Advisory Panel on Rare Disease In-Person Meeting: Fall 2019

September 16, 2019

Matt Cheung, PhD, RPH
Chair

Cindy Luxhoj, MUP
Co-Chair

Gyasi Moscou-Jackson, PhD, MHS, RN
Program Officer, HDDR

Arpi Terzian, PhD, MPH
Program Officer, CEDS

pcori
PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

Webinar Information

Dial-in number (US): 1 877 309 2071

Access Code: 506-392-048 (muted)

Webinar URL:

https://attendee.gotowebinar.com/register/1064488182025914371

Webinar ID: 370-970-459

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.
  - No public comment period is scheduled

Please visit www.pcori.org/events for more information.

COI Statement

Welcome to the Rare Disease Advisory Panel in-person meeting.

I want to remind everyone that disclosures of conflicts of interest of members of the Advisory Panel are publicly available on PCORI's website. Members of the Rare Disease Advisory Panel are reminded to update your conflict of interest disclosures if the information has changed, in addition to completing your annual disclosure. You can do this by contacting your staff representative, Amanda Ruesch.

Finally, if the Rare Disease Advisory Panel will deliberate or take action on a matter that presents a conflict of interest for you, please inform one of the co-chairs so we can discuss how to best address the issue.

9/16 In-Person Meeting Agenda

Start Time	Agenda Items	Discussion Leader
9:00 AM	Welcome and Setting the Stage	Matt Cheung Cindy Luxhoj
9:20 AM	Introduction of New RDAP Members	Cindy Luxhoj
9:30 AM	Recap of Previous Meetings	Matt Cheung
9:50 AM	RDAP Speakers: Experience with the Patient & Caregiver Journey from Diagnosis to Treatment and Beyond	Julie Gortze Cindy Luxhoj
10:50 AM	**BREAK** (10 minutes)	
11:00 AM	Discussion: Rare Disease Portfolio Synthesis	Gyasi Moscou-Jackson Amanda Ruesch
12:00 PM	**LUNCH** (60 minutes)	

9/16 In-Person Meeting Agenda (continued)

Start Time	Agenda Items	Discussion Leader
1:00 PM	Discussion: PCORnet Rapid Cycle Research Opportunities	Maryan Zirkle
2:10 PM	**BREAK** (10 minutes)	
2:20 PM	Spotlight: Panelist Work Outside of RDAP	Matt Cheung Cindy Luxhoj
3:10 PM	RDAP Future Planning	Matt Cheung Cindy Luxhoj
3:40 PM	Recognition: Panel Members Rolling Off	Cindy Luxhoj
3:50 PM	Closing and Next Steps	Matt Cheung Cindy Luxhoj
4:00 PM	Adjourn	

Introductions

Introductions

Please briefly state the following:

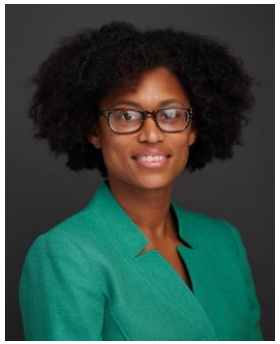
- Name
- Position title and organization
- Stakeholder group you represent

Members of the Advisory Panel on Rare Disease

- **Maureen Smith, MEd**; Patients
- **Kathleen Gondek, MS, PhD**; Industry
- **Matt Cheung, PhD, RPh** (Chair); Payers
- **Cindy Luxhoj, MUP** (Co-Chair); Patients
- **Julie Abramson**; Patients
- **Marcia Rupnow, MS, PhD**; Industry
- **Stephen Mathai, MD**; Researcher
- **Roxanna Bendixen, PhD, MS**; Researcher
- **Sherene Shalhub, MD, MPH**; Clinicians
- **Scott Berns, MD, MPH**; Clinicians
- **Julie Gortze, RN**; Patients
- **Kathleen Babich*, MA**; Patients
- **Tilicia Mayo-Gamble, PhD, MPH, MA**; Patients
- **Vanessa Boulanger, MSc**; Patients
- **Naomi Aronson, PhD**; Ex-Officio Member from PCORI's Methodology Committee

**Attending via Webinar*

Rare Disease Advisory Panel – PCORI Staff

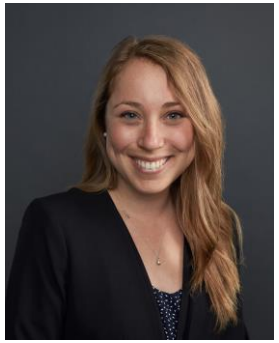


Gyasi Moscou-Jackson, PhD, MHS, RN
Program Officer,
Healthcare Delivery and Disparities Research

Arpi Terzian, PhD, MPH
Program Officer,
Clinical Effectiveness and Decision Science

Nora McGhee, PhD
Senior Manager
Office of the Chief Science Officer

Amanda Ruesch, MPH
Program Assistant
Office of the Chief Science Officer



Allison Rabinowitz, MPH
Senior Program Associate
Clinical Effectiveness and Decision Science

Sarah Philbin, MPH
Senior Program Associate
Clinical Effectiveness and Decision Science

Robin Goins
Senior Administrative Assistant
Healthcare Delivery and Disparities Research

Welcome: New RDAP Panelists

Welcome: New RDAP Member

Saira Sultan, JD

- Stakeholder group: Policy Maker
- Founder of The Haystack Project, an organization dedicated to giving voice to the experiences and perspectives of patients with rare and ultra-rare conditions to influence research and policymaking
- Also works with patient groups suffering from more common conditions, including cancer, pain, and addiction

Welcome: New RDAP Member

Doug Lindsay

- Stakeholder Group: Patients/Caregivers and Patient Advocates
- Got sick at age 21 and spent the next 11 years homebound and bed-bound until he aided in his own diagnosis, developed novel treatments to keep his rare autonomic-adrenal condition at bay, and eventually developed the innovative surgery used to fix him
- Provides in-depth Personal Medical Consulting services to rare disease and complex condition patients who have found themselves trapped in the medical system

Recap of Previous Meetings

Looking Back: Meeting Recap

Winter 2018: In-Person Meeting

- Orientation for New Panelists, including a compilation of RDAP completed activities & panel member recommendations
- Dissemination & Implementation (D&I) presented on current activities, followed by presentations from 3 PCORI-funded rare disease principal investigators on their D&I plans; Panel members offered a list of recommendations to overcome expected challenges in D&I of PCORI Rare Disease evidence
- Panel members offered recommendations to increase the feasibility of PCORnet for rare disease research
- Discussed ideas for rare disease research to inform legislature and be included in PCORI re-authorization considerations
- Panel members discussed outstanding and new topics for future activities of RDAP

Looking Back: Meeting Recap

Summer 2019: Webinar Meeting

- Based on completed on-line survey and prioritization, Panel members reviewed the top 4 ideas to expand PCORI's rare disease research portfolio and the top 4 future panel activities
- Panel members provided more details behind the top 4 new research ideas for PCORI to move forward to expand its portfolio. It was decided that "allowing adaptive research design" need not be addressed as PCORI has already adopted it
- For future topics for RDAP, members recommended:
  - Engaging, gathering, and discussing input from patients, researchers, and advocacy organizations on the diagnostic journey to identify cross-cutting rare disease research ideas
  - Consider utilizing workshops at PCORI or other rare disease meetings to further engage rare disease partners

