Advisory Panel on Rare Disease (RDAP) Virtual Meeting: Spring 2021

May 14, 2021

Scott Berns, MD, MPH, FAAP
Chair, RDAP

Doug Lindsay, BS
Co-Chair, RDAP

Nora McGhee, PhD
Senior Program Officer, CEDS
Staff Co-Lead, RDAP

Carly Khan, PhD, MPH, RN
Program Officer, HDDR
Staff Co-Lead, RDAP

Rohini Mohanraj, MHA
Program Associate, Research Infrastructure
Panel Coordinator, RDAP

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RDAP Chairs

Scott Berns, MD, MPH
Chair, Advisory Panel on Rare Disease
Chief Executive Officer, National Institute for Children's Health Quality

Doug Lindsay, BS
Co-Chair, Advisory Panel on Rare Disease
Personal Medical Consultant and Founder of Doug Says LLC.

Housekeeping

- Please note that today's webinar is being recorded for posting on PCORI's website.
- Members of the public are invited to listen to the teleconference and view the webinar.
- Meeting materials can be found on the PCORI website. The recording of the webinar will also be made available to the public after this event.
- 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 Spring 2021 virtual 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, Rohini Mohanraj.

Finally, if the Rare Disease Advisory Panel will deliberate or act 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.

Meeting Agenda

Start Time	Agenda Items	Presenters & Discussion Facilitator
1:00 PM	Welcome, Introductions, and Setting the Stage	Scott Berns, Doug Lindsay
1:10 PM	Introduction of RDAP PCORI staff, RDAP Program Staff Update, Upcoming Activities	Carly Khan, Nora McGhee
1:15 PM	Rare Disease Research Awards: Study Highlights and Discussion	Jason Gerson, Penny Mohr, Daniel Herman, Jinbo Chen, Michelle Denburg, Aneta Jovonovska, Rebecca Levondosky Michael O'Rorke, Josh Mailman, Anne Berg, Sandi Lam, Tracy Dixon-Salazar, Tom Carton, Anitha John, Scott Leezer, Scott Berns
2:10 PM	**BREAK**	
2:25 PM	Engagement Awards and Rare Disease Organizations: An Update and Project Highlights	Karen Martin, Jennifer Canvasser, Doug Lindsay

Meeting Agenda

Start Time	Agenda Items	Presenters & Discussion Facilitator
3:10 PM	Identifying Our National Priorities for Health: Relevance for Rare Disease Populations	Laura Lyman Rodriguez, Scott Berns
3:40 PM	An Update on the Cost-Data Provision	Andrew Hu, Doug Lindsay
3:55 PM	BREAK	
4:10 PM	COVID Connect: PCORI's Response to the COVID-19 Pandemic	Claudia Grossmann, Scott Berns
4:40 PM	Acknowledgments and Recap	Scott Berns, Doug Lindsay
4:50 PM	Farewell to Departing Members	Doug Lindsay
5:00 PM	Adjourn	

RDAP Panelist Introductions

RDAP Members

CLINICIANS

Scott Berns (Chair)
- CEO, National Institute for Children's Health Quality

Nancy Rose (American College of Medical Genetics and Genomics)

Sherene Shalhub (University of Washington)

Laura Tosi (Children's National Hospital)

RESEARCHERS

Roxanna Bendixen (University of Pittsburgh)

POLICY MAKERS

Saira Sultan (Connect4Strategies)

INDUSTRY

Salman Hussain (Charles River Associates)

PATIENTS, CAREGIVERS, AND PATIENT ADVOCATES

Doug Lindsay (Co-Chair)
- Personal Medical Consultant

Sarah Bacon (Patient, advocate, and writer)

Vanessa Boulanger (Amyloidosis Research Consortium)

Danielle Boyce (COPD Foundation)

Julie Gortze (Rare New England)

Matthew J. Edick (Michigan Public Health Institute)

Tilicia Mayo-Gamble (Georgia Southern University)

EX-OFFICIO MEMBER

Naomi Aronson (BCBSA)

RDAP Staff Introductions

Rare Disease Advisory Panel – PCORI Staff

Carly Khan, PhD, MPH, RN
Program Officer
Healthcare Delivery and
Disparities Research

Nora McGhee, PhD
Senior Program Officer
Clinical Effectiveness
and Decision Science

Rohini Mohanraj, MHA
Program Associate
Research Infrastructure

PCORI Rare Disease-related Updates

- March 2021: The PCORI Board of Governors approved $19 million to fund four CER studies focused on rare diseases in response to the Conducting Rare Disease Research Using PCORnet® targeted funding announcement

- Advisory panel applications are under review – we anticipate new members will join us for the Winter 2021 Meeting

- Save the Date: The 2021 PCORI Virtual Annual Meeting will be held Wednesday, November 17th – Friday, November 19th 2021

Rare Disease Research Awards: Study Highlights and Discussion

Conducting Rare Disease Research using PCORnet®

Overview of Funding Announcement and Awards

Penny Mohr, MA
Interim Program Director, Research Infrastructure

Conducting Rare Disease Using PCORnet

- Signaling that PCORI gives special attention to the conduct of rare disease research, PCORI's authorizing legislation mandated the establishment of this panel - RDAP.

