Cycle 3 2015)
Excludes Methods

Rare Disease Portfolio

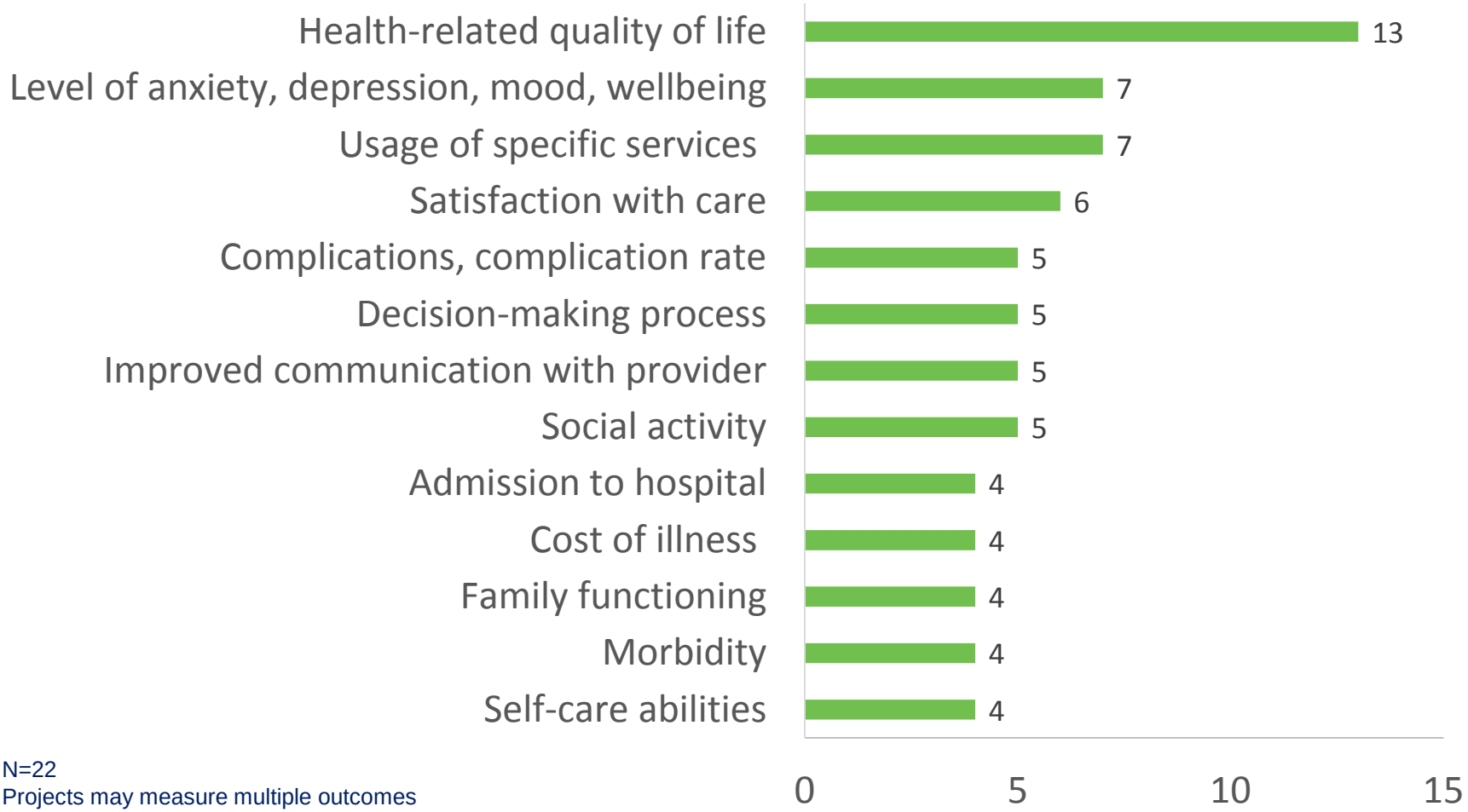
Number of projects with each sample size, by design



N=22
Active and completed projects through April 2016 (Spring 2015 PCS)
Excludes Methods

Rare Disease Portfolio

Number of projects with each outcome



N=22
Projects may measure multiple outcomes
Active and completed projects through September 2016 (Cycle 3 2015)
Excludes Methods

Rare Disease Portfolio

Projects that Target Multiple Rare Diseases

Decision Support for Parents Receiving Genetic Information about Child's Rare Disease

David E. Sandberg, PhD
University of Michigan at Ann Arbor
Ann Arbor, MI

Enhancing Genomic Laboratory Reports to Enhance Communication and Empower Patients

Marc Williams, MD
Weis Center For Research-Geisinger Clinic

Rare Disease Portfolio

Specific conditions

Acute Myeloid Leukemia
Cerebral palsy
Chiari type I malformation (CM) & syringomyelia (SM)
Disorders of Sex Development
Duarte galactosemia
Eosinophilic Esophagitis
Hydrocephalus
Idiopathic Subglottic Stenosis
Lupus nephritis
Non-CF bronchiectasis
Pediatric Crohn's Disease
Pediatric Transverse Myelitis
Polyarticular Juvenile Idiopathic Arthritis
Sickle Cell Disease
Spinal Cord Injury and Spina Bifida
Systemic Scleroderma
Urea cycle disorders

Projects awarded through September 2016 (Cycle 3 2015)
Excludes Methods

RDAP Discussion

- What can PCORI do to increase outreach with the rare disease community and increase the number of rare disease research applications it receives?
- What is the unique role of PCORI in contribution to RD research?
  - What should and shouldn't PCORI be doing in rare disease?
- What are the challenges and opportunities associated with cross cutting studies that enroll individuals with a variety of rare diseases that share one or more common features?

Break

We will resume at 11:15 AM ET

PCORI's Engagement Funding Opportunities: A Focus on Rare Diseases

Lia Hotchkiss

Director, *Eugene Washington Engagement Awards*

Engagement Award Program Overview

- Programmatic funding opportunity, launched in Feb 2014
- Supports projects that will build a community better able to participate in PCOR/CER and serve as channels to disseminate study results
- Engagement Award projects will produce deliverables that are useful to awardees, PCORI, and the broader PCOR community for increasing patient and stakeholder engagement in PCOR/CER

Engagement Awards

- Engagement Award (EA) projects
  - build our knowledge base about how patients and other stakeholders want to participate in PCOR/CER or receive research findings;
  - implement training or skill development initiatives to build capacity for engaging in PCOR/CER; and/or
  - strengthen channels for disseminating PCOR/CER findings.

- Engagement Award Initiative Notice (EAIN) meetings/conferences
  - align with PCORI's mission and strategic plan, and facilitate expansion of PCOR/CER in areas such as:
    - research design and methodology
    - research development
    - dissemination and implementation

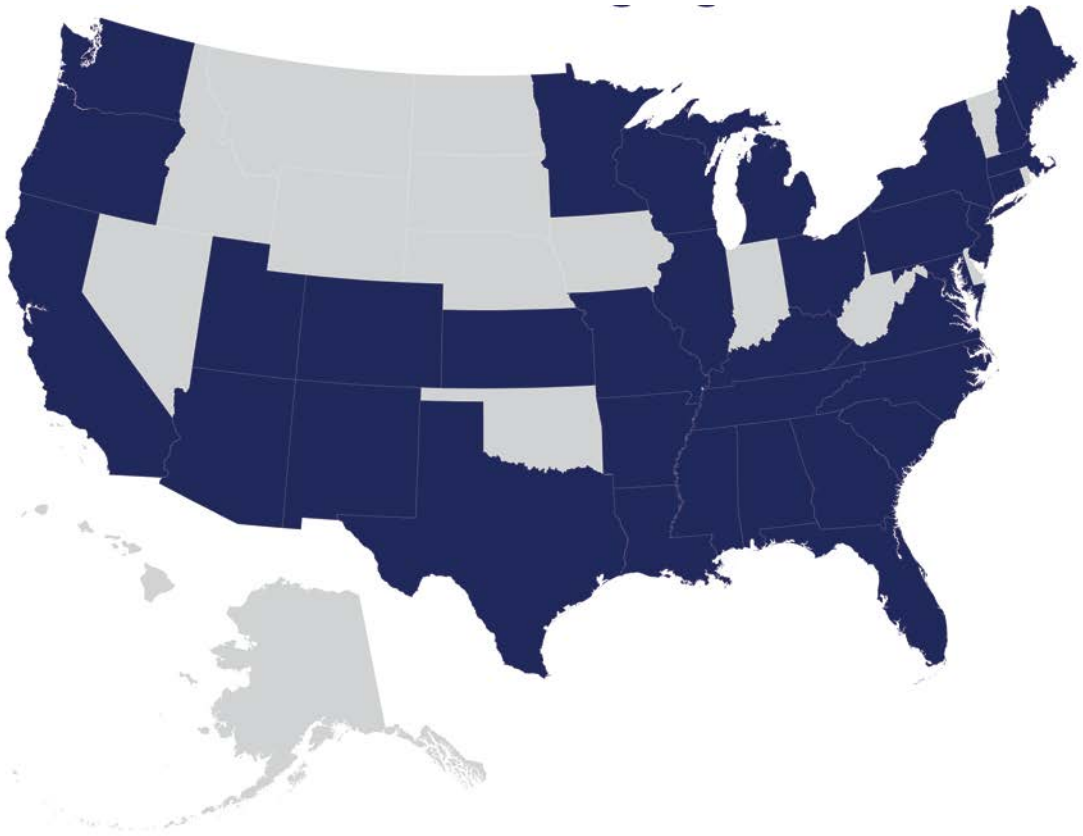
Awards of **up to $250,000** per project, up to **two years** in duration

Strengthening PCOR Nationwide: Engagement Award Program

Number of awards:
EA projects: 91
Conference support: 61

Amount awarded:
$29.6 M

Number of states where we are funding awards:
35 states



Examples of Engagement Award Projects Focused on Rare Disease Community

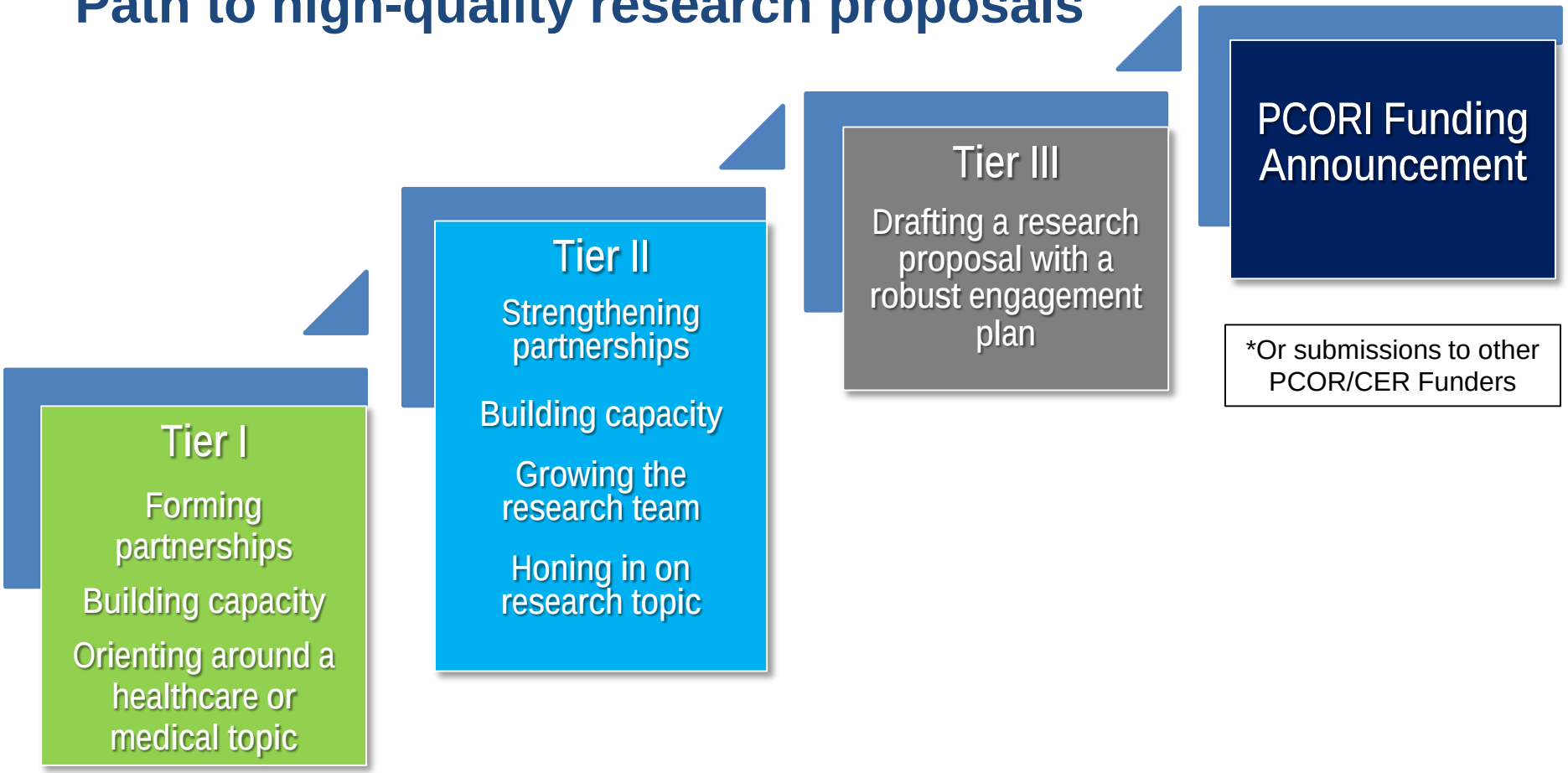
- **A Foundation for Building Strength: *Novel Stakeholder Engagement for Nemaline Myopathy, Patient Centered Research***
  - Ongoing collaboration with stakeholder groups to further encourage patient-centered research, foster better patient-physician communication, and develop a path to treatment for the rare disease
  - Collaborators: The Hospital for Sick Children; NINDS/NIH; Boston Children's Hospital; MIT NEW DIGS; NORD; University of Florida Muscle Lab; multi-stakeholder volunteers and conference panelists
- **University of Arkansas for Medical Sciences: *Developing and Testing "Best Practices" in Training for Academic/Community Research Partnerships***
  - Objective is to develop online training with usability and value to research partners, focusing on unique needs of rare and/or genetic condition patient-partner trainees
  - Expert panel partially comprised of rare disease representatives (patients with and providers focused on rare cancer and sickle cell disease) working to test the training modules

Examples of Engagement Awards for Conference Support Focused on Rare Disease Community

- **Alstrom Syndrome International**: It's Time to Talk! It's Time to Listen!
- **Aplastic Anemia & MDS International Foundation**: Patient Advisory Committee for Clinical Trials
- **Cure CMD**: Building Momentum Through Congenital Muscular Dystrophy Stakeholder Participation
- **Foundation for Prader-Willi Research**: Real World Data Research Readiness - Engaging the Prader-Willi Syndrome Community
- **Hydrocephalus Association**: Translation to Transform
- **Sturge-Weber Foundation**: The SWF Patient Engagement Conference
- **NEC Society**: Necrotizing Enterocolitis Symposium: A Transdisciplinary Approach to Improved NEC Outcomes
- **Phelan-McDermid Syndrome Foundation**: Phelan-McDermid Syndrome Patient-Centered Outcomes Workshop
- **PXE International**: Discovering What Matters Most
- **Sickle Cell Disease Association of America**: The National Sickle Cell Advocate Network
- **Vanderbilt University**: Tennessee Sickle Cell Disease Network