Patient & Caregiver Journey: from Diagnosis to Treatment and Beyond

Julie Gortze

Cindy Luxhoj, MUP

One Rare Disease Patient Experience

1 in 30 Million

ME

The 1 out of 10

Julie Gortze RN
September 16, 2019
PCORI RDAP Meeting

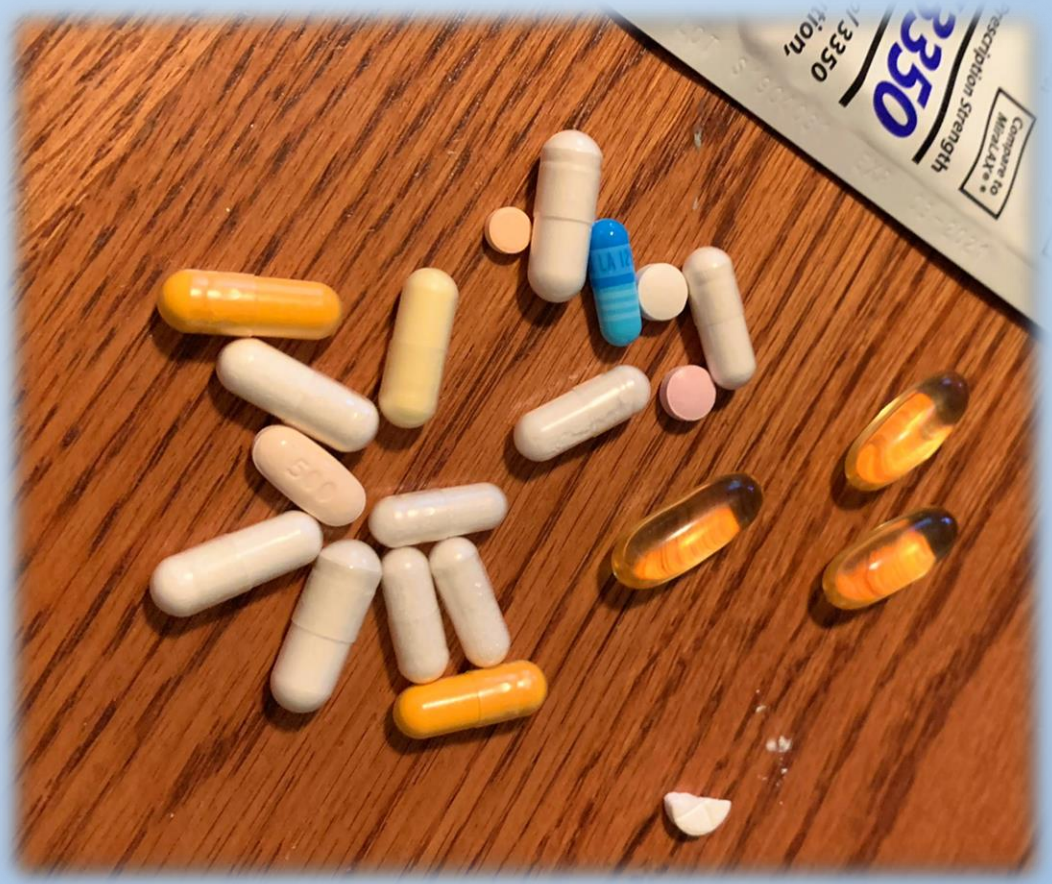
Congrats on being my 1st rare disease patient story audience!

Julie’s Medical Journey

Daily Medication Regimen

Let’s Play!

What’s covered by insurance and what’s not??



This is just the AM dose!

Julie’s Personal Burden

- Fear (even terror)
- Invalidation
- Pain
- Anxiety
- Hopelessness
- Mistrust
- Loneliness
- Financial Insecurity

On top of symptoms!

Julie’s Medical Journey Cost Throughout the Years

- Several Medical Facilities
- Multiple Doctors
- Numerous Medical Tests
- Various Diagnoses and Misdiagnoses
- Incorrect Treatments and Medications

Healthcare Provider Perception

As an RN: No education on rare diseases

As a new RD Patient: Colleagues unaware

Throughout decades of symptoms, no understanding by:
Pediatrics; Primary Care; Cardiology; ENT; GI; Neurology; Emergency; Ophthalmology; Surgical; Anesthesiology; Obstetrics; Gynecology; Rheumatology

Classic Rare Disease Patient Profile

- Consulted with many physicians in various institutions
- Multiple ER visits and hospitalizations
- Health care teams were clueless
- Decades to diagnosis (47 years!)
- Misdiagnoses with incorrect treatments
- Insurance company battles
- Symptoms blamed on anxiety and even recreational drug use
- No answers for future expectations

Questions?

Thank you for listening!