- The ability to conduct robust rare disease comparative effectiveness research (CER) is often limited by small numbers

The vision behind this funding initiative: Harness the scale of PCORnet to enable improved comparative effectiveness research in rare disease

PCORnet as a Resource for Rare Disease Research

Scale

- Power in numbers so that studies are possible in rare conditions

Data

- Collected at point of care, reflective of patient experience within health system,
- Collected from a variety of specialties and clinics to support understanding progression of disease
- Linkable with claims data and/or PROs for more complete picture of patient experience

Access

- To patients behind the data, allowing contact for study participation through ethically approved channels

Engagement

- Involvement of patients with lived experience to help develop patient-centered questions and studies

RDAP Contribution to PFA

- Long expressed an interest in use of PCORnet for rare disease research
- In 2019, PCORI worked with RDAP to identify priority areas of research
- Some priority questions identified by the RDAP include:
  - Retrospective evaluation of use patterns and presentation prior to diagnosis of rare disease, mechanisms to accelerate diagnosis;
  - Effective symptom management;
  - Mechanisms to improve case management, care coordination services;
  - Transitions of care (across the lifespan, between primary and specialty care);
  - Monitoring and screening for rare disease complications post-diagnosis.
- Particular interest in pediatric rare disease

Cycle 2 2020 – Rare Disease PCORnet Targeted PFA

PFA Overview

- **Objectives:**
  - Utilize PCORnet resources to perform an observational research study that will answer one or more important research questions about the care of patients with rare diseases
  - Enhance the capabilities for the conduct of multi-site rare disease research by creating partnerships, methods, tools, and linkages that will facilitate future comparative effectiveness studies

- **Funding Announcement Issued:** May 2020

- **Awards Announced:** March 2021

- **Maximum project period:** 3 years

Awarded Projects

- **Comparative Effectiveness of Palliative Surgery vs. Additional Anti-seizure medications for Lennox-Gastaut Syndrome**
  PI: Anne Berg, PhD Rare Disease Partners: Lennox-Gastaut Syndrome Foundation
  - What is the comparative effectiveness of palliative surgery versus additional medications on improving outcomes in Lennox-Gastaut Syndrome (LGS)?
- **Preserving Kidney Function in Children with Chronic Kidney Disease (PRESERVE)**
  PI: Christopher Forrest, MD, PhD Rare Disease Partners: Families Associated with GLEAN Network
  - What is the comparative effectiveness of different blood pressure monitoring practices and medication strategies among children with chronic kidney disease (CKD) on preserving kidney function?
- **Utilizing PCORnet to Support Transition from Pediatric to Adult Centered Care and Reduce Gaps in Recommended Care in Patients with Congenital Heart Disease**
  PI: Thomas Carton, PhD, MS Rare Disease Partners: Adult Congenital Heart Association
  - What are the effects of gaps in recommended care (cardiology visits) on patient prioritized outcomes for adults with non-complex and complex subtypes of congenital heart disease (CHD)?
- **Comparative Effectiveness Research for Neuroendocrine Tumors (CER-NET)**
  PI: Michael O'Rorke, PhD Rare Disease Partners: Five major NET Patient Associations
  - What is the optimal therapy selection and sequencing for patients with gastroenteropancreatic (GEP) or lung neuroendocrine tumors (NETs)?

Ann & Robert H. Lurie Children's Hospital of Chicago

LGS FOUNDATION
LENNOX-GASTAUT SYNDROME

Northwestern Medicine
Feinberg School of Medicine

Comparative Effectiveness of Palliative Surgery versus Additional Anti-Seizure Medications for Lennox-Gastaut Syndrome