Pipeline to Proposals (P2P) Program

Path to high-quality research proposals

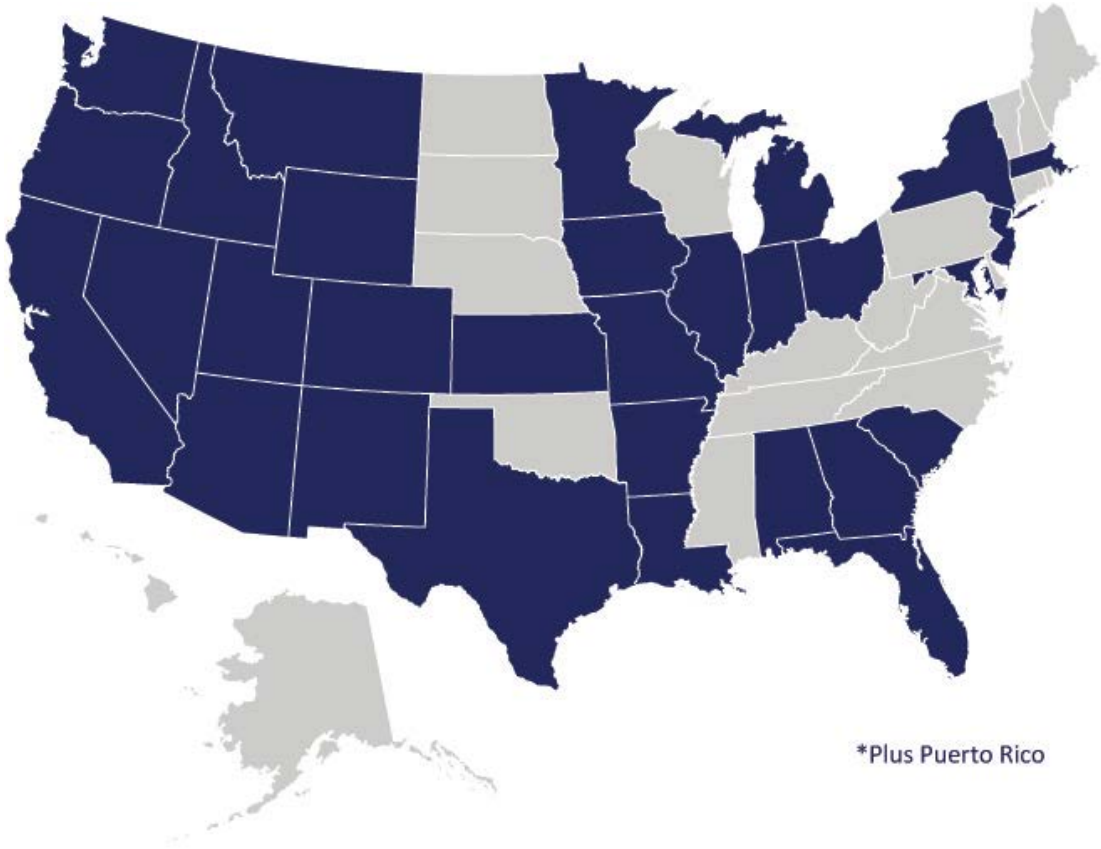


Strengthening PCOR Nationwide: P2P Program

Number of projects:
Tier I – 77
Tier II - 27

Amount awarded:
$4.7 M

Number of states where we are funding projects:
30 states and Puerto Rico



P2P Funded Rare Disease Projects

- **<u>Tier I – Stakeholder Engagement and Partnership Development for $15,000 over a 12-month period</u>**
  - Advancing Reproductive Health Wellness between Patients with **Cystic Fibrosis**, Their Caregivers, Healthcare Providers and Researchers
    - Emily M. Godfrey MD MPH FAAFP with University of Washington
  - Community Engagement to Improve Prevention and Treatment of **Congenital Cytomegalovirus Infection**
    - Laura Gibson, MD with University of Massachusetts Medical School
  - Developing Comparative Effectiveness Research in **Dravet Syndrome**
    - Mary Anne Meskis with Dravet Syndrome Foundation
  - Building an Empowered Patient Community: Tackling Health Care for All Affected by **Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome**
    - Amy C. Lossie, PhD with Beautiful You MRKH Foundation, Inc.

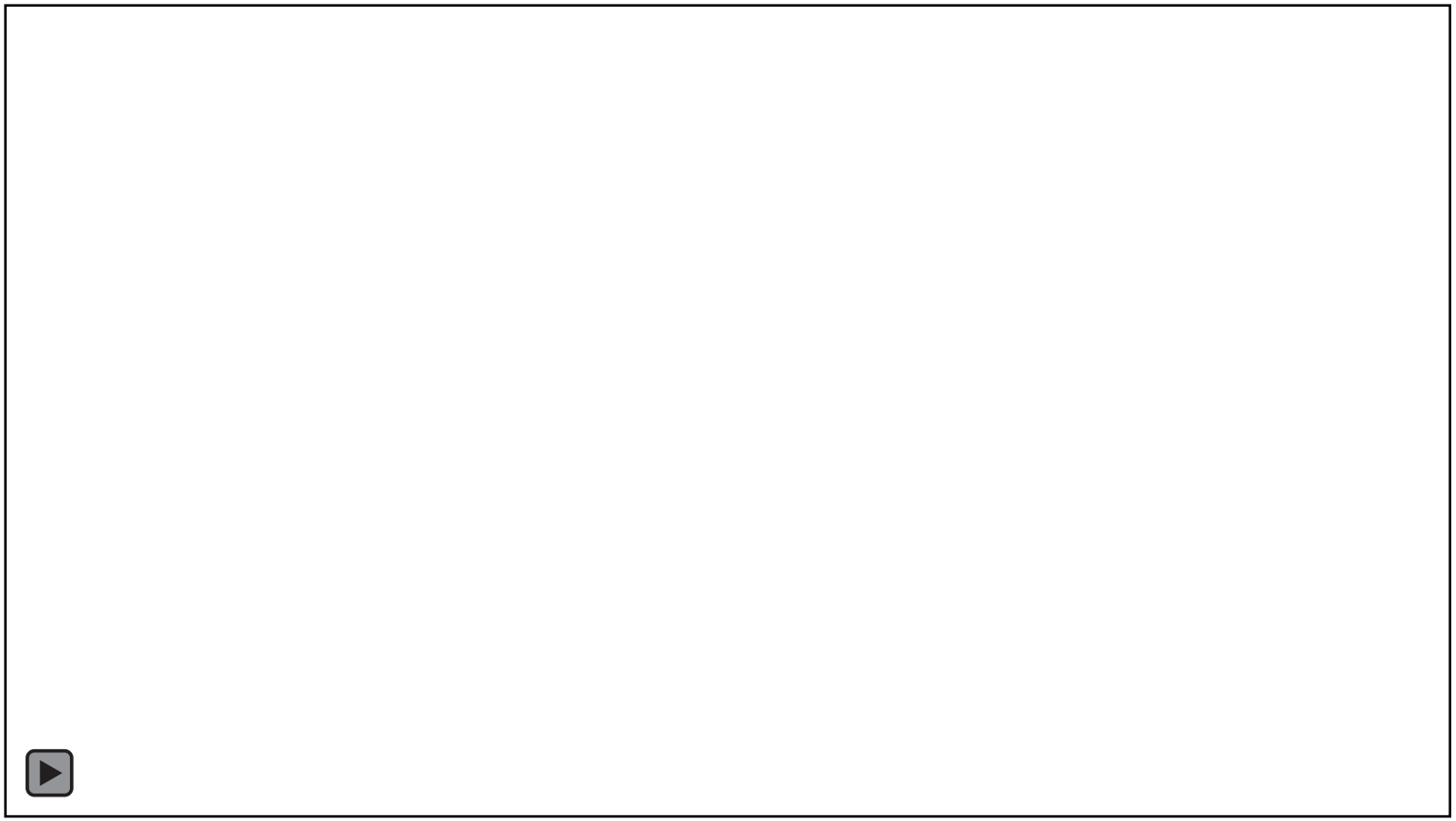
P2P Funded Rare Disease Projects

- **<u>Tier II – CER Question Development and Research Partnership Maturation for $25,000 over a 12-month period</u>**
  - Bridging **Rare Disease** Patients and Data through Novel Research Partnerships, Indiana
    - Catherine Fairchild, JD with Parkview Health
  - We'll Take the Village: Engaging the Community to Better Health
    - Mary Bentley LaMar with The **Sickle Cell** Association of New Jersey
  - What's the SCOOP? Discovering Quality-of-Life Outcomes That Matter to **Squamous Cell Carcinoma of the Oropharynx (SCOOP)** Patients and Their Families
    - Steven Chang, MD with Henry Ford Health System

P2P Funded Rare Disease Projects

- **<u>Tier III – CER Proposal Development for $50,000 over a 12-month period</u>**
  - **CysticLife**
    - Ronnie Sharpe with CysticLife
  - Addressing Obesity in Latino Adolescents with **Spina Bifida**/Supporting Latino Families with Children with Spina Bifida
    - Ruth Bush with Spina Bifida San Diego

From the Field…



Engagement Funding NOT Intended to Support:

- Projects solely intended to improve patient engagement in healthcare service delivery,
- Projects intended to increase the number of patients who agree to be research subjects or participants
- Research studies
- Planning or pilot studies
- Projects designed solely to validate tools or instruments
- Delivery of health care
- Development of registries or recruitment of research or registry participants
- Development of decision support tools or clinical practice guidelines
- Meetings that don't focus on PCOR or CER
- Full-fledged projects to translate PCORI research findings into products and/or disseminate PCORI research results
- Projects proposed by PCORI-funded investigators to prepare for applying (i.e. "bridge funding") to the Limited PCORI Funding Announcement: Dissemination and Implementation

For More Information

Engagement Awards Program

- Web Page: www.PCORI.org/eugene-washington-awards
- Email Address: ea@pcori.org
- Contact Number: 202-370-9312

Pipeline to Proposals Program

- Web Page: http://www.pcori.org/funding-opportunities/programmatic-funding/pipeline-proposal-awards
- Email Address: p2p@pcori.org

Questions?

Thank You

Lia Hotchkiss, MPH

Director, Eugene Washington PCORI Engagement Awards Program

202-494-3441

lhotchkiss@pcori.org

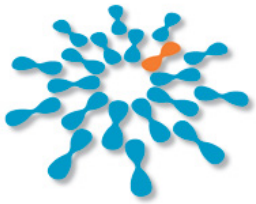
Presentation of Results

Eleanor Perfetto, PhD

Professor, *Pharmaceutical Health Services Research, School of Pharmacy, University of Maryland*

Senior Vice President, *Strategic Initiatives, National Health Council*

PI, *PCOR Training: A Program for Rare Disease Patient Advocates*



NORD
National Organization for Rare Disorders

UNIVERSITY of MARYLAND
SCHOOL OF PHARMACY

PCOR Training: A Program for Rare-Disease Patient Advocates

Highlighting Training Materials and Resources

November 27, 2016

Eleanor M. Perfetto, PhD, MS
University of Maryland, Baltimore
National Health Council

Training Objectives

1. To increase rare-disease community capacity to engage in patient-centered outcomes research (PCOR) through PCOR training specifically designed for this community
2. Disseminate tools developed and evaluation of findings

Team Members

- PCORI Staff
  - Lia Hotchkiss
  - Scott Yoo
- Award Team
  - Dr. Eleanor M. Perfetto
  - Joella Trenchard
  - Dr. C. Daniel Mullins
  - Elisabeth Oehrlein
  - Dr. Chinenye Anyanwu
  - Lisa Phelps (NORD)
  - Laverne McCoy
  - Dr. Divya Shridharmurthy
  - Dr. Jasel Martin

Patient Advisory Board

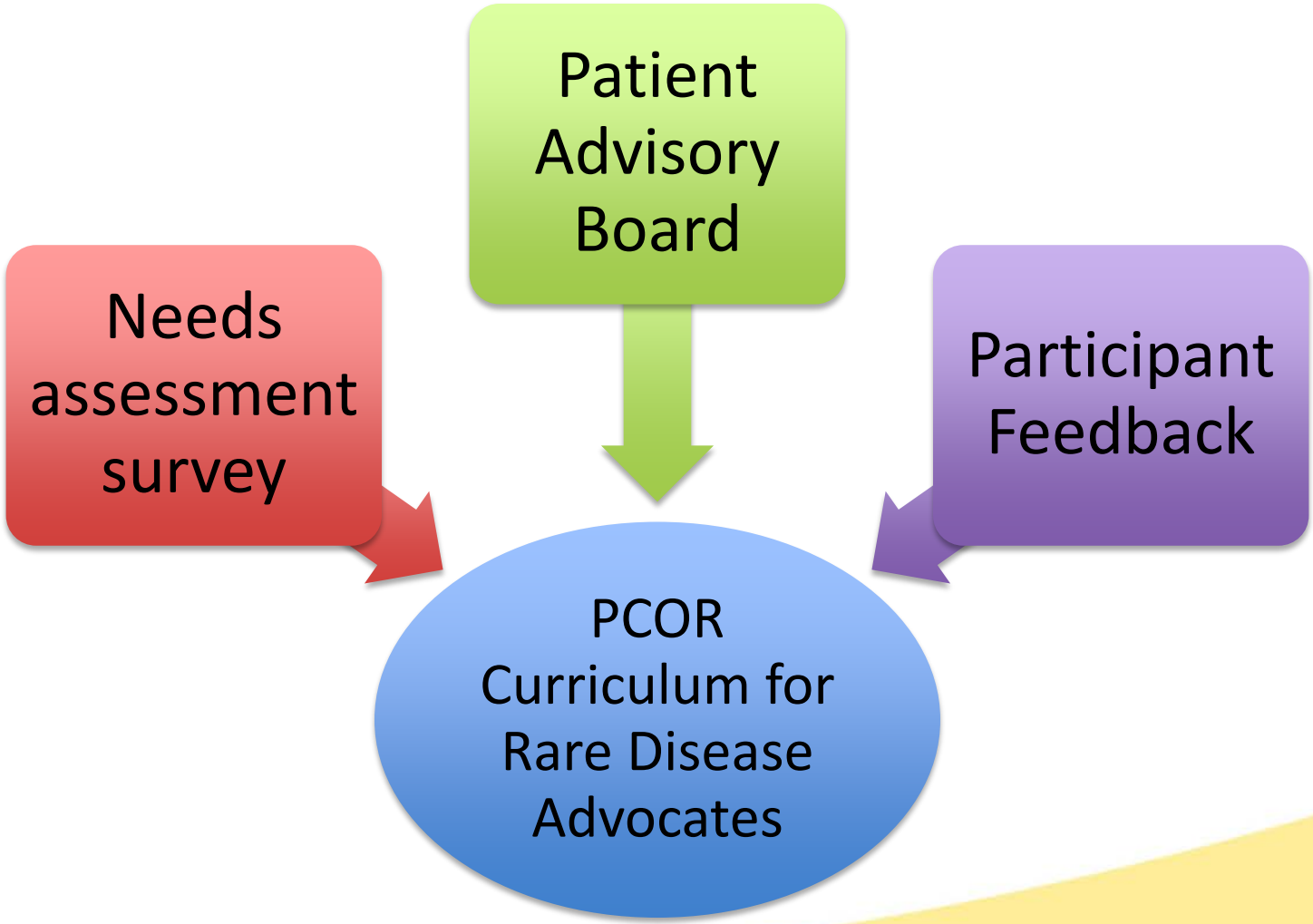
- *Jacqueline Kraska, National Organization for Rare Disorders (Co-chair)*
- *C. Daniel Mullins, PhD, University of Maryland School of Pharmacy (Co-chair)*
- *Ron Bartek, Friedreich's Ataxia Research Alliance (FARA)*
- *Marc Boutin, JD, National Health Council*
- *Christine Brown, National PKU Alliance*
- *Laurie Burke, The Lora Group*
- *Mary Dunkle, National Organization for Rare Disorders*
- *Rachel Gomel, PSC Partners Seeking a Cure*
- *Reta Honey Hiers, R.N.C., Tarlov Cyst Disease Foundation*
- *Jack Kelly, Lymphangiomatosis & Gorham's Disease Alliance*
- *Lisa Phelps, National Organization for Rare Disorders*
- *Christopher Scalchunes, Immune Deficiency Foundation (IDF)*

Curriculum and Tools on PCORI Website

- Curriculum
  - Day 1 - PCOR 101 Half-day session
  - Day 2 - PCOR 201 Full-day session
- Resources
  - Acronym list
  - Glossary
  - Successful partnerships checklist
  - PCOR for rare disease resource list
- Post-training Research Club Teleconferences (3)
  - Assignments including pre-reading assignments

http://www.pcori.org/research-results/2015/pcor-training-program-rare-disease-patient-advocates

Curriculum and Tools Development



Needs Assessment Survey Results

How familiar are you with patient-centered outcomes research?