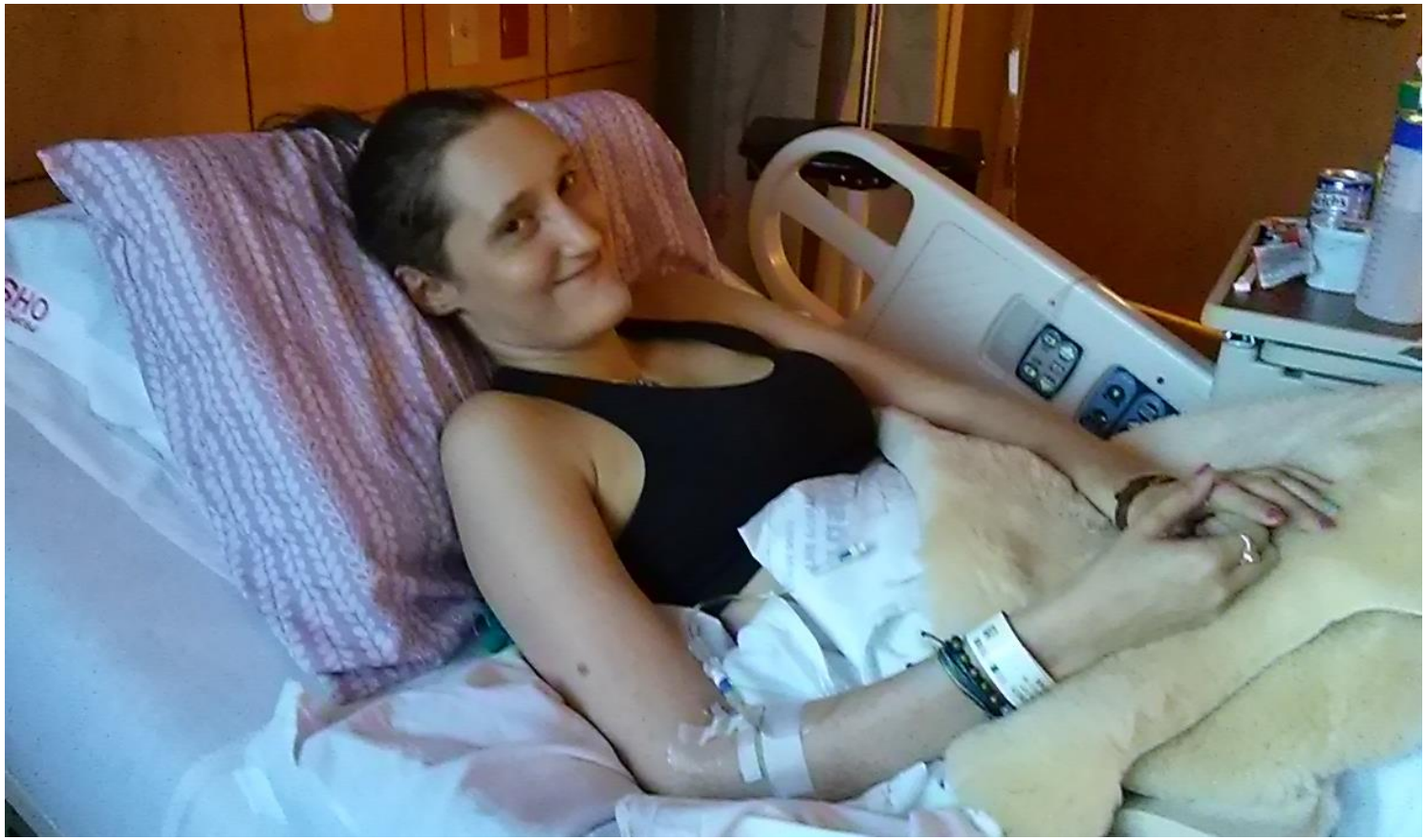
Julie@rarenewengland.org

The Caregiver's Journey

Cindy Luxhoj

Mother

- Confusion
- Uncertainty
- Explanation
- Connection
- Support
- Isolation
- Boundaries
- Expectations



Caregiver

- Questions
- Listening
- Treatment
- Specialists
- Choices
- Coverage
- Self-care
- Journey
- Letting go

Founder

- Connection
- Universality
- Understanding
- Experience
- Health
- Balance
- Resources
- Research

PCORI's Guidance

- Evidence-based
- Interventions
- Questions
- Decisions
- Real-world

Lessons Learned

- Life
- Love
- Hope

Discussion

- What is the definition of “cross-cutting” research for rare disease communities?
- Which themes resonate with your experience?
- Based on your experience, are there other themes in the diagnostic journey that have not been shared that you think should be explored?
  - e.g. research methods, healthcare delivery processes, social care, access to treatments, patient-provider communication tools or models, etc.

BREAK

10:50AM – 11:00AM

PCORI's Research Portfolio Exploration: Focus on Rare Disease (Update)

Gyasi Moscou-Jackson, PhD, MHS, RN

Program Officer,

Healthcare Delivery and Disparities Research

Amanda Ruesch, MPH

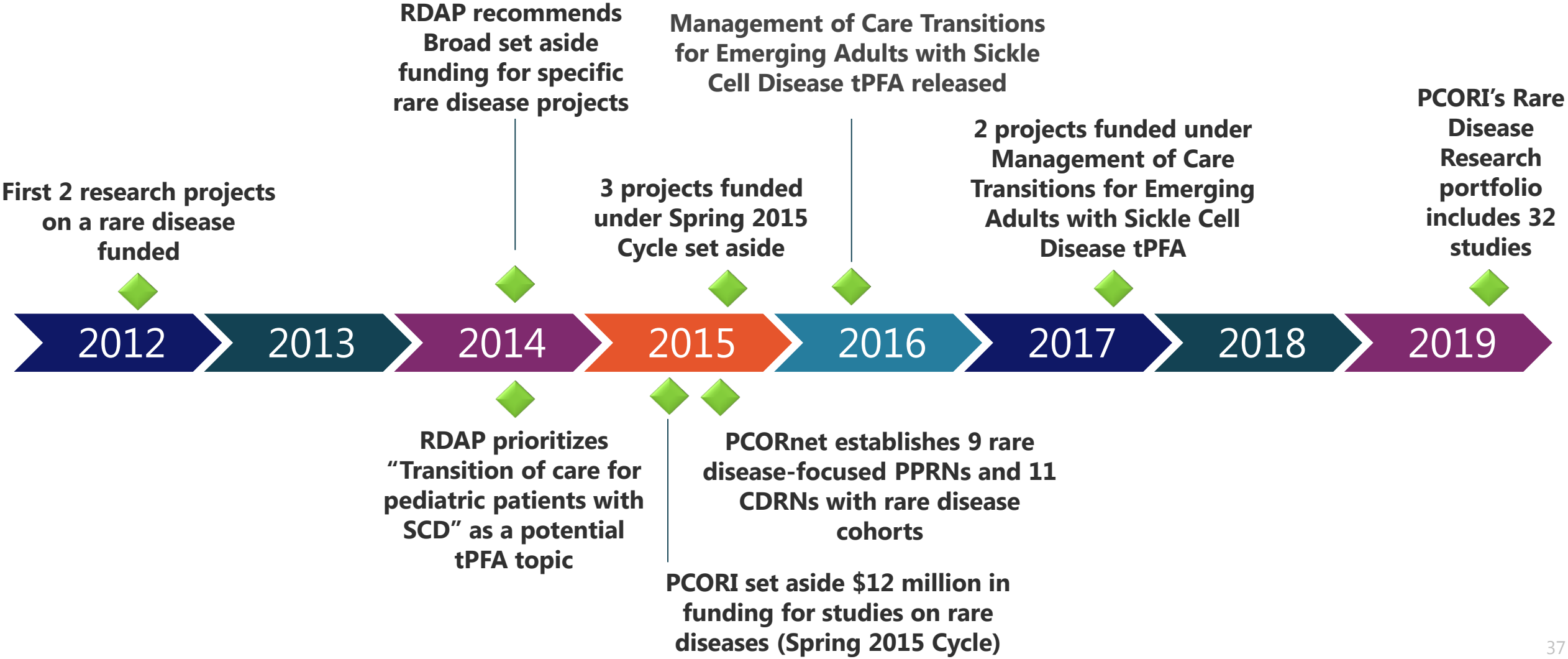
Program Assistant,

Office of the Chief Science Officer

Background

- PCORI utilizes NIH's definition of a rare disease, defined as a condition that affects **less than 200,000** in the United States.
- It is estimated that approximately **25-30 million** Americans have a rare disease (CDC).
- The exact cause of many rare diseases is still unknown and most rare diseases have no curative treatments.
- PCORI is committed to funding relevant research about what works best for individual patients.
  - This includes a commitment to funding research that addresses outstanding questions about the **prevention, diagnosis, and treatment of rare diseases**.

Timeline of PCORI Activities Related to Rare Disease



PCORI's Investment in Research Studies on Rare Disease

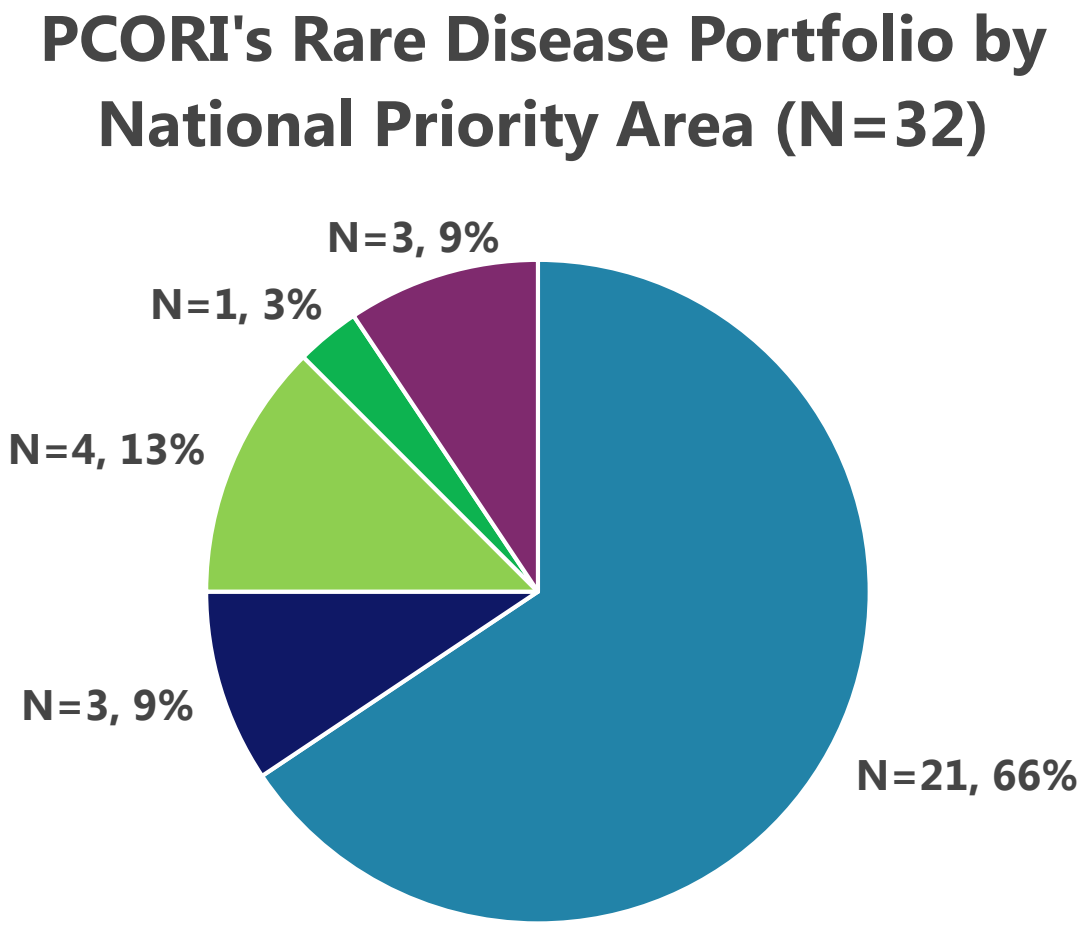
$87 MILLION

SUPPORTING **32**

RESEARCH STUDIES ON RARE DISEASE

As of September 2019

PCORI's Rare Disease Portfolio Spans the National Priority Areas



As of September 2019

Number of Funded Research Studies by Rare Disease (N=32)

Condition	Number of Studies	Condition	Number of Studies
Sickle Cell	7	Hydrocephalus	1
Cerebral Palsy	2	ID/ASD	1
Acute Myelitis Leukemia	2	Kawasaki Disease	1
Rare Disease (Non-specific)	2	Lupus Nephritis	1
Chiari Type I Malformation	1	Myasthenia Gravis	1
Crohn's Disease	1	Neuroblastoma	1
Disorders of Sex Development	1	Non-CF Bronchiectasis	1
Down Syndrome	1	Pulmonary Fibrosis	1
Duarte Galactosemia	1	Scleroderma	1
Duchenne Muscular Dystrophy	1	Subglottic Stenosis	1
Eosinophilic esophagitis	1	Transverse Myelitis	1
		Urea Cycle Disorder	1

N = 32 studies

As of September 2019

Topics Studied (N=32)

Cross-cutting CER Topics Relevant to Patients with Rare Diseases

14 studies focus on cross-cutting research topics

Shared Decision Making

- Decision support tools
- Shared decision-making toolkit
- Personalized care plans
- End of life decision-making

Patient self-directed care

- Mobile interventions for pain management
- Online disease self-management tool

Health service delivery models

- Interventions to improve treatment adherence
- Transitional care models

Methods for conducting research in rare diseases

- Self-phenotyping
- Engagement of patients and caregivers in clinical guideline development
- Study designs that are feasible for research on small samples (i.e. snSMART)

STUDY PROFILE

Comparing Transitional Care for Teens and Young Adults with Sickle Cell Disease with and without Peer Mentoring

Patient Population: 700 teens and young adults (16 to 25 years) with Sickle Cell Disease

Design: Cluster randomized controlled trial

Comparators:

- Structured education-based program
- Structured education-based program plus peer mentoring

Primary Outcomes: change in average number of acute care visits per year

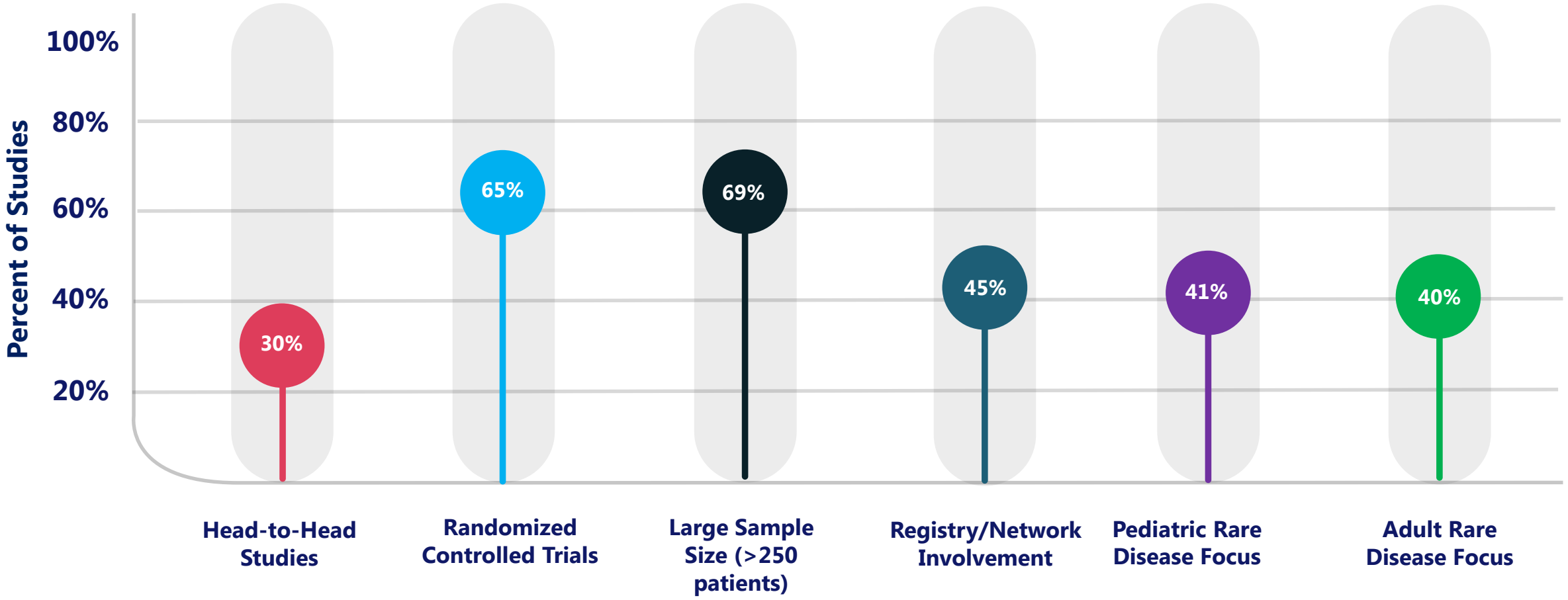
Outcome Assessment: Up to 2-year follow-up for primary outcome