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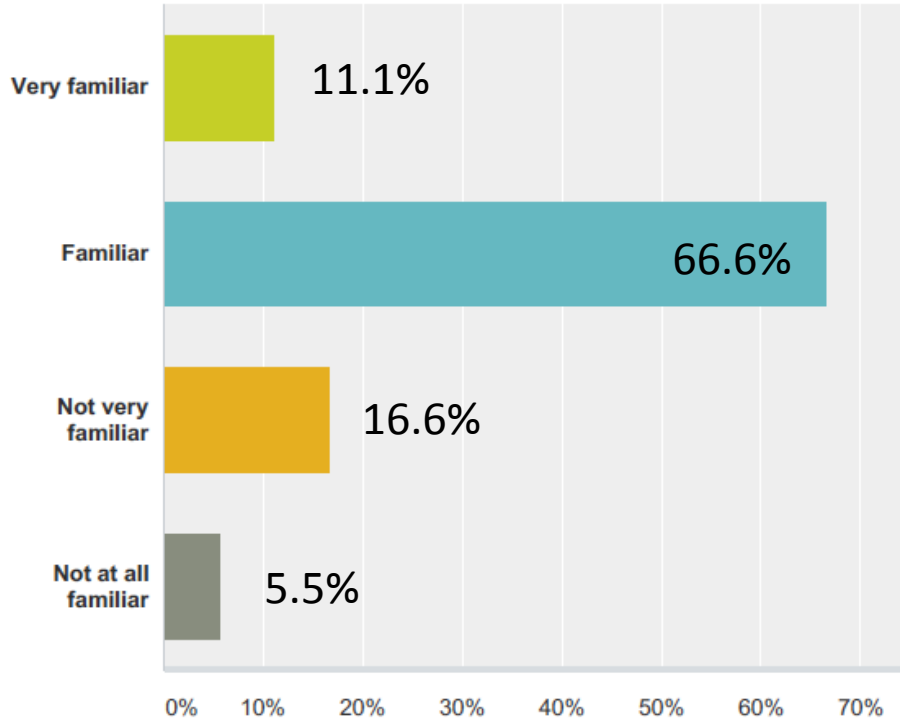
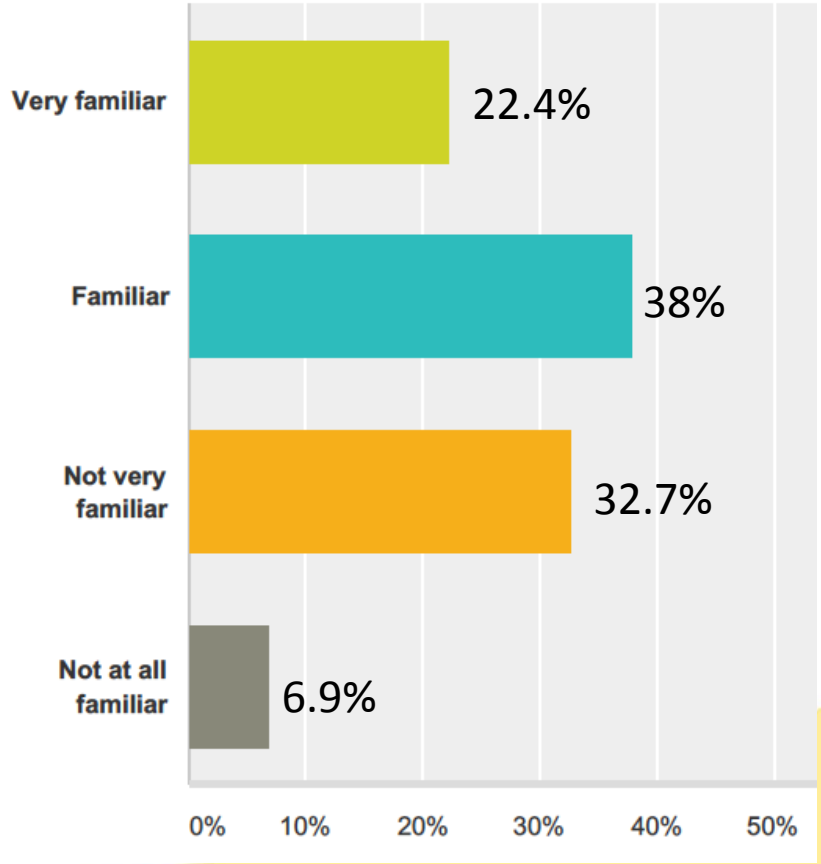


2015

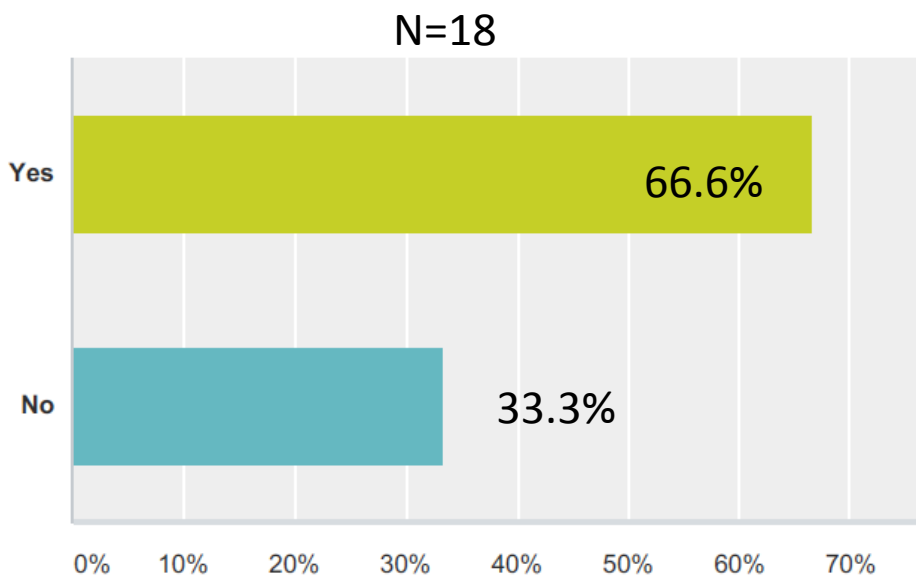
N=58



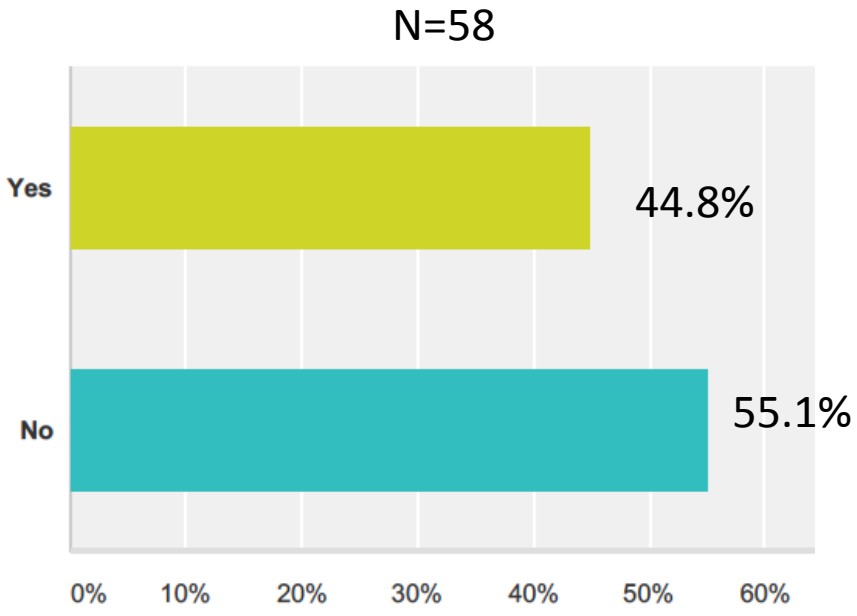
2016

Needs Assessment Survey Results

Has your organization interacted with the Patient-Centered Outcomes Research Institute (PCORI)?



2015



2016

Day 1- ‘PCOR 101’ Participant Objectives

At the end of the half-day track, participants will be able to:

- Act as PCOR ambassadors for the rare-disease community
- Describe the role patients/patient organizations play in PCOR
- Clearly convey the importance of including the patient voice in research

- Describe strategies for partnering with PCOR stakeholders, including leveraging patient group assets
- List and describe funding sources for PCOR projects and identify funding opportunities

Day 1 - 'PCOR 101' Half-Day Agenda

- ***Session 1*** *- Patient-Centered Outcomes Research: What it Means to the Rare Disease Community*
- ***Session 2*** *- Reorienting the Research Environment: Advocating for the Value of the Patient's Voice*
- ***Session 3*** *- Partnerships and Funding for PCOR Projects*
- ***Session 4*** *- Patient-Centered Outcomes Research and Policy*

http://www.pcori.org/research-results/2015/pcor-training-program-rare-disease-patient-advocates

Day 2- ‘PCOR 201’ Participant Objectives

At the end of this full-day special session, participants will be able to

- Demonstrate an understanding of patient-engagement methods
- Develop communication and networking skills and strategies to promote partnership
- Identify key features of successful PCOR proposals and projects

Day 2 - 'PCOR 201' Agenda

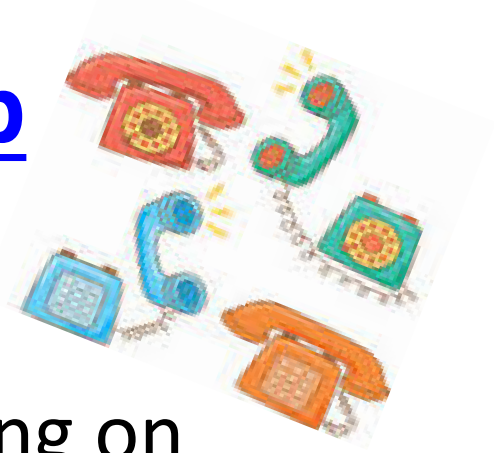
- ***Session 1*** *- The Natural History of Disease: The Trump Card?*
- ***Session 2*** *- The Different Levels of Patient Engagement—Case Examples of Making the Most of Limited Resources*
- ***Session 3*** *- Putting Your Best Foot Forward: Establishing Partnerships with PCOR Stakeholders*
- ***Session 4 -*** *Optimizing Funding Applications (Panel Discussion)*

http://www.pcori.org/research-results/2015/pcor-training-program-rare-disease-patient-advocates

Resources Created

- Acronym list
- Glossary
- Successful partnerships checklist
- PCOR for rare disease resource list

Three Monthly Research Club Teleconferences



- Designed to continue active learning on PCOR, informed by participant feedback
- Discuss new opportunities to engage in PCOR
- Topics Discussed:
  - ✓ PICOTS Framework: How to Write a Research Question
  - ✓ Applying for a Eugene Washington Award
  - ✓ Patient-Reported Outcomes

Lessons Learned

- Patient groups need more support and help with infrastructure than other investigators or co-investigators
- Team effort to overcome barriers
- Importance of an active and engaged advisory group of NORD members
- Champion(s) at all partner organizations

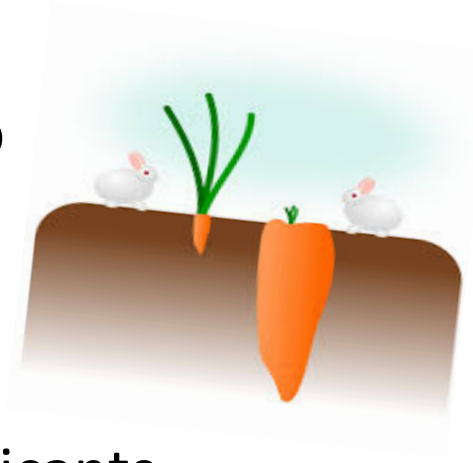
Funded Participants

- Training participants funded as the primary contractor:
  - Cholangiocarcinoma Foundation
  - Platelet Disorder Support Association
- Indirect "participants" funded as the primary contractor:
  - Foundation for Prader-Willi Research

    (Dr. Strong participated in our training as a speaker on her experience working with industry)
  - National Alopecia Areata Foundation

    (Used our materials and Perfetto an advisor to the project)

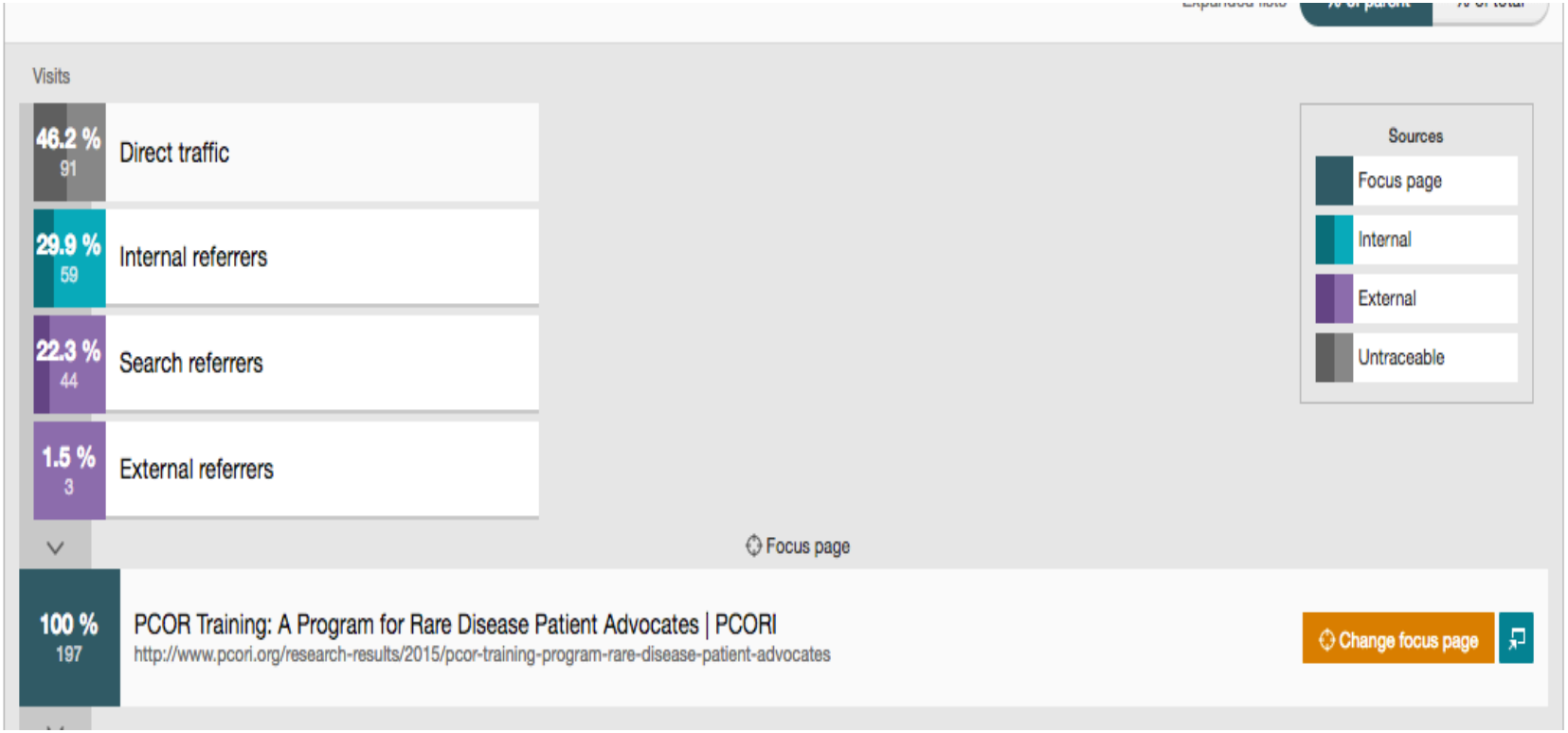
Underestimate of Our Impact?



We are sure this represents an underestimate of the impact of the training:

- The list is for funded projects but not for applicants
  - We don't know how many trainees have submitted proposal as a primary or sub contractor.
- The organizations listed are the primary contractors
  - We don't know how many of our trainees are subcontractors on funded projects.
- We don't know if other applicants or funded projects are using our materials in their proposals/projects
- We have anecdotal information that participants are using their new skills to obtain other, non-PCORI funding.

PCORI Training URL Traffic



59.9 % 118	Visit ended
9.6 % 19	UMD-NORD-Day-One-PCOR-What-It-Means-Rare-Disease-Community http://www.pcori.org/sites/default/files/UMD-NORD-Day-One-PCOR-What-It-Means-Rare-Disease-Community.pdf
6.6 % 13	Patient-Centered Outcomes Research for Rare Disease Patients on Vimeo https://vimeo.com/156000384
6.6 % 13	UMD-NORD-Day-Two-Different-Levels-Patient-Engagement http://www.pcori.org/sites/default/files/UMD-NORD-Day-Two-Different-Levels-Patient-Engagement.pdf
5.1 % 10	UMD-NORD-Day-Two-Natural-History-Disease http://www.pcori.org/sites/default/files/UMD-NORD-Day-Two-Natural-History-Disease.pdf
5.1 % 10	UMD-NORD-Day-One-Agenda http://www.pcori.org/sites/default/files/UMD-NORD-Day-One-Agenda.pdf
5.1 % 10	UMD-NORD-Day-One-Reorienting-Research-Environment http://www.pcori.org/sites/default/files/UMD-NORD-Day-One-Reorienting-Research-Environment.pdf
4.6 % 9	UMD-NORD-Telecon-One-PICOTS-Research-Question http://www.pcori.org/sites/default/files/UMD-NORD-Telecon-One-PICOTS-Research-Question.pdf
4.6 % 9	UMD-NORD-Successful-Parntership-List http://www.pcori.org/sites/default/files/UMD-NORD-Successful-Parntership-List.pdf
4.1 % 8	UMD-NORD-Telecon-Two-Patient-Reported-Outcomes http://www.pcori.org/sites/default/files/UMD-NORD-Telecon-Two-Patient-Reported-Outcomes.pdf
3.6 % 7	UMD-NORD-Day-One-Partnerships-Funding-for-PCOR-Projects http://www.pcori.org/sites/default/files/UMD-NORD-Day-One-Partnerships-Funding-for-PCOR-Projects.pdf
2.5 % 5	Answering the Right Questions on Vimeo https://vimeo.com/171956528
2.5 % 5	UMD-NORD-Glossary http://www.pcori.org/sites/default/files/UMD-NORD-Glossary.pdf
1.5 % 3	UMD-NORD-Telecon-Two-Patient-Reported-Outcomes-Deck http://www.pcori.org/sites/default/files/UMD-NORD-Telecon-Two-Patient-Reported-Outcomes-Deck.pdf
1.5 % 3	Research & Results | PCORI http://www.pcori.org/research-results
1.0 % 2	UMD-NORD-Day-One-PCOR-Policy-Panel-Discussion http://www.pcori.org/sites/default/files/UMD-NORD-Day-One-PCOR-Policy-Panel-Discussion.pdf
1.0 % 2	UMD-NORD-Day-Two-Agenda http://www.pcori.org/sites/default/files/UMD-NORD-Day-Two-Agenda.pdf
1.0 % 2	UMD-NORD-PCOR-Rare-Disease-Resource-List http://www.pcori.org/sites/default/files/UMD-NORD-PCOR-Rare-Disease-Resource-List.pdf
1.0 % 2	About Us | PCORI http://www.pcori.org/about-us
1.0 % 2	http://rarediseases.org/event/pcor-training-for-nord-members/
1.0 % 2	UMD-NORD-Day-Two-Putting-Your-Best-Foot-Forward http://www.pcori.org/sites/default/files/UMD-NORD-Day-Two-Putting-Your-Best-Foot-Forward.pdf
1.0 % 2	UMD-NORD-Day-Two-Optimizing-Funding-Applications http://www.pcori.org/sites/default/files/UMD-NORD-Day-Two-Optimizing-Funding-Applications.pdf
1.0 %	Funding Opportunities | PCORI

% of URL clicks for each deliverable

Conclusion

Rare disease patient advocates reported that the training provided them with the skills they need to engage in PCOR projects and/or develop their own PCORI submissions. They are more confident in their discussions and relationships with researchers and medical product developers. The resources developed may be useful to other audiences.

Link to training materials and resources

http://www.pcori.org/research-results/2015/pcor-training-program-rare-disease-patient-advocates

Questions?

Thank You!

Eleanor M. Perfetto, PhD, MS

Professor, Pharmaceutical Health Services Research,
University of Maryland, Baltimore (UMB)
School of Pharmacy

RDAP Discussion

- How should these materials be disseminated?
- Do you find these materials useful?
- Is there anything missing from this training?

COI Form Updates

Jayne Jordan

Special Assistant, General Counsel

Lunch

We will resume at 1:00 PM ET

PCORnet Rare Disease Patient Powered Research Networks (PPRNs)

Sharon Terry, MA

President and CEO, *Genetic Alliance*

Co-PI, *PCORnet Coordinating Center*

PI, *Community-Engaged Network for All (CENA)*

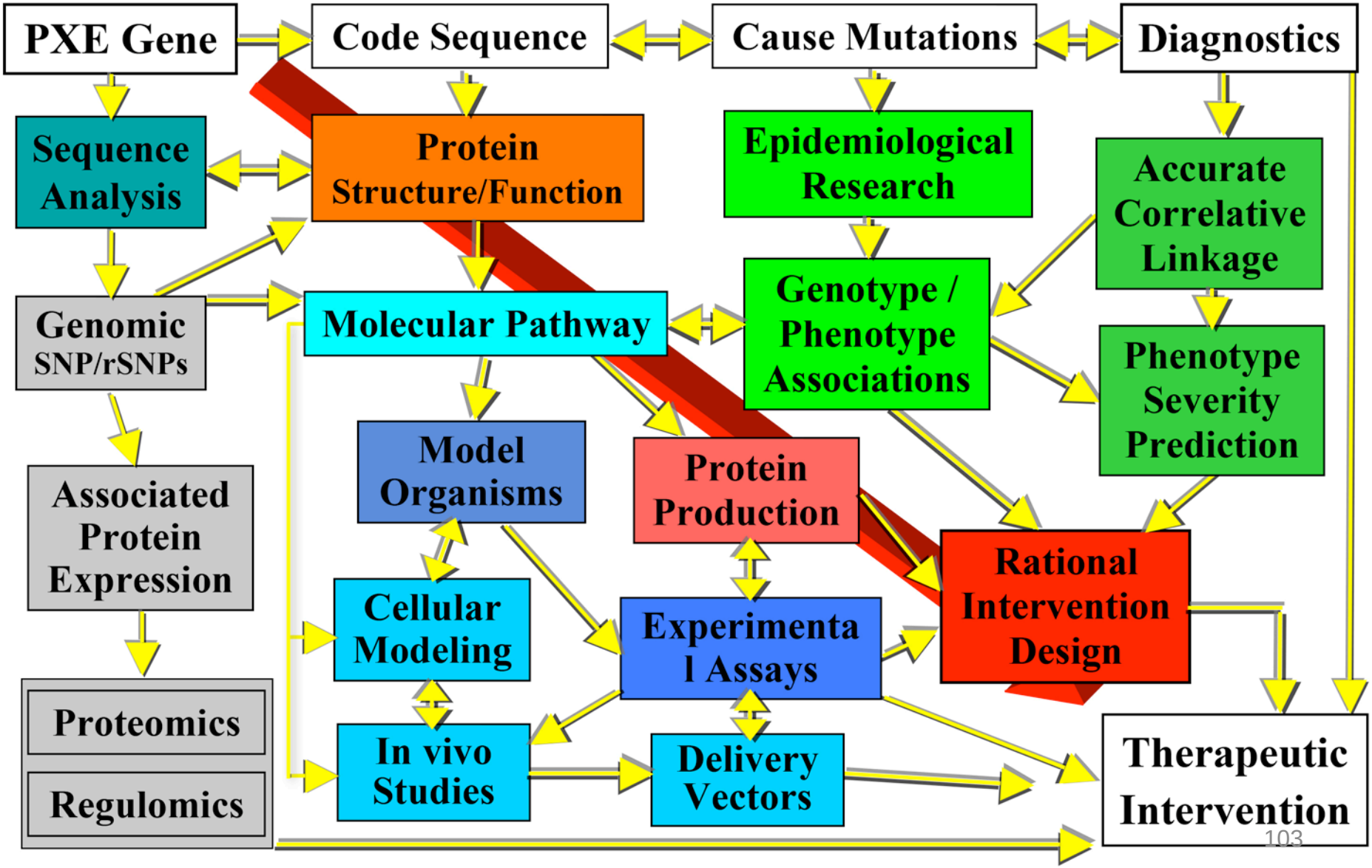
PCORnet People-Powered Outcomes Research

Adrian Hernandez, Rich Platt, and Sharon Terry, Co-principal Investigators, PCORnet Coordinating Center

pcornet® | The National Patient-Centered Clinical Research Network

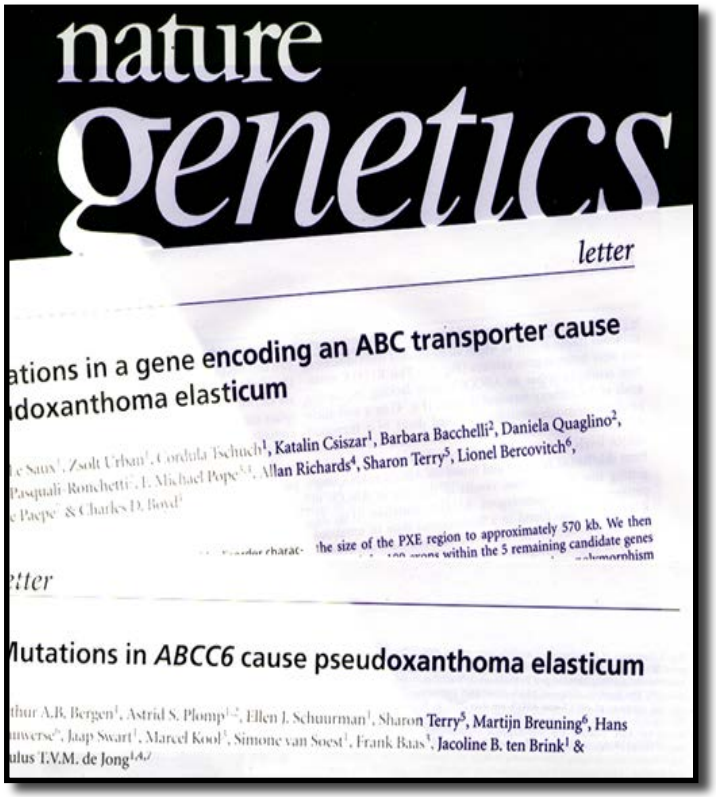
Elizabeth and Ian diagnosed with pseudoxanthoma elasticum (PXE) 1994

PXE International Strategy

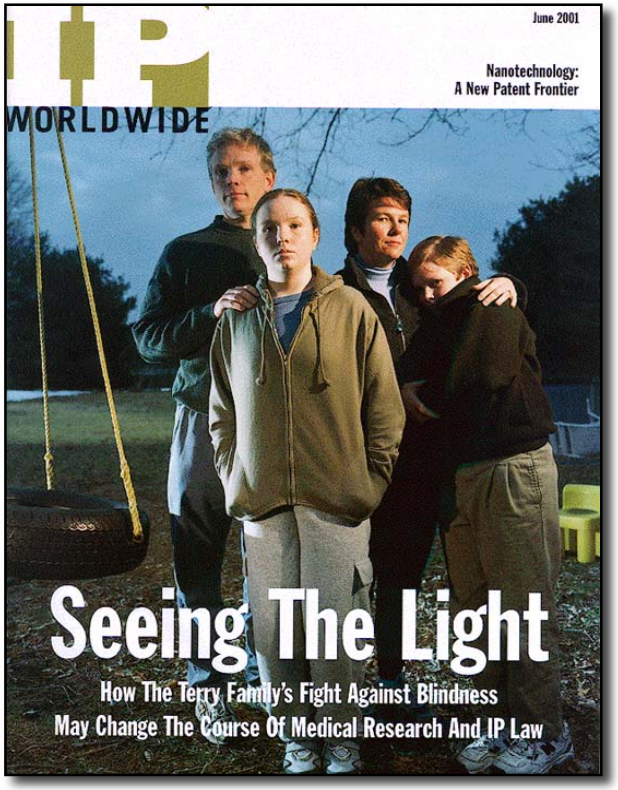


PXE international

Gene Discovery



BioBank



Testing

Clinical Diagnostic Test Development via FDA & CLIA Regulatory Strategies

Human Clinical Trials

Drug Screening & Development Approaches

Therapeutics

--Small Molecules

--Nonsense mutants

Patenting

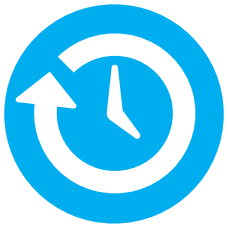
Licensing & Intellectual Property Management

Our national clinical research system is well-intentioned but flawed

We are not generating the evidence we need to support the healthcare decisions that patients and their doctors have to make every day.

- High percentage of decisions are not supported by evidence
- Health outcomes and disparities are not improving
- Current clinical research system faces several problems:

Doesn't answer questions that matter most to people



Too slow

Too expensive

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We can improve

What if we could have at our fingertips **trustworthy, high-quality data** from health systems, people and partnerships to **bring people the real-world answers they seek?**

What if we could **decrease the time it takes to get clinical insights?**

What if we could achieve **significant cost savings** over a traditional clinical study?

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PCORnet: the National Patient-Centered Clinical Research Network

PCORnet is **a large, highly representative, national patient-centered clinical research network**.

Our **vision** is to support a learning U.S. healthcare system and to enable **large-scale clinical research** conducted with **enhanced quality and efficiency**.

Our **mission** is to enable faster, more trustworthy clinical research that **helps people make informed health decisions**.

With PCORnet, we have developed a nationwide functional research network that…

- **Engages** people, clinicians, and health system leaders throughout
- **Creates** infrastructure, tools, and policies to support rapid, efficient clinical research
- **Utilizes** multiple data sources including electronic health records, insurance claims data, data reported directly by people, and other data sources

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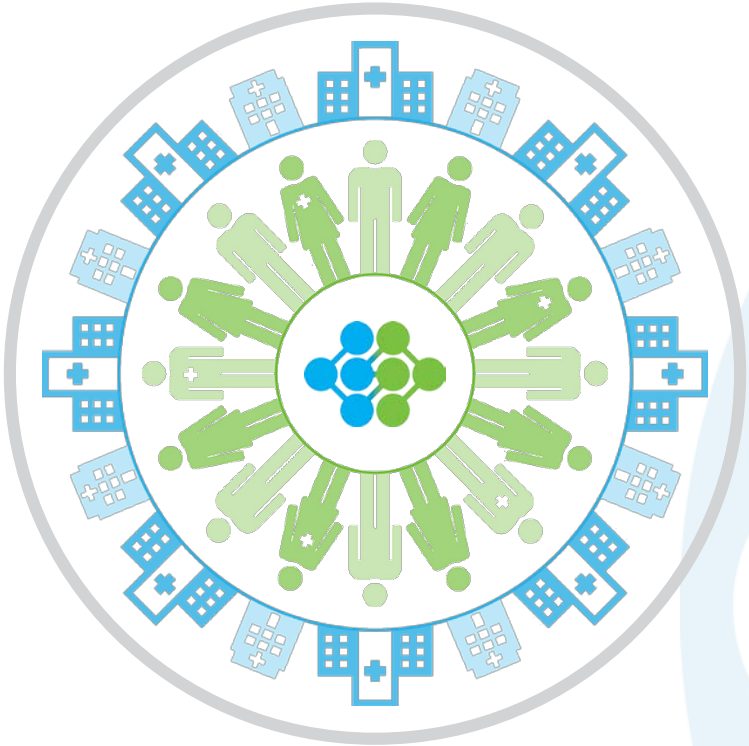
PCORnet embodies a “community of research” by uniting people, clinicians & systems



20
Patient-Powered Research Networks (**PPRNs**)

+

13
Clinical Data Research Networks (**CDRNs**)

=

PCORnet
A national infrastructure for people-centered clinical research

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PPRNs

American BRCA Outcomes and Utilization of Testing Patient-Powered Research Network (ABOUT Network)
University of South Florida

ARthritis patient Partnership with comparative Effectiveness Researchers (AR-PoWER PPRN)
Global Healthy Living Foundation

CCFA Partners Patient Powered Research Network
Crohn's and Colitis Foundation of America

Collaborative Patient-Centered Rare Epilepsy Network (REN)
Epilepsy Foundation

Community and Patient-Partnered Centers of Excellence for Behavioral Health
University of California Los Angeles

Community-Engaged Network for All (CENA)
Genetic Alliance, Inc.