Potential Impact for Rare Disease:

Could change practice by ensuring that young adults with rare diseases have the support and structure they need to successfully transfer care from a pediatric to adult provider.

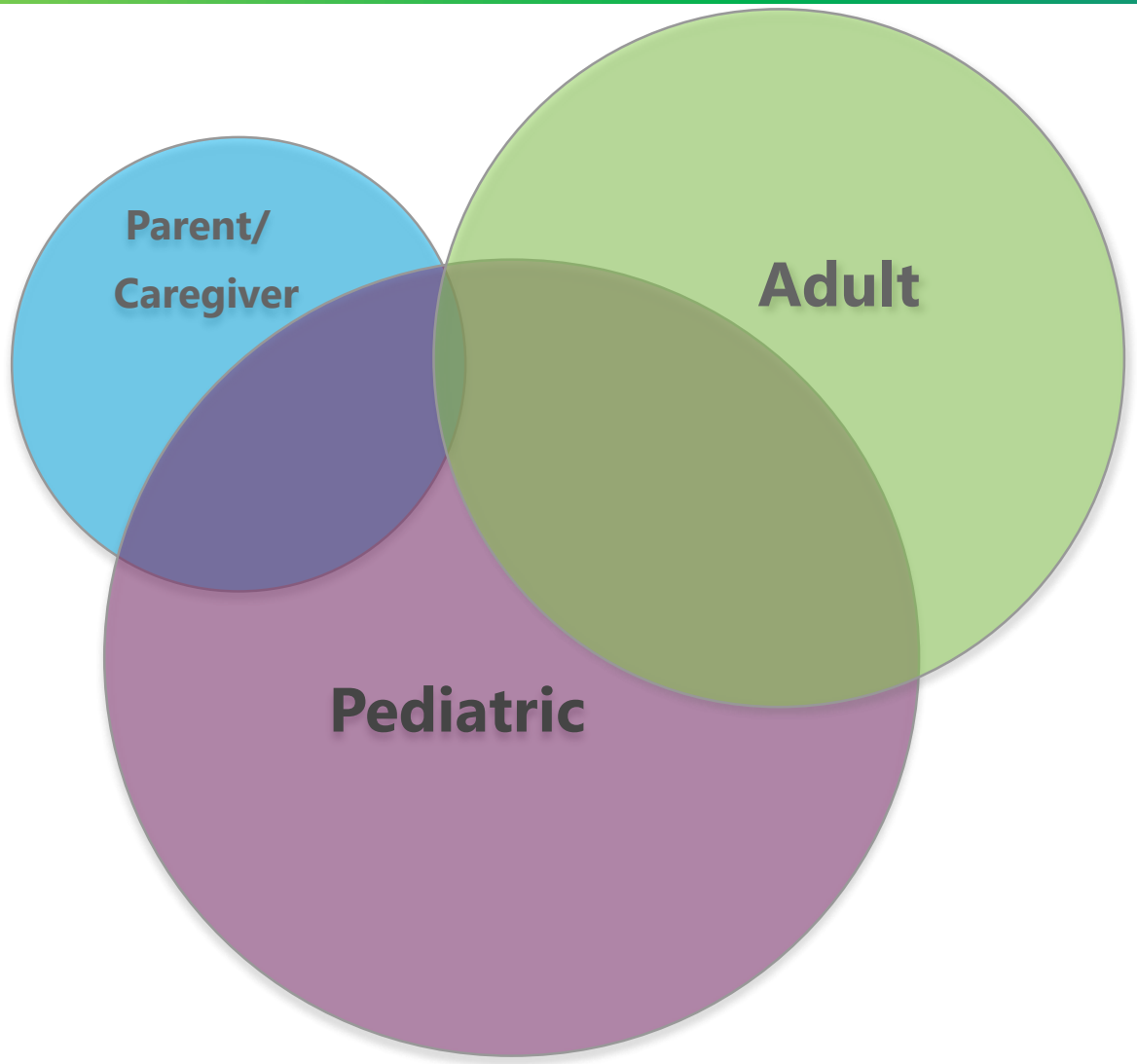
Ifeyinwa Osunkwo, MD, MPH and Raymona Lawrence, DrPH, MPH
Carolinas Medical Center
Charlotte, NC
Management of Care Transitions for Emerging Adults with Sickle Cell Disease, awarded September 2017

Key Portfolio CER Highlights* (N=29)