COPD Patient Powered Research Network
COPD Foundation

DuchenneConnect Registry Network
Parent Project Muscular Dystrophy

Health eHeart Alliance
University of California, San Francisco (UCSF)

ImproveCareNow: A Learning Health System for Children with Crohn's Disease and Ulcerative Colitis
Cincinnati Children's Hospital Medical Center

Interactive Autism Network
Kennedy Krieger Institute

Mood Patient-Powered Research Network
Massachusetts General Hospital

Multiple Sclerosis Patient-Powered Research Network
Accelerated Cure Project for Multiple Sclerosis

National Alzheimer's and Dementia Patient and Caregiver-Powered Research Network
Mayo Clinic

NephCure Kidney International
Arbor Research Collaborative for Health

Patients, Advocates and Rheumatology Teams Network for Research and Service (PARTNERS) Consortium
Duke University

Phelan-McDermid Syndrome Data Network
Phelan-McDermid Syndrome Foundation

PI Patient Research Connection: PI-CONNECT
Immune Deficiency Foundation

Population Research in Identity and Disparities for Equality Patient-Powered Research Network (PRIDEnet)
University of California San Francisco

Vasculitis Patient Powered Research Network
University of Pennsylvania

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CDRNs

[Accelerating Data Value Across a National Community Health Center Network (ADVANCE)]
Oregon Community Health Information Network (OCHIN)

[Chicago Area Patient Centered Outcomes Research Network (CAPriCORN)]
The Chicago Community Trust

[Greater Plains Collaborative (GPC)]
University of Kansas Medical Center

[Kaiser Permanente & Strategic Partners Patient Outcomes Research To Advance Learning (PORTAL) Network]
Kaiser Foundation Research Institute

[Research Action for Health Network (REACHnet)]
Louisiana Public Health Institute (LPHI)

[Mid-South CDRN]
Vanderbilt University

[National PEDSnet: A Pediatric Learning Health System]
The Children's Hospital of Philadelphia

[New York City Clinical Data Research Network (NYC-CDRN)]
Weill Medical College of Cornell University

[OneFlorida Clinical Data Research Network]
University of Florida

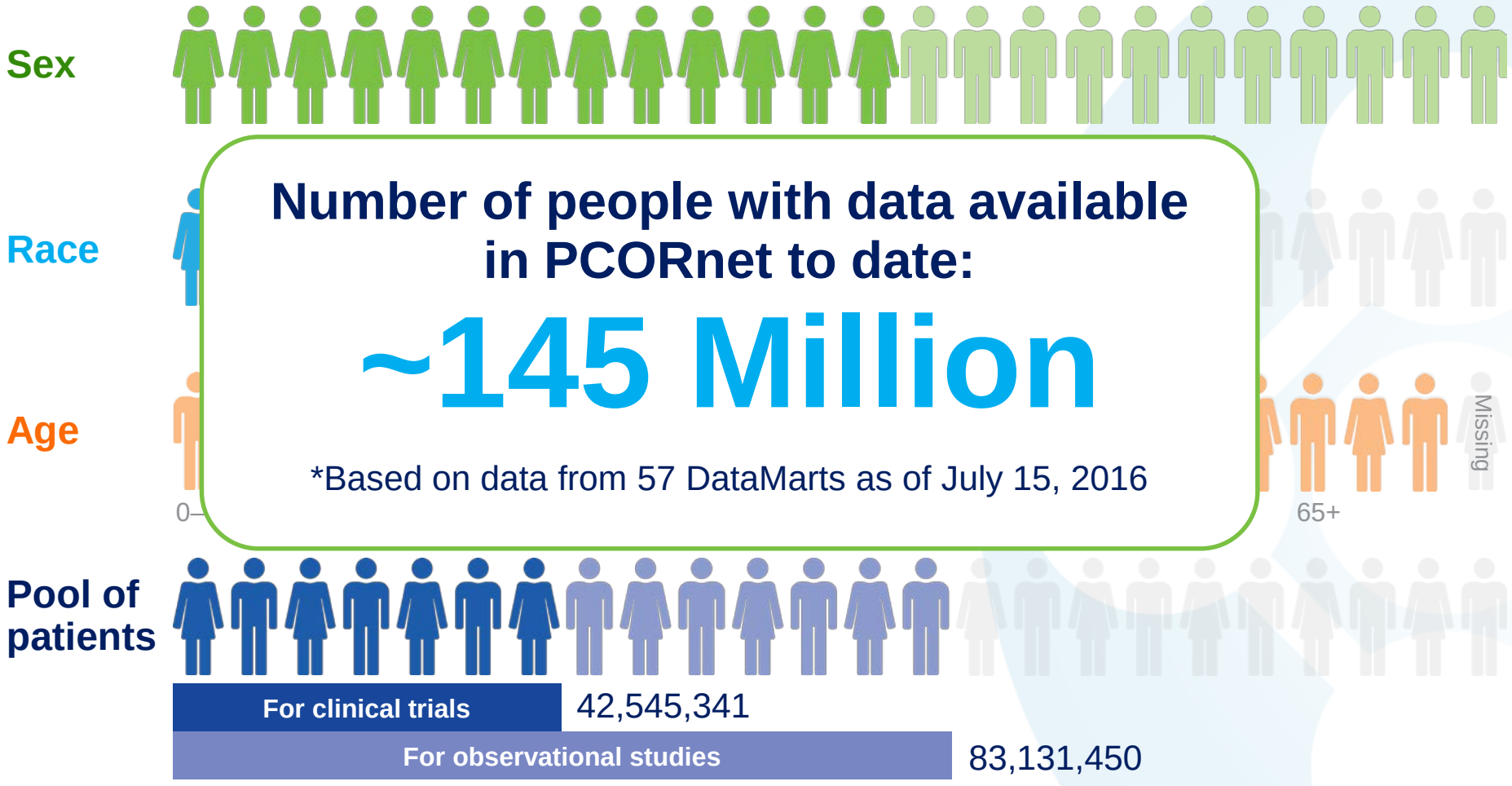
[Patient-Centered Network of Learning Health Systems (LHSNet)]
Mayo Clinic

[Patient-oriented SCAlable National Network for Effectiveness Research (pSCANNER)]
University of California, San Diego (UCSD)

[PaTH: Towards a Learning Health System]
University of Pittsburgh

[Scalable Collaborative Infrastructure for a Learning Healthcare System (SCILHS)]
Harvard University

Resulting in a national evidence system with unparalleled research readiness



Number of people with data available in PCORnet to date:

~145 Million

*Based on data from 57 DataMarts as of July 15, 2016

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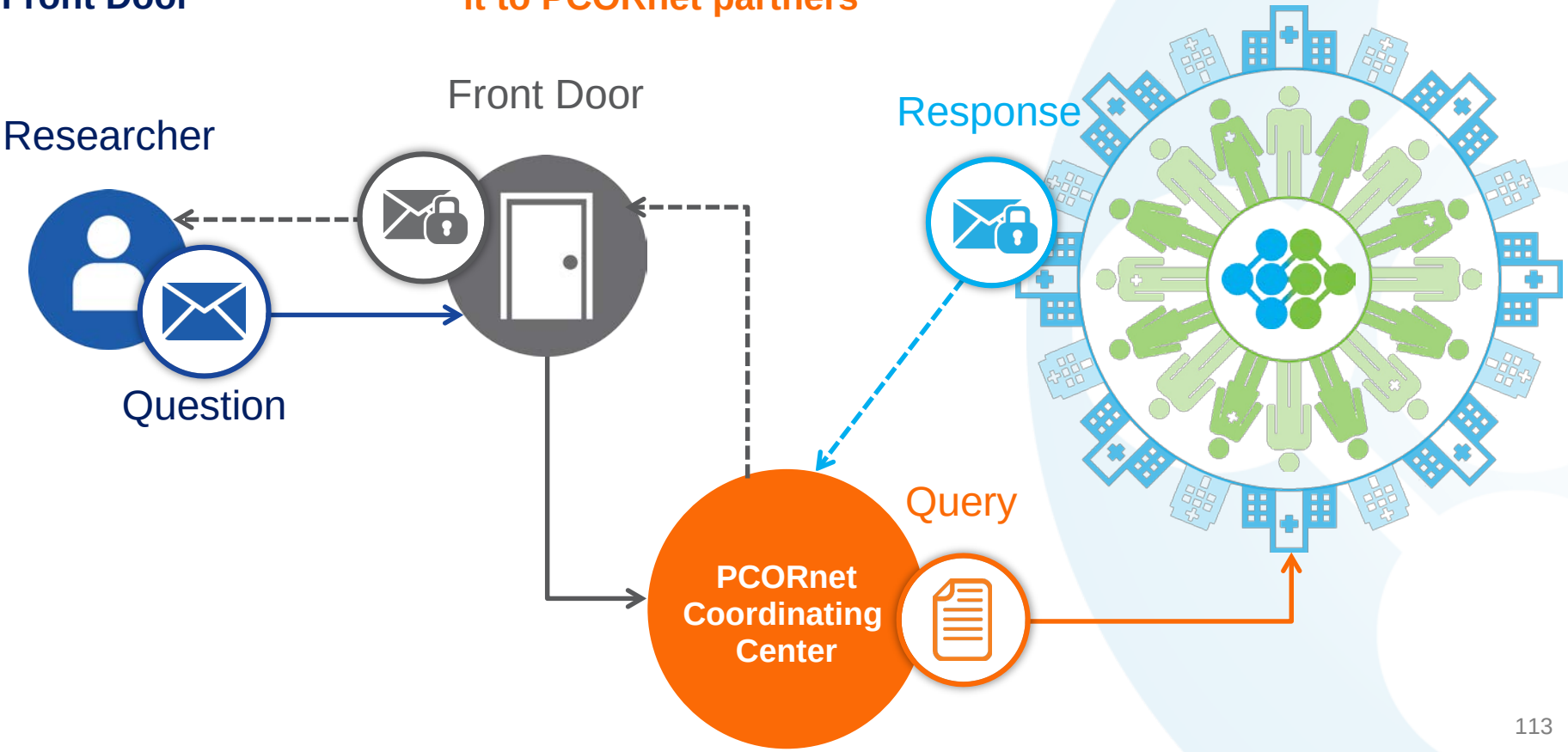
Here's how PCORnet's distributed research network works

The Researcher sends a question to the PCORnet Coordinating Center through the Front Door

The Coordinating Center converts the question into a query with an underlying executable code, and sends it to PCORnet partners

PCORnet partners review the query and provide a response, which is sent back through the Front Door to the Researcher



CDRNs Disease Cohorts

Network	Common Cohort	Rare Cohort
ADVANCE	Diabetes	Alpha 1 Antitrypsin deficiency
CAPriCORN	Anemia; Asthma	Sickle cell disease; Recurrent C. Difficile colitis
Great Plains Collaborative	Breast Cancer	Amyotrophic Lateral Sclerosis (ALS)
LHSnet	Heart Failure	Osteogenesis Imperfecta
Mid-South CDRN	Coronary Heart Disease (CHD)	Sickle Cell Disease (SCD)
NYC-CDRN	Diabetes	Cystic fibrosis
OneFlorida CDRN	Hypertension	Duchenne muscular dystrophy
PEDSnet	Inflammatory bowel disease	Hypoplastic left heart syndrome
PORTAL	Colorectal Cancer	Severe Congenital Heart Disease
pSCANNER	Congestive Heart Failure	Kawasaki Disease
PATH	Atrial Fibrillation	Idiopathic Pulmonary Fibrosis

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CDRNs Disease Cohorts

Network	Common Cohort	Rare Cohort
REACHnet	Diabetes	Sickle Cell Disease (SCD); rare cancers, including: cancers of lip, tongue, salivary glands, gum, mouth; stomach cancers; cancers of the peritonium and retroperitoneum; thymus cancer; cancers of bone and articular cartilage; cancer of body of uterus; cancer of ovary and other uterine adnexa; cancer of other female genital organs; cancer of testis and other male genital organs; cancer of endocrine glands and related structures (excluding ovary, testis, and thymus)
SCILHS	Osteoarthritis	Pulmonary arterial hypertension

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PPRNs Conditions

Network	PI	Condition	Proposed PPRN Population Size
ABOUT Network	Rebecca Sutphen	Hereditary Breast and Ovarian Cancer	100,000
AD PCPRN	Ronald Peterson	Alzheimer's Disease & Dementia	
AR-PoWER PPRN	Seth Ginsberg	Arthritis (rheumatoid arthritis, spondyloarthritis, osteoarthritis), musculoskeletal disorders (osteoporosis), and inflammatory conditions (psoriasis)	50,000
CCFA Partners PPRN	Michael Kappelman	Inflammatory bowel diseases (Crohn's disease and ulcerative colitis)	30,000
CENA	Sharon Terry	Allergies, Alström syndrome, arylsulfatase A deficiency, asthma, Austin disease, breast cancer, celiac disease, Cogan-type oculomotor apraxia, Dekaban-Arima syndrome, dyskeratosis congenital, familial pulmonary fibrosis, fibrolamellar hepatocellular carcinoma, Gaucher disease, hepatitis, inflammatory breast cancer, Jacobs syndrome, Joubert syndrome, juvenile nephronophthisis, Klinefelter syndrome, Meckel-Gruber syndrome, metachromatic leukodystrophy (MLD), mucosulfatidosis, multiple sulfatase deficiency, pseudoxanthoma elasticum (PXE), saposin B deficiency, Senior-Loken syndrome, sex chromosome aneuploidy, telomere biology disorder/syndrome, trisomy X, Varadi-Papp syndrome, X and Y chromosome variations.	50-50,000

PPRNs Conditions

Network	PI	Condition	Proposed PPRN Population Size
COPD PPRN	David Mannino	Chronic Obstructive Pulmonary Disease (COPD), Bronchiectasis, Chronic Bronchitis, Emphysema, refractory (non-reversible) asthma	50,000
CPPRN	Kenneth Wells	Behavioral health in under-resourced communities	
DuchenneConnect	Ann Lucas	Duchenne and Becker muscular dystrophy	4,000
Health eHeart Aliance	Mark Pletcher	Hypertension, Congestive Heart Failure, Atrial Fibrillation, Coronary Heart Disease, High Cholesterol/ Statin Eligibility, Cardiac Arrest Survivors and Congenital Heart Disease	100,000
ImproveCareNow	Peter Margolis	Pediatric Inflammatory Bowel Disease, Crohn's disease, ulcerative colitis	15,000
Interactive Autism Network	Kiely Law	Autism spectrum disorder (ASD)	
MoodNetwork	Andrew Nierenberg	Major Depressive Disorder (MDD) and Bipolar Disorder (BP), mood disorders	50,000
MS-PPRN	Robert McBurney	Multiple Sclerosis	20,000
NephCure Kidney Network	Elizabeth Cope	Primary Nephrotic Syndrome (Focal Segmental Glomerulosclerosis [FSGS], Minimal Change Disease [MCD], and Membranous Nephropathy [MN])	

PPRNs Conditions

Network	PI	Condition	Proposed PPRN Population Size
PARTNERS	Laura Schanberg	Pediatric Rheumatic Disease	9,000
PI-CONNECT	Kathleen Sullivan	Primary Immunodeficiency Diseases	1,250
PMS_DN	Megan O-Boyle	Phelan-McDermid Syndrome	737
PRIDEnet	Mitchell Lunn	Health and healthcare disparities for Sexual and gender minorities (SGMs)	
REN	Jan Buelow	Rare epilepsies/epileptic encephalopathy	1,500
Vasculitis PPRN	Peter Merkel	Vasculitis including, Behçet's disease, Central nervous system vasculitis, Cryoglobulinemic vasculitis, Eosinophilic granulomatosis with polyangiitis (Churg Strauss syndrome), Giant cell arteritis, Granulomatosis with polyangiitis (Wegener's), Henoch-Schonlein purpura (IgA vasculitis), Microscopic polyangiitis, Polyarteritis nodosa, Takayasu's arteritis, Urticarial vasculitis, other types of vasculitis	500

Patient/People-Powered Research Networks

250,000 people

More than 100 disease, many rare

pcornet®


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FAQs
Resources
Contact
Log In
Home
A project of
MASSACHUSETTS GENERAL HOSPITAL
MoodNetwork
Improving Lives and Empowering People with Mood Disorders

Mood PPRN

Phelan-McDermid Syndrome INTERNATIONAL REGISTRY

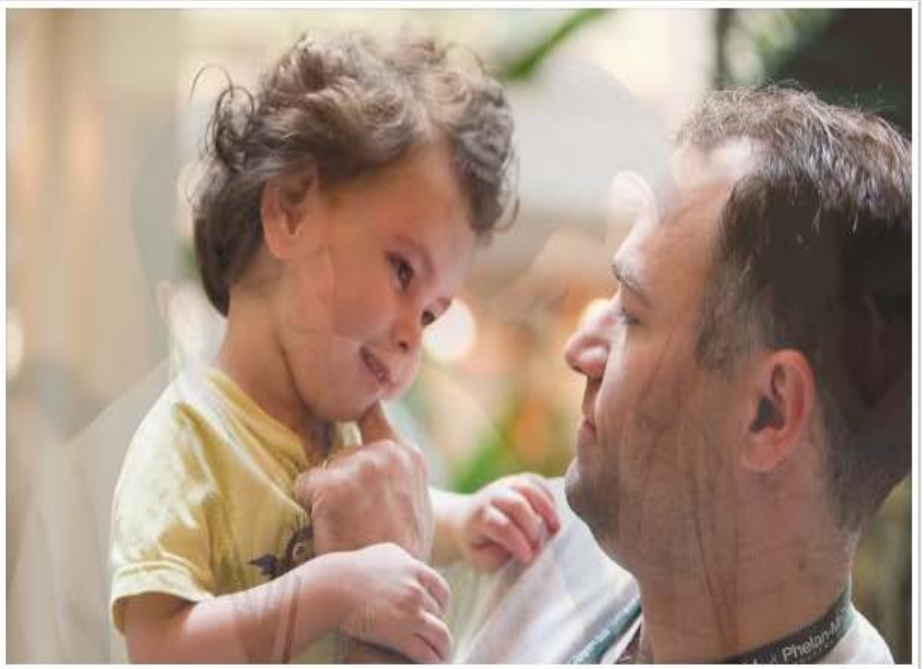
Select English

Username ••••••

Login

Forgot login?

Home | About | Glossary | Clinical Trials | Help



Welcome to the Phelan-McDermid Syndrome International Registry. The purpose of this registry is to consolidate information from individuals with Phelan-McDermid Syndrome into a single database, which will be utilized by researchers to understand Phelan-McDermid Syndrome better.

Click here to register now!

Newsflash

NEW

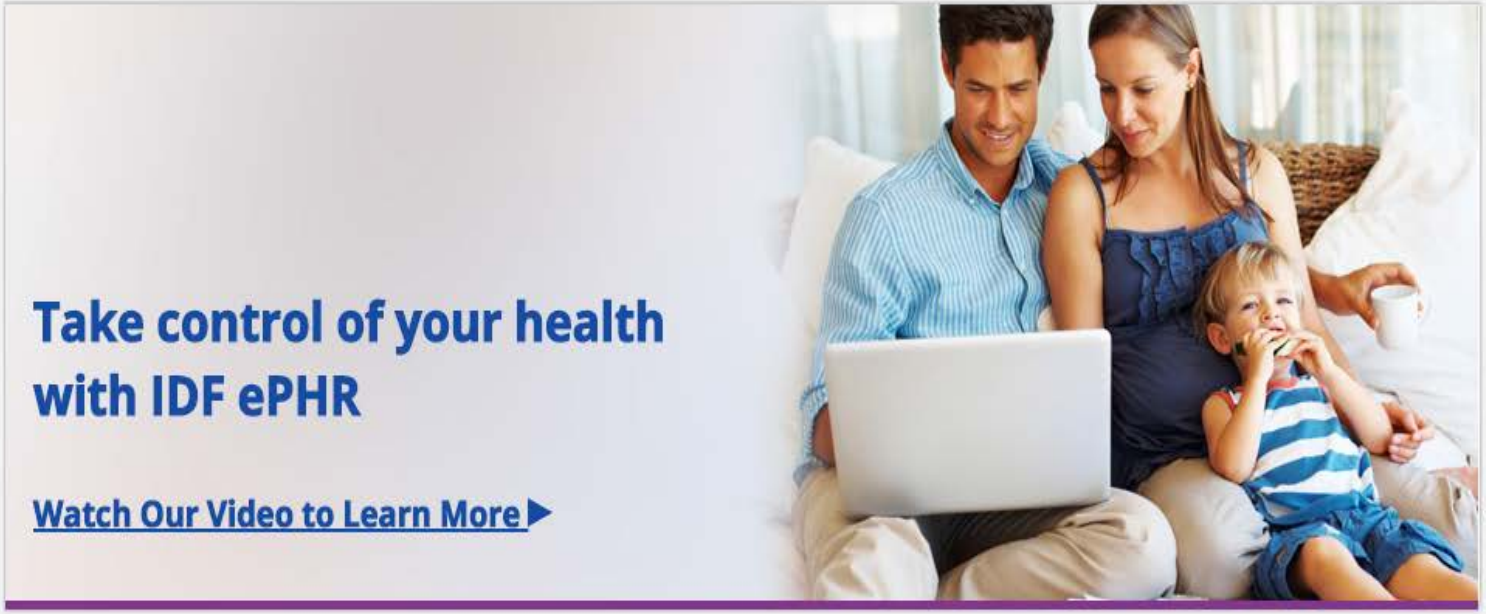
SIGN IN TO RECONSENT

Welcome to the Phelan-McDermid Syndrome International Registry. Please sign in to review and complete the new Informed Consent. There are new data sharing options, including the option to participate in the Phelan-McDermid Syndrome Data Network (PMS_DN). Once you have answered the 8 questions in the informed consent, check "Yes, I agree to participate in the Registry." If you have any questions, please contact the Registry Coordinator at PMSIR@PMSF.org.

ePHR
Immune Deficiency Foundation

The electronic personal health record for the primary immunodeficiency community

Immune Deficiency Foundation



IDF ePHR uses a unique new online system to transform how patients with primary immunodeficiency diseases (PI) manage their health. It offers a place for you to keep your information safe, secure and private at no cost. But it goes so much further!

Through innovative technology, IDF ePHR provides you with easy-to-use, convenient tools to help you live a healthy life. This one central location offers the latest advancements in health record system management.

So, no excuses! It's time to join many other patients and families in the PI community and simplify your life with the new IDF ePHR!

Sign-In | Create Account

Did you create an account before September 1, 2014?
CLICK HERE to reset your password and get started!

If you have already reset your password, click SIGN IN .

SIGN IN

Vasculitis Patient-Powered Research Network

A partnership of the Vasculitis Clinical Research Consortium and the Vasculitis Foundation

Hear from our Co-Principal Investigator, George Casey, about how the V-PPRN will transform vasculitis research.

▶ Play Video

We are patients, researchers, clinicians, advocates, and family members all working to improve healthcare for patients with vasculitis through high-quality clinical research.

Become part of our Research > Join The Network!

CHAT!

The Challenge

The Solution

DuchenneConnect

Home | Register | News | Webinars | About DMD/BMD | Clinical Trials

Translate

Parent Project Muscular Dystrophy

LEADING THE FIGHT TO END DUCHENNE

FUND RESEARCH

DONATE TO DUCHENNECONNECT

User Name

Password

Login

Forgot username / password?

Create account

NEWSFLASH

Please Complete the NEW GI/GU Module

Click here to register now!

Decode Duchenne

A new genetic testing program that allows patients with Duchenne or Becker muscular dystrophy to have access to genetic testing. Decode Duchenne is administered by DuchenneConnect and Parent Project Muscular Dystrophy and is funded by Sarepta Therapeutics.

How Your Data Is Being Used

Have you ever wondered how your data in DuchenneConnect is used? Learn about studies and clinical trials that have utilized the DuchenneConnect data and/or recruited DuchenneConnect registrants.

Login

REN
Rare Epilepsy Network

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Welcome to the Rare Epilepsy Network. **We need your help.**

Solve the Puzzle

CENA: Dyskeratosis Congenita Outreach

Dyskeratosis Congenita Outreach, Inc.

May you never be alone

A Telomere Biology Disorder

Researchers around the world are working hard to piece together the puzzle behind Dyskeratosis Congenita (DC) and related Telomere Biology Disorders (TBD). By sharing our respective stories, we contribute key pieces of information. That's why DC Outreach is sponsoring the first-ever, participant powered DC and TBD registry. By learning how these conditions affect each individual and - with express permission – making it easier for top researchers to contact us about new studies, we can help accelerate our quest to more fully understand DC and in developing better therapies in the future.

You are in control of who gets your data

We use Private Access to give you control over any information you provide through the DCO registry. With Private Access, only you decide who can access your information and for what purpose. Choosing your settings is easy. You don't need to be a computer whiz, and we offer suggestions from knowledgeable guides if you wish.

PRIVACY ASSURED with PrivateAccess

It's quick and simple!

1. Sign Up! (or sign in) — Start Now!
2. Take Health Survey
3. Let Researchers Find YOU!
4. Share this with others!

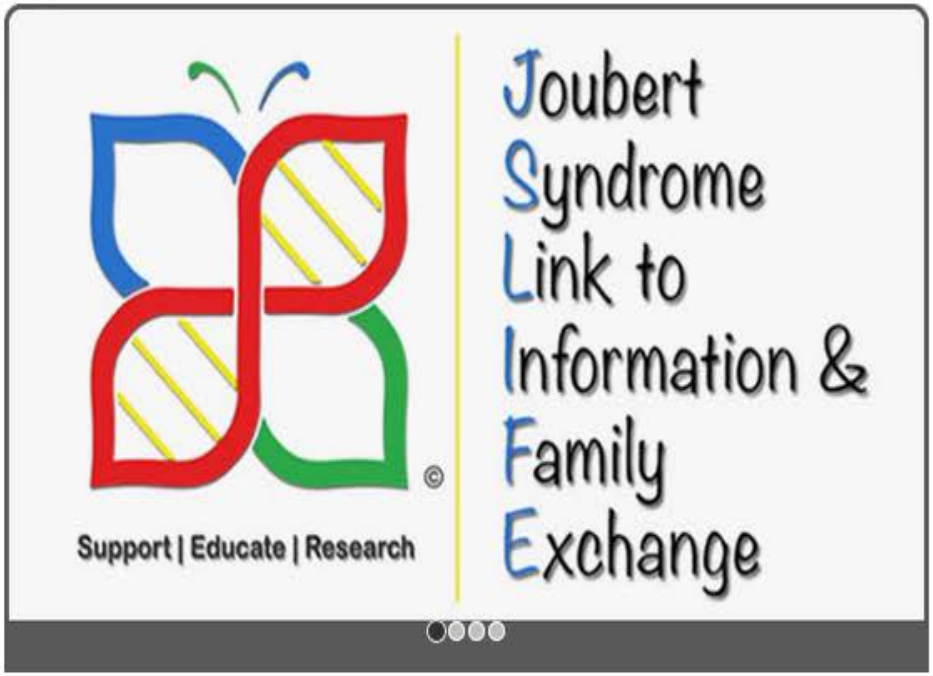
MY DOCTOR OR DISEASE ADVOCACY GROUP RECOMMENDED THIS SERVICE AND PROVIDED ME WITH A REFERRAL CODE:

Enter referral code here | Submit

Privacy Policy Terms of Service Give Feedback

Together we can advance research and improve care

For over 22 years, JSRDF has played an important role in educating physicians and their support teams as well as increasing public awareness about Joubert syndrome. The goal of JS-LIFE (Joubert Syndrome Link to Information & Family Exchange) is to extend this tradition of leadership by enabling access to a much broader research community of clinical, neuroimaging, and genetic studies. This information will help us discover patterns within the data collected and bridge gaps in knowledge among researchers studying Joubert syndrome, other related disorders and ciliopathies. Please join us.



Privacy and Sharing in Perfect Balance

To help us protect your individual privacy in accordance with rules that you establish, we harness the power of Private Access with the specific goal of helping you make your health information available to top researchers - under your own terms.

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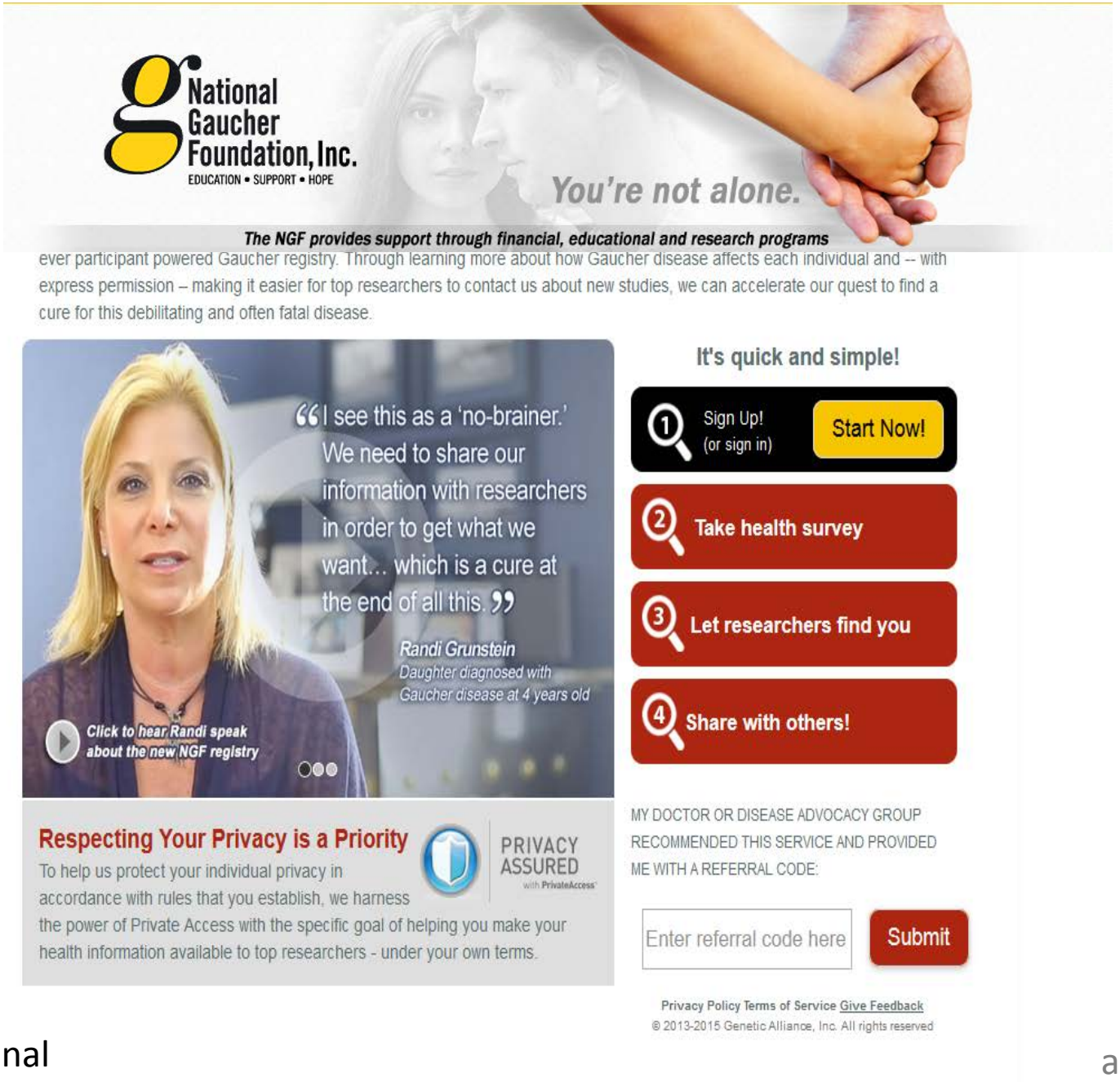

CENA: National Gaucher Foundation

PXE international

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Surveys submitted so far toward the goal of 4,300!