*Methods studies excluded; As of September 2019

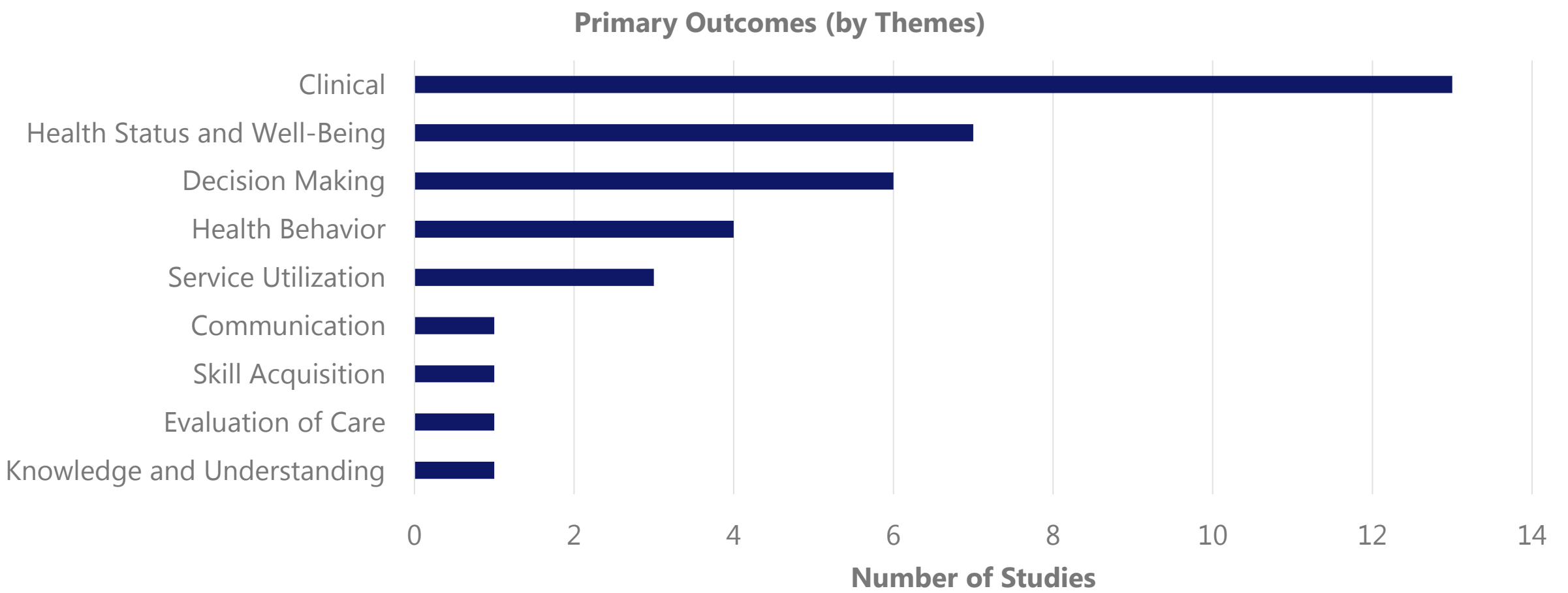
Populations of Interest



- **Pediatric (n=17)**
  - Pediatric only, n=12
- **Adult (n=14)**
  - Adult only, n=11
- **Parent/Caregiver (n=6)**
  - Parent/Caregiver only, (n=3)

As of September 2019

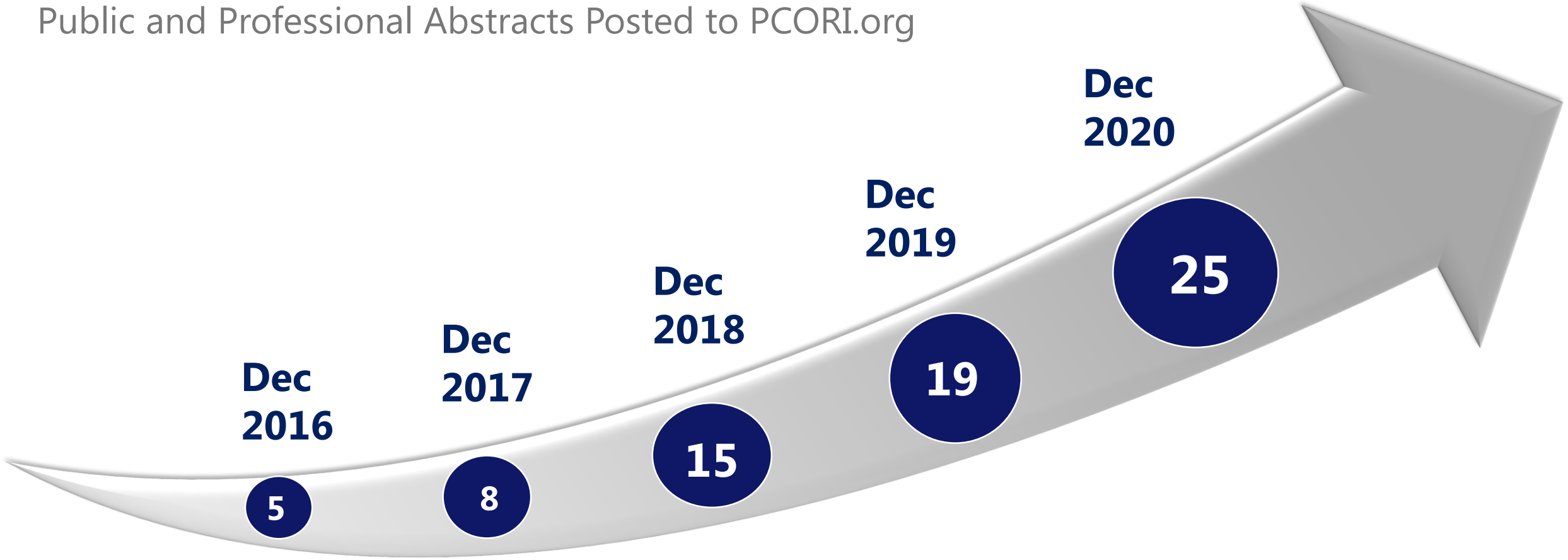
Focus on Patient and Caregiver-Centered Outcomes (N = 29)



**Not mutually exclusive; As of September 2019*

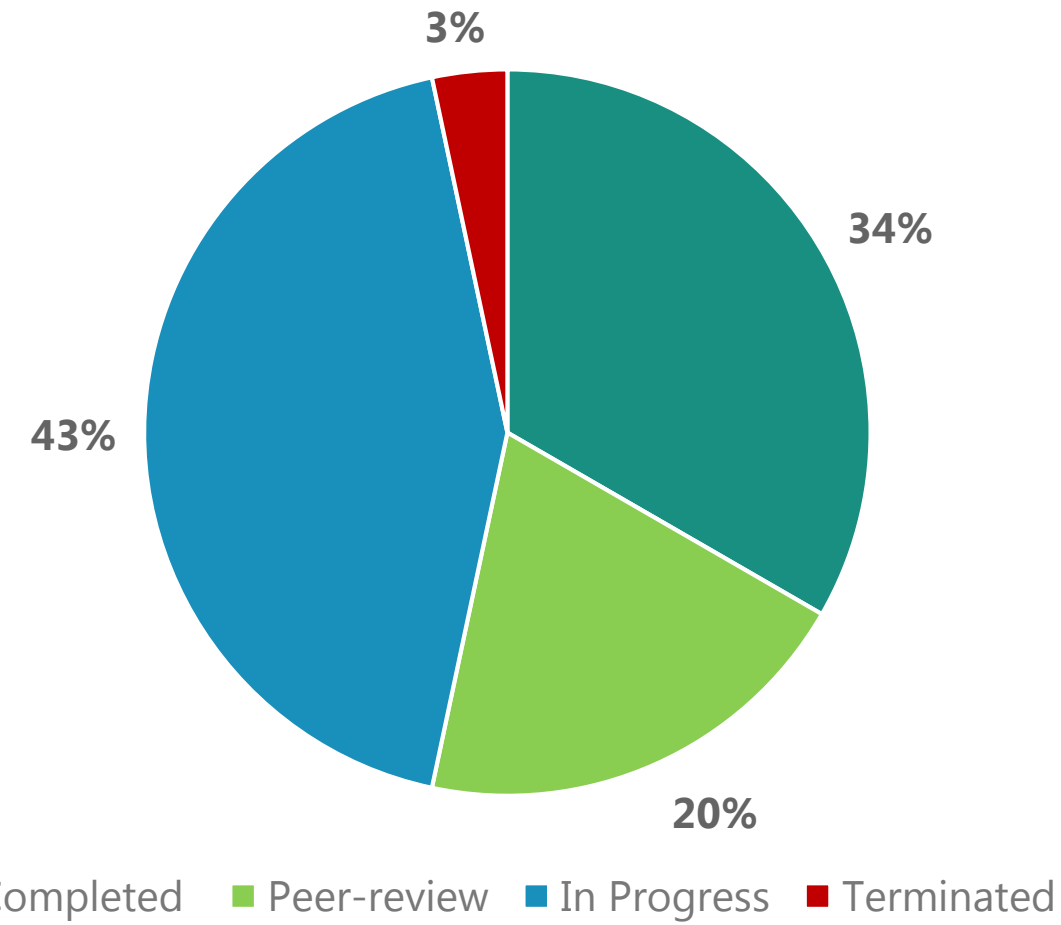
Cumulative Number of Studies with Posted Results by Year