502 | 3798

We do not understand how PXE progresses, or how to slow it down. It is time to change that! Please join our effort to complete the puzzle!

Respecting Your Wishes is Our Priority

To help us protect your individual privacy in accordance with rules that you yourself establish, we harness the power of Private Access with the specific goal of helping you make your health information available to top researchers for your condition - under your own terms.

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Discussion

- How can we connect the dots around data infrastructure for RDAP?

pcornet®

The National Patient-Centered Clinical Research Network

FDA Rare Disease Initiatives

Gayatri Rao, MD, JD

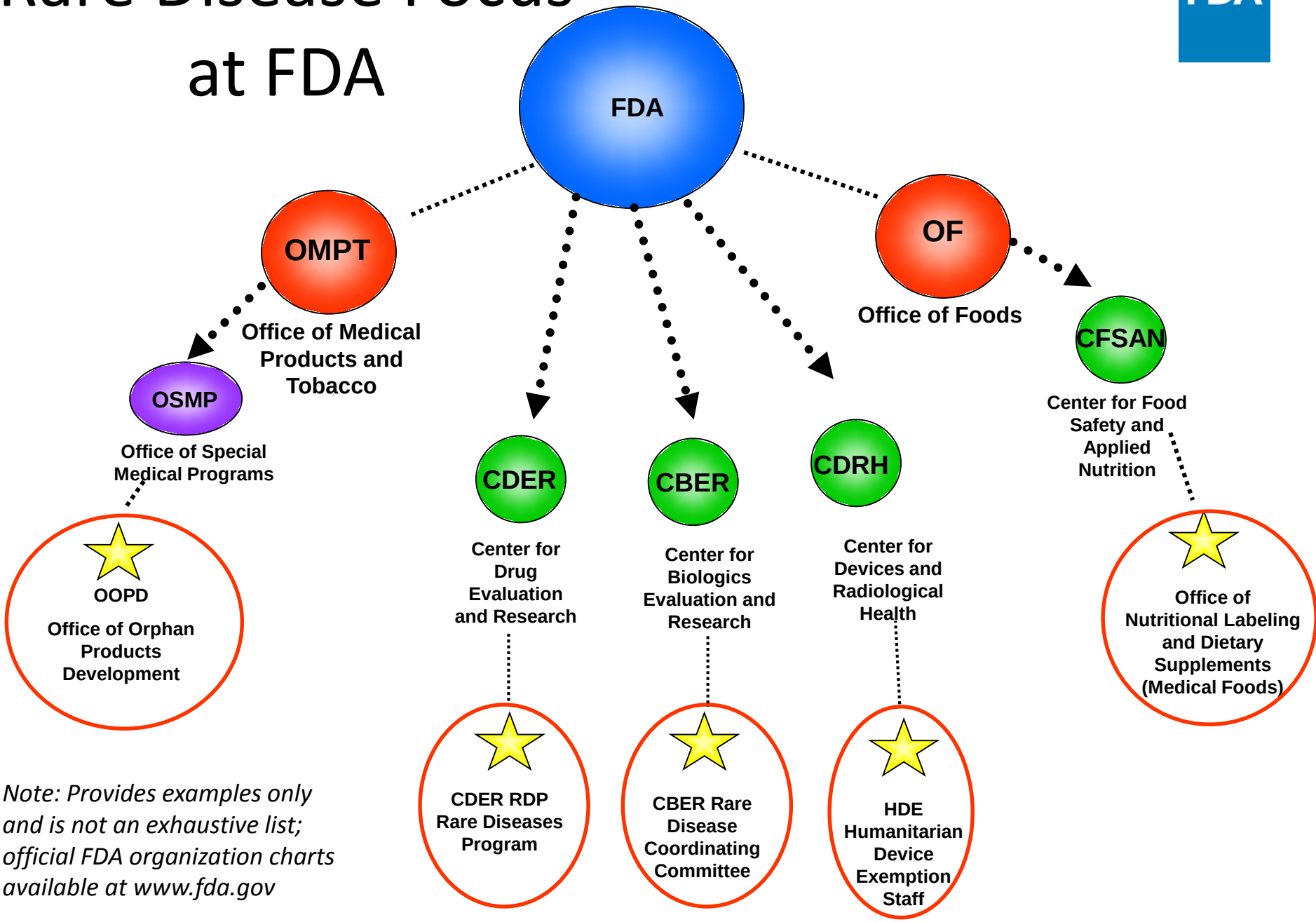
Director, *Orphan Projects Development*

FDA

FDA Rare Disease Activities

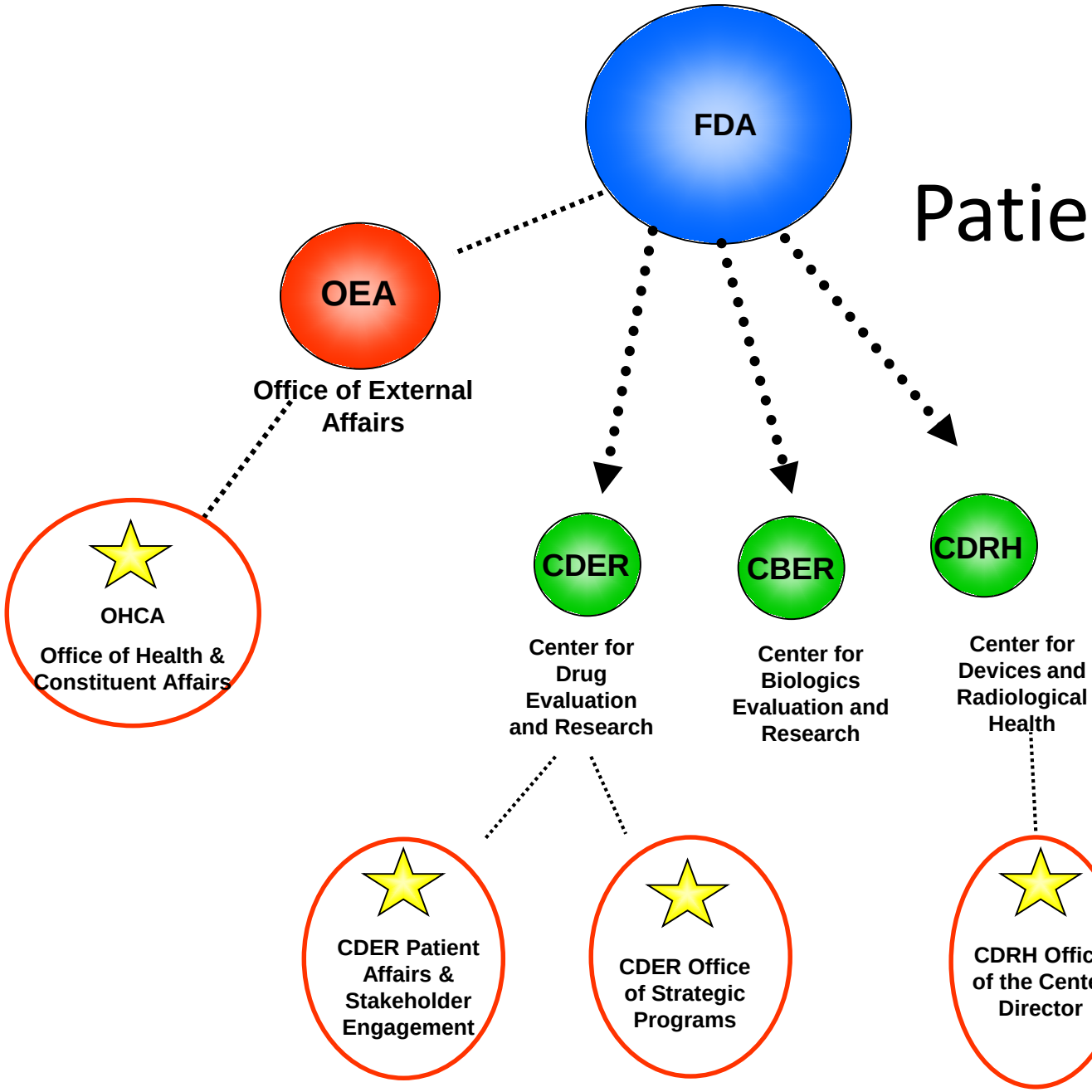
Gayatri R. Rao, MD, JD
Director, Office of Orphan Products Development
PCORI: Advisory Panel on Rare Disease

Rare Disease Focus at FDA



Note: Provides examples only and is not an exhaustive list; official FDA organization charts available at www.fda.gov

Patient Focus at FDA



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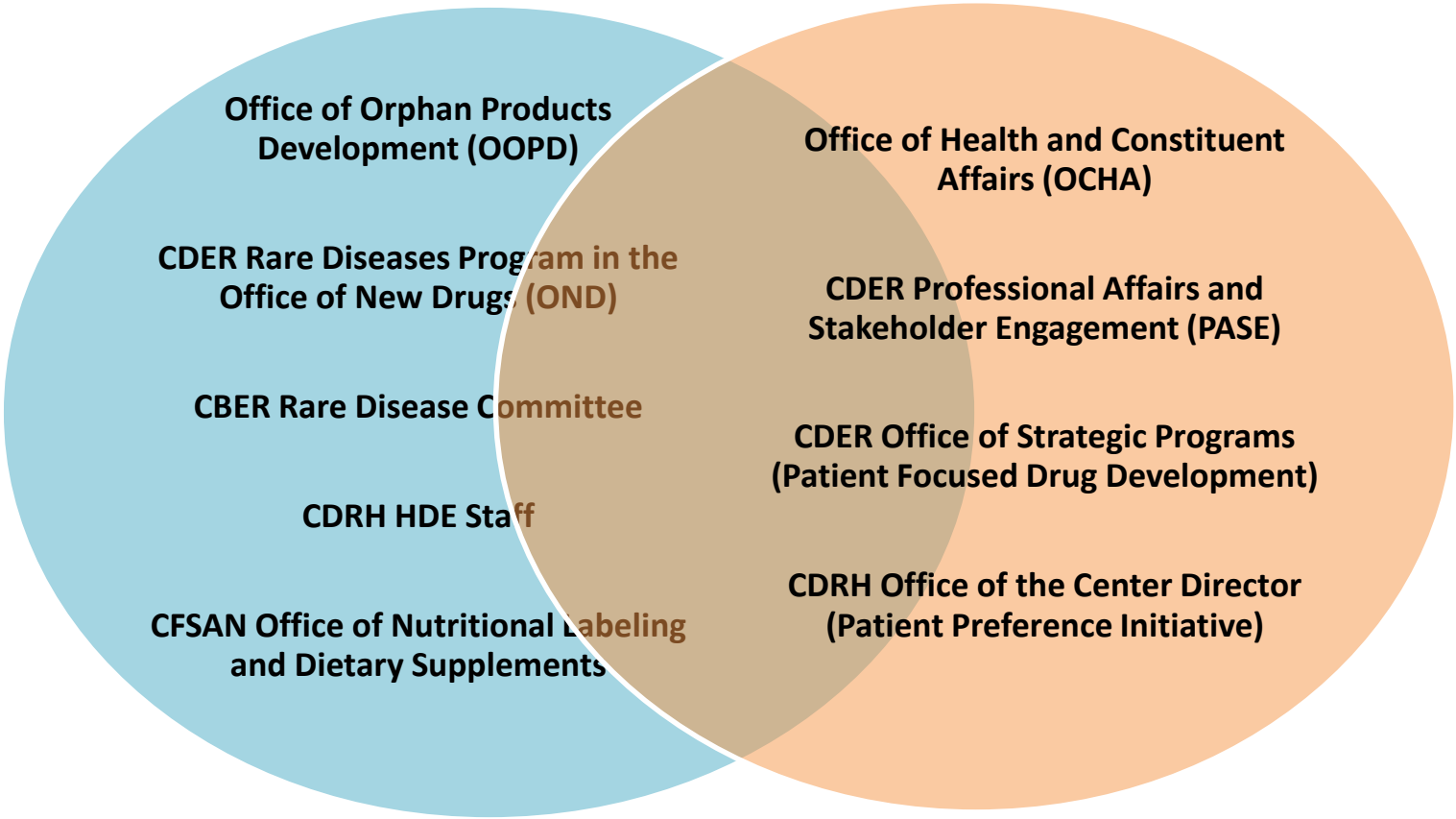
Engagement with Rare Disease Patients & Patient Advocacy Stakeholders

FDA

FDA's Rare Disease Council



FDA's Patient Council

Note: Provides examples only and is not an exhaustive list

Rare Disease Patient Engagement

FDA

Provide information related to, e.g., :
(1) Background on FDA
(2) Product Development
(3) Expanded Access
(4) Drug Shortage
(5) Personal Importation

Provide patient perspectives on product development, e.g.,
(1) Patient Representative Program
(2) Patient Focused Drug Development
(3) Patient Preference Initiative

Patients & Patient Advocacy Groups

The Essential Role of the Patient Voice

- Presence of patients as part of the process of evaluating drugs and medical devices is critical
- Patients have a direct stake in the review process and should have a role in all phases of product development
- They are in a unique position to provide essential insights to help with FDA's mission, including:
    - Learning more about the what it is like to live with a disease or condition;
    - Understanding more about the adequacy of treatment options;
    - Discovering opportunities to develop new and more effective treatments and responses to disease; and
    - Designing a more effective benefit-risk framework to better evaluate the safety and effectiveness of medical products.

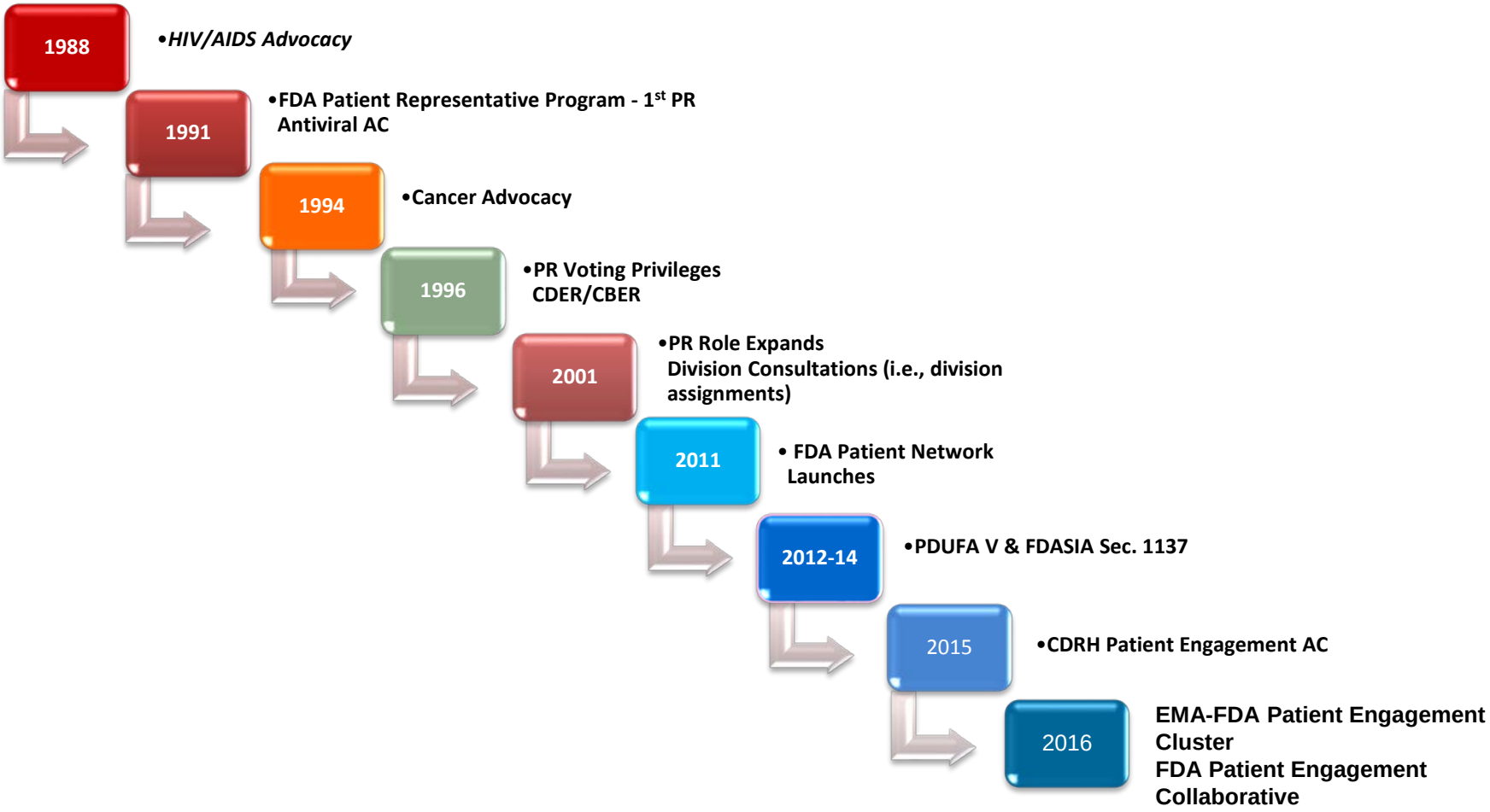
Patient Engagement Can Lead To…

- Faster recruitment and improved retention in trials
- Reducing the time for product development
- Cutting cost of drug development
- Development of meaningful endpoints and measurements
- Outcomes more directly focused on patient needs
- Medical products that better reflect outcome and quality of life measures most important to patients
- Increased reporting of adverse events, post market surveillance, and better treatment management

Challenges to Meaningful Patient Engagement

- Understanding of trial design (meaningful endpoints and data, measuring outcomes, control arms)
- Understanding the regulatory framework, standards, and requirements (level of evidence)
- Legal and practical limitations facing sponsors (promotion v. education and engagement)
- Division within patient communities
- Different objectives or agendas among organizations
- Disagreement on meaningful measurement

FDA Patient Involvement Milestones



FDASIA, Section 1137

- Directs FDA to develop and implement strategies to solicit the views of patients during the medical product development process and consider the perspectives of patients during regulatory discussions, including:
  - Fostering participation of patient representatives in appropriate Agency meetings with medical product sponsors and investigators; and,
  - Exploring ways of identifying Patient Representatives (since they must have little or no financial interest in the medical products industry).