Public and Professional Abstracts Posted to PCORI.org



As of September 2019

Rare Disease Studies with Results



As of September 2019, 10 rare disease studies have been completed, with an additional 6 undergoing PCORI peer-review.

Interviews with PCORI staff have led to early **lessons learned** regarding CER studies conducted in rare diseases

Lessons Learned from Completed Research Studies

- Many lessons learned are similar to that of common diseases but may be more challenging to navigate in studies of rare diseases.
  - Experienced investigators increase study success
  - Challenges with research environment can impede study implementation
  - Challenges in study conduct and analysis
    - Intervention delays and slow recruitment impact study timeline
    - Incomplete adherence to treatments and low retention
    - Missing data

Lessons Learned from Completed Research Studies

- **Study Planning Initiation**
  - Limited preliminary data from which to draw estimates or assess feasibility.
  - Longer study durations (i.e., > 3-5 years) may improve challenges with site start up and recruitment.
  - Investigators face tension between choosing a study design that minimizes bias and limited sample of patients.
  - When treatment options are limited or perceived to be more effective, patients (or parents) are less likely to participate or be randomized.

- **Stakeholder Engagement**
  - Stakeholder engagement can be a barrier and facilitator to conducting the study.

Lessons Learned from Completed Research Studies

- **Participant recruitment and retention**
  - Finding appropriate comparison groups may be a challenge for observational studies
  - Recruitment feasibility is increased by:
    - Access to existing patient registries/existing networks,
    - Investigator rapport with the community,
    - Use of internet-based recruitment,
    - Access to specialized clinics serving the population of interest, and
    - Focus on common rare diseases.
  - Participant retention decreased by high-burden interventions and disease severity.

Engagement & Infrastructure Complements to Our Portfolio

Research Infrastructure Awards	Awarded Amount
PFA: Partnerships to Conduct Research within PCORnet (PaCR) **Project Title:** Improving Outcomes in Limited Juvenile Idiopathic Arthritis **PI:** Laura Schanberg, MD	**$7M**
Engagement Awards	
Total number of awards: 50	**$7M**
Engagement Award Project: 8 Example: *Tennessee Sickle Cell Disease Network Project, Velma Murray, PhD; $250K*	$1.8M
Engagement Award Conference: 42 Example: *2019 NEC Symposium, Erin Umberger, M.Arch; $50K*	$5.2M

Discussion Questions

- Which cross-cutting CER evidence gaps should PCORI examine in the future? (e.g., care coordination, delivery models, diet/nutrient interactions, etc.)
  - Are there evidence gaps we sufficiently explored?
  - Should PCORI seek more studies on caregivers and/or adults? If so, which topics may be ready for CER?
- Based on your experience, under what circumstances might usual care be an appropriate comparator?
- Based on your experience, what other facilitators increase the success of rare disease studies?
- What are potential strategies to overcome the challenges encountered? Are there any challenges we can explore further with the RDAP?
- Other thoughts?

LUNCH

12:00PM – 1:00PM

Overview: What is PCORnet?

Maryan Zirkle, MD, MS, MA

Associate Director, Research Infrastructure

PCORI set out to help improve the conduct of clinical research

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?

PCORnet® : The National Patient-Centered Clinical Research Network

The National Network was created to be **a large, highly representative, national patient-centered clinical research network**.

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

The **mission:** 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
- **Uses** multiple data sources including electronic health records, insurance claims data, data reported directly by people, and other data sources
- **Enables** people and systems to work collaboratively

Harnessing the Power of Partnerships

9
Clinical Research Networks (CRNs)

2
Health Plan Research Networks (HPRNs)

Patient Partners

Coordinating Center (CC):
Duke (DCRI), Harvard Pilgrim Healthcare Institute (HPHCI)

Central Office: People Centered Research Foundation (PCRF)

CDRN Rare Disease Cohorts

CDRN	Rare Condition(s)
ADVANCE	Alpha-1 antitrypsin
CAPriCORN	Sickle cell disease
	Recurrent clostridium difficile
GPC	ALS
STAR	Sickle cell disease
INSIGHT	Cystic fibrosis
OneFlorida	Duchenne muscular dystrophy
PaTH	Idiopathic pulmonary fibrosis
PEDSnet	Autism
REACHnet	Sickle cell disease

Overview: Rapid Cycle Research & Projects (RCRP) Initiative

Award of **up to $7.0 M** for projects that utilize the PCORnet infrastructure for:

- Descriptive analyses intended to help assess the feasibility of studies addressing **stakeholder-prioritized topics** (prep to research)
- Implementing small, rapid, observational studies including analyses on PCORI- **prioritized stakeholder-driven topics**

Responsive to PCORI's need for rapid evidence generation

- PCORnet was identified as an infrastructure to support rapid research
- Rapid, descriptive observational research will be designed to lead to high-impact pragmatic studies

Approved RCRP Implementation Process

Where do the topics come from?	PCORI Staff in Science or Engagement PCORI Strategy Committees Stakeholder groups working with PCORI (e.g., clinicians, payers)
How do we determine if PCORnet is the best resource?	In collaboration with PCORI Research Infrastructure Staff and the PCORnet Coordinating Center/Data Operations Center
What is the review process?	PCORI staff is authorized to award funding for specific topics ONLY after the feasibility assessment and rapid review/approval process jointly developed by SOC and RTC (up to $7M in 2017)
Should we consider building off the preliminary findings, who would fund this?	Any larger studies that would develop from the RCRP findings would go through the usual Board approval process

RCRP in ACTION: How Rapid is it?

Protocol and Budget Finalized & Contract Executed

Descriptive Analysis

OR

Small Observational Studies

Up to 700K in funding

Pre-Award Review Process:
<3 months

Post-Award Contracting:
<1 month

Awards:
~ 9 months

~ 1 year from Topic to Results

RCRP in ACTION: Approved Projects

Topic Area	Research Question	Project Completion Date
PCSK9	Trends and current prevalence of use of PCSK9 Inhibitors in persons who are eligible for statin therapy	April 2018 **COMPLETED**
Cancer	Patterns of use of molecular and genetic biomarkers/targeted cancer treatment for new onset solid tumors	Sept 2018 **COMPLETED**
Hep C	Do pts with Hep C who are prescribed newer direct acting antiviral meds experience higher rates of adverse events than pts with Hep C who are untreated?	Jan 2019 (includes modified peer review) **COMPLETED**
T2DM	Trends in use of newer agents in treatment of T2D and the potential for outcomes studies	Feb 2019 **COMPLETED**
Opioids	Evaluate and Demonstrate the Fitness for Use of PCORnet for surveillance of the opioid epidemic	Oct 2019
PROMs	Implementation of Patient-Reported Outcomes Measurement in Routine Clinical Practice for Heart Failure Patients in PCORnet	Dec 2019

RCRP Examples

Cancer: *Patterns of use of molecular and genetic biomarkers/targeted cancer treatment for new onset solid tumors*

PROJECT COMPLETED

Stakeholder(s): ***PCORI Staff, NCI, FDA***

- **CDRN Lead**
  - Greater Plains Collaborative (GPC)
- **Study Design**
  - **Descriptive: Retrospective database study with chart review for a sample of patients**
- **Project Objective**
  - Document the patterns of use of molecular biomarkers and molecular-guided cancer therapies for patients with solid tumors in real-world settings.
- **Population**
  - Electronic medical record data for 86,154 patients diagnosed with a single primary solid tumor between 2013 and 2016.
  - 11 PCORnet CDRNs across 10 states

- **Conclusions**
  - The percentage of patients that received molecular testing ranged from 26 percent for patients with endocrine cancers to 76 percent for those with breast cancer.
  - Information from the shared database showed that about 5 percent of all patients in the 11 networks received molecular targeted therapy after their tests. Patients age 60 years and older had the lowest use of this therapy.
    - Patients with breast, lung, or head and neck cancer had the highest use of this therapy.
  - In 2 of the health systems examined, fewer than 20 percent of patients with colorectal cancer received molecular targeted therapy. But all patients who had molecular targeted therapy had test results that supported that treatment decision.
  - The project team compared two sets of data. The first set only included data from the database. The second set included data from the database plus Medicare insurance data. Using database data alone found that just 49 percent of patients had molecular tests, Medicare plus database data showed that 60 percent of these patients had the tests. Using database data only showed that about 4 percent of the patients had molecular therapy compared with about 7 percent of patients found using Medicare plus database data.

Hep C: *Do patients with Hep C who are prescribed direct acting antiviral meds(DAAs) experience high rates of adverse events than patients with Hep C who are untreated?*

PROJECT COMPLETED

Stakeholder(s): ***FDA, PCORI Staff***

- **CDRN Lead**
  - Patient Outcomes Research To Advance Learning (PORTAL)
- **Study Design**
  - **Retrospective observational study**
- **Project Objective**
  - Assess the rates of adverse events in patients with HCV infection exposed to DAAs compared with those not exposed.
- **Population**
  - Electronic medical record data for 33,808 patients diagnosed with Hepatitis C between 2012 and 2017.
  - 3 Health Systems/sites within PORTAL
- **Conclusions**
  - DAAs were not linked with patients having more adverse events. Overall, compared with patients who had not received DAAs, patients who received DAAs had lower rates of:
    - Death
    - Reduced liver function
    - Acute-on-chronic liver event
    - Arrhythmia
    - Multiple organ failure
    - Hospital or emergency room visits
  - Safety concerns based on analyses of the US Food and Drug Administration's Adverse Events Reporting System did not appear to be confirmed, suggesting that dispensed direct-acting antivirals may be safe for patients with hepatitis C.

Discussion Topics

- Specify a **disease/condition** for RCR Project
- Identify an **external stakeholder** to support question prioritization
- Identify a **research question** with external stakeholder

BREAK

2:20PM – 2:30PM

Panelist Work Outside of the RDAP

Spotlight

In a few sentences, please summarize your relevant rare disease efforts outside of PCORI.

- Goals:
  - Discuss member activities outside of the panel that could be relevant to the work of the panel
    - Including activities that are completed, especially those that have led to a deliverable output
  - Increase awareness of potential topics to explore at future meetings

Planning Ahead: The Future of RDAP

Planning Session: PCORI Goals

PCORI Strategic Goals

1. Increase quantity, quality, and timeliness of useful, trustworthy research information available to support health decisions
2. Speed the implementation and use of patient-centered outcomes research evidence
3. Influence research funded by others to be more patient-centered

Planning Session: RDAP Scope of Work

RDAP Function and Scope of Work

- Provide input to PCORI on research needs of the rare disease community and on specific issues and concerns in conducting research on rare diseases
- Identify existing infrastructure (registries, tools, etc.) that can be a resource for conducting research
- Provide ongoing feedback and advice on evaluating and disseminating PCORI's research portfolio on rare diseases
- 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

Planning Session: Developing Goals

- Based on the RDAP function and scope of work, which topics/activities could the RDAP take action on during the coming year?
  - Which of these topics/activities do you consider as the highest priority for the RDAP in the coming year? In future years?
  - Which of these topics/activities should be considered low priority?

Recognition of Departing Panel Members

Thank you!

We'd like to give special thanks to those members whose terms end this year:

- Maureen Smith
- Kathleen Gondek
- Matt Cheung

Panelist Recognition: Maureen Smith, MEd

- Represented: Patients, Caregivers, and Patient Advocates

- Smith has a long history as a patient advocate after being diagnosed with a rare disorder at the age of 8
- Memberships include:
  - Board of Directors of the Canadian Organization for Rare Disorders (CORD) since 2009; current Secretary
  - Canadian Institute of Health Research's (CIHR) Institute of Genetics' Health Services, Policy and Ethical, Legal and Social Issues Research Priority & Planning Committee
  - Patient member of Ontario's Committee to Evaluate Drugs

- Assisted with the development of the core outcome set initiative
- Provided the patient perspective of the challenges associated with rare diseases

Panelist Recognition: Kathleen Gondek, MS, PhD

- Represented: Industry
- Vice President of Global Health Economics Outcomes Research and Epidemiology, Shire PLC
- Over two decades of experience in the pharmaceutical industry with numerous co-authored publication in peer-reviewed journals
- Gondek brought a wealth of knowledge and perspective on the advancement and availability of treatments for patients with rare diseases

Panelist Recognition: Matt Cheung, PhD, RPh

- Represented: Payers

- Adjunct Professor of Pharmacy Practice at University of the Pacific
- Served as Chair of the Advisory Panel on Rare Disease since August 2019
- Cheung has been an involved and hard-working Chair, providing expert leadership and direction for the RDAP. His initiatives have included:
  - Creation and upkeep of the RDAP meeting crosswalk
  - The development of the core outcome set
  - The RDAP Website Review Workgroup

Acknowledgements & Recap

Next Steps

- New RDAP Chair nomination approval by Science Oversight Committee on September 17th and the Board of Governors in November 2019.
- Determine next meeting date – Winter 2020?