FDA Patient Representative Program

- Patients take an active role on FDA Advisory Committees and consult with agency components
- Over 200 patients and caregivers serve as patient representatives, including patients with rare diseases
  - Go through an application process (with a conflict of interest review) and training process

http://www.fda.gov/ForPatients/About/ucm412709.htm

Patient Focused Drug Development (PFDD)

- Created to more systematically obtain patient perspective on certain diseases and treatments
  - 5 year effort under PDUFA V
  - CDER and CBER commitment to convene at least 20 meetings on specific disease areas, not limited to rare diseases
- Meetings follow a tailored design depending on current state of drug development, needs of the patient population, and specific interests of FDA review divisions

PFDD Meetings

Fiscal Year 2013	Fiscal Year 2014	Fiscal Year 2015	Fiscal Year 2016-2017
• Chronic fatigue syndrome/ myalgic encephalomyelitis • HIV • Lung cancer • **Narcolepsy**	• **Sickle cell disease** • Fibromyalgia • **Pulmonary arterial hypertension** • **Inborn errors of metabolism** • **Hemophilia A, B, and other heritable bleeding disorders** (CBER) • **Idiopathic pulmonary fibrosis**	• Female sexual dysfunction • Breast cancer • **Chagas disease** • Functional gastrointestinal disorders • Parkinson's disease and **Huntington's disease** • **Alpha-1 antitrypsin deficiency** (CBER)	• **Non-tuberculous mycobacterial lung infections** • Psoriasis **To be conducted** • Patients who have received an organ transplant • Neuropathic pain associated with peripheral neuropathy • Alopecia areata • Autism • **Hereditary angioedema**(CBER) • Sarcopenia

***Rare Diseases**

PFDD Outcomes

- **Voice of the Patient** report prepared for each meeting
- Input can support FDA staff to, e.g.,
  - Conduct benefit-risk assessments for products under review
  - Advise drug sponsors on drug development programs
- Input may also support drug development more broadly
  - Identify unmet needs
  - Identify or develop clinical outcome tools (e.g., PROs)

Externally-Led PFDD Meetings

- Many more disease areas than can be addressed by FDA in PFDD meetings (e.g., >7,000 rare diseases)
- FDA welcomes patient organizations to identify and organize patient-focused collaborations to generate public input in other disease areas
  - Recommend using the process established through PFDD as a model
  - FDA open to participation
  - Note, externally-led PFDD meetings and resulting products (e.g., surveys, reports) not considered FDA sponsored/endorsed
- Example: Myotonic Dystrophy, September 15, 2016

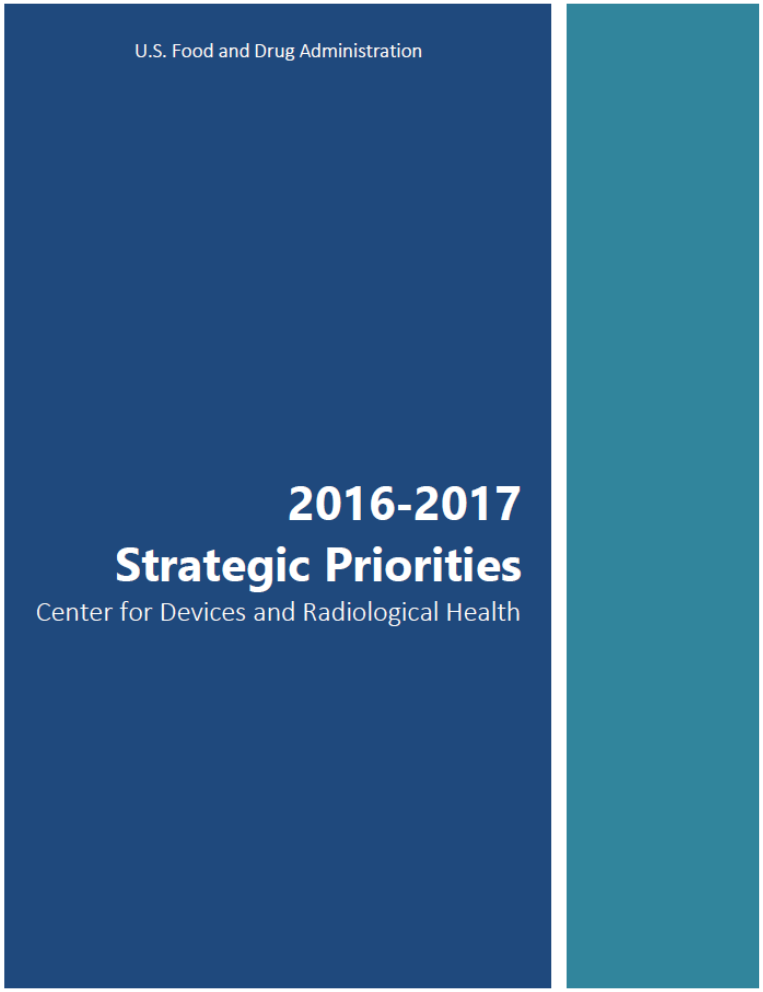
Proposed Recommendations under PDUFDA VI

- Addresses enhancing the incorporation of the Patient's Voice in drug development, including:
  - Guidance development to bridge PFDD meetings, to fit-for-purpose tools to collect meaningful patient for use in regulatory decision making
    - Public workshop to precede guidance development to gather stakeholder input
- Public meeting held on August 15, 2016, to discuss proposed recommendations
- Present proposed recommendations to Congress

For More Information on PFDD:

- FDA website on Externally-Led PFDD Meetings
  - http://www.fda.gov/ForIndustry/UserFees/PrescriptionDrugUserFee/ucm453856.htm
- FDA website on FDA's PFDD Meetings
  - http://www.fda.gov/ForIndustry/UserFees/PrescriptionDrugUserFee/ucm347317.htm
  - The previously conducted meetings include all of the meeting materials, such as agendas, discussion questions.
- Email patientfocused@fda.hhs.gov
  - FDA CDER's Office of Strategic Programs is leading FDA's PFDD effort

CDRH-Patient Engagement



- *"We believe that if CDRH is to successfully achieve a mission and vision in the service of patients, we must interact with patients as partners, and work together to advance the development and evaluation of innovative devices, and monitor the performance of marketed devices."*

CDRH Patient Preference Initiative

- Goal: To develop a systematic way of eliciting, measuring, and incorporating patient preference information, where appropriate, into the medical device Total Product Life Cycle
- Includes:
  - Guidance Development – e.g., Final Guidance on Patient Preference Information
  - Creation of the Patient Engagement Advisory Committee (PEAC)

Guidance on Patient Preference Information (PPI)

- Encourages the submission of PPI, if available, by sponsors or other stakeholders
- Outlines recommended qualities of patient preference studies, which may result in valid scientific evidence
- Provides recommendations for collecting and submitting PPI
- Discusses inclusion of PPI in FDA decision summaries and device labeling

http://www.fda.gov/downloads/MedicalDevices/DeviceRegulationandGuidance/GuidanceDocuments/UCM446680.pdf

CDRH Patient Engagement Advisory Committee (PEAC)

To help assure the needs and experiences of patients are incorporated into our considerations, the PEAC will:

1. Advise CDRH on ways to include and foster participation of patients where appropriate throughout the total product lifecycle
2. Advise CDRH on patient perspectives about current and new approaches or policies for integrating patient input in regulatory decision-making
3. Serve as a resource to CDRH as a body of experts in patient experience, needs, and the activities of the patient community

FDA/EMA Patient Engagement Cluster

- FDA and the European Medicines Agency (EMA) created a new working group on patient engagement in June 2016
- Focus on specific topic areas where the FDA and EMA can benefit from a greater exchange of information and strengthen collaboration, including:
  - Strategies for encouraging sponsors to collect information from patients
  - Comparison of patient/caregiver input between agencies
  - Sharing input with sponsors to create meaningful changes
  - Comparing process for including patient/caregiver input in regulatory decisions
  - Metrics – measuring impact of patient engagement

Patient Engagement Collaborative

(name pending approval)

- Creation of external stakeholder group to provide ongoing counsel about input to and monitoring of patient participation in regulatory processes and policy development across the medical product Centers.
- A forum to exchange information, ideas and experiences on general matters of interest to patients/patient advocates related to patient engagement at FDA.
- FDA's OHCA is leveraging CTTI's steering committee members who are representatives of patient advocacy organizations to help develop the framework for this collaborative

Building a Strong Future of Patient Engagement

- The Process must involve the development of rigorous, objective methodology for measuring patients' experiences and perspectives in a reliable and representative way.
- Can include:
  - development of methods for identifying key impacts and elements of disease experience
  - Translation of those elements in clear, reliable valid measurement tools to capture patient experience in clinical trials
- We are committed to look for new and better ways to integrate the patient voice.

Acknowledgements

OCHA and CDER OSP for sharing perspectives and slides


FDA
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ADMINISTRATION

RDAP Discussion

- How can PCORI work with the FDA to improve the evidence available on treatment and management approaches for rare diseases?
- Are there other organizations that PCORI should engage regarding rare disease funding activities?

Rare Disease Methodology Paper

Danielle Whicher, PhD, MS
Program Officer, Clinical Effectiveness Research

Naomi Aronson, PhD
Ex-Officio Member from PCORI's Methodology Committee

An Overview of the Impact of Rare Disease Characteristics on Research Methodology

Danielle Whicher & Naomi Aronson

October 27, 2016

Overview

- History
- Purpose and Approach
- Paper Overview
- Usefulness / dissemination

History

- RTI landscape review (May 2015)* provided background on current literature discussing registry and research methods for rare diseases
- RDAP members identified several areas where additional information and guidance was needed
  - One identified area: Understanding how features of a rare disease impact decisions about appropriate study designs
- Group members
  - Naomi Aronson (lead)
  - Yaffa Rubenstein
  - Mark Skinner
  - Lisa Heral
  - Jim Wu

*Available at: http://www.pcori.org/sites/default/files/PCORI-Report-Landscape-Review-On-Rare-Disease-May-2015.pdf

Purpose and Approach

- **Purpose**: To raise awareness of the available methodological and analytic approaches relevant to conducting rare disease research
- **Approach**:
  - Reviewed the literature on:
    - Methodological approaches to conducting research on rare diseases
    - How registries and other research infrastructure can facilitate rare disease research
  - Reviewed research methods used in PCORI's rare disease portfolio
  - Requested feedback from RDAP members and PCORI staff

Paper Overview

- Identified and summarized 3 articles that developed algorithms/provided guidance on the relationship between rare disease or intervention characteristics and study design decisions (Appendix A)
- Described study design and analytic approaches mentioned in the literature that might be relevant to addressing research challenges posed by rare diseases (Appendix B)
- Summarized literature describing the utility of existing infrastructure for supporting rare disease research
- Provided an overview of the research methods used by PCORI-funded rare disease projects and PCORnet PPRNs focusing on one or more RDs (Appendix C)
- Suggested areas for further development

Uses for this report

- Promote the use of and challenge investigators to consider a broader range of research methods for RD projects
- Inform investigators of infrastructure to support RD research created through PCORnet
- Inform methodologists of areas where additional methods research and guidance is needed
- Raise awareness among stakeholders, including researchers, payers, patients/patient advocates, and clinicians, of the available methodological and analytic approaches relevant to conducting rare disease research

Discussion

- What next steps are needed?

Panelist Recognition

Thank You!

- We would like to give a special thanks to those members whose terms end this year:
  - Jacqueline Alikhanni
  - Mardi Gomberg-Maitland
  - Philip Ruff

- We would also like to thank Marshall Summar for serving as the chair of the panel since the beginning of the RDAP

Panelist Recognition – Jacqueline Alikhaani

- Volunteer Heart Survivor Spokesperson, American Heart Association – Greater Los Angeles
- Represented: Patients, Caregivers, and Patient Advocates
- Cardiovascular patient and survivor of a rare congenital heart disease that was misdiagnosed for 48 years and is found in less than 1 percent of the population. She is also a mother and guardian of a family member with multiple disabilities
- Advocated on behalf of all patients with chronic, disabling, or life-threatening medical conditions
- Provided a unique patient perspective related to her rare disease and the issues she has encountered over the years
- Holds a BA from the University of Southern California and is an MBA candidate at Pepperdine University
- Served as a member of the Advisory Panel on Rare Disease from April 2014 - April 2016

Panelist Recognition – Mardi Gomberg-Maitland

- Associate Professor of Medicine and Director, Pulmonary Hypertension Program, The University of Chicago
- Represented: Clinicians
- She is an expert clinician and researcher in the field of pulmonary vascular disease with a focus on pulmonary arterial hypertension, a rare disease that affects approximately 15- 50 people per million and for which there is no cure
- Provided the panel with in-depth clinical expertise on approaches to managing rare disease clinical research and working with patients with rare diseases
- Received her BA from Yale in Economics and Political Science, an MD with distinction in cardiovascular research from the Albert Einstein College of Medicine, and an MSc in Clinical Epidemiology from Harvard School of Public Health
- Served as a member of the Advisory Panel on Rare Disease from April 2014 - April 2016

Panelist Recognition – Philip W. Ruff

- Director, Global Market Access, Shire Pharmaceuticals
- Represented: Industry
- Part of the Shire Comparative Effectiveness Research advisory team. Worked at several commercial companies previously, including Medeval, the UK's largest Phase I contract research organization, Abbott Laboratories, Amgen, and Millennium Pharmaceuticals
- Provided the panel with in-depth industry perspectives on comparative effectiveness research and rare disease research, also providing a very important international perspective
- Received a BSc and PhD in Chemistry from the University of Leicester, is a Chartered Chemist and Member of the Royal Society of Chemistry, and holds the EU Chartered Scientist designation
- Served as a member of the Advisory Panel on Rare Disease from April 2014 - April 2016

Wrap Up and Next Steps

Adjourn

Thank you for your participation!