Anne Berg PhD
Sandi Lam MD MBA
Tracy Dixon-Salazar PhD

Presenting to:

pcori®

Patient-Centered Outcomes Research Institute

Lennox Gastaut Syndrome

LGS first diagnosed at 2-5 years

In the Unites States:
- 13,400 children
- 34,300 adults

Living with LGS

Maslow's Hierarchy of Needs

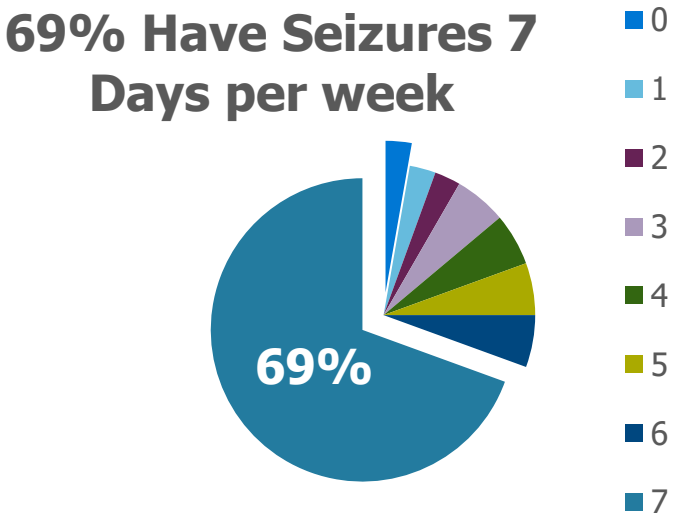
Maslow (1943). "A theory of human motivation."

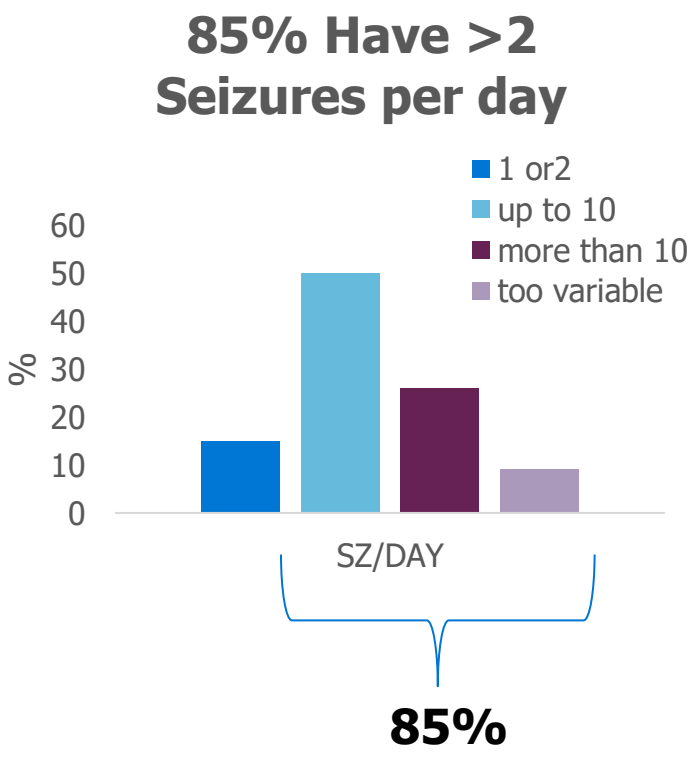
LGS Family & Caregiver Hierarchy of Needs

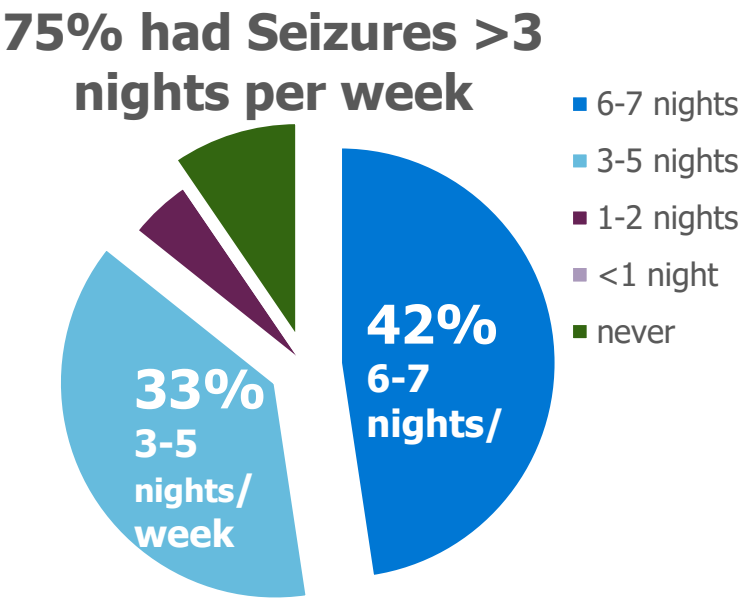
Dixon-Salazar, et al. (2021).
LGS Patient Focused Drug Development Meeting (PFDD)

LGS is relentlessly refractory

Ann & Robert H. Lurie Children's Hospital of Chicago







Northwestern Medicine® Feinberg School of Medicine

Parent-driven natural history survey

Emergencies are common

In previous 6 months

>30% had Seizure emergencies

30% ED visit

- 15% Multiple ED visits

51% used any rescue medications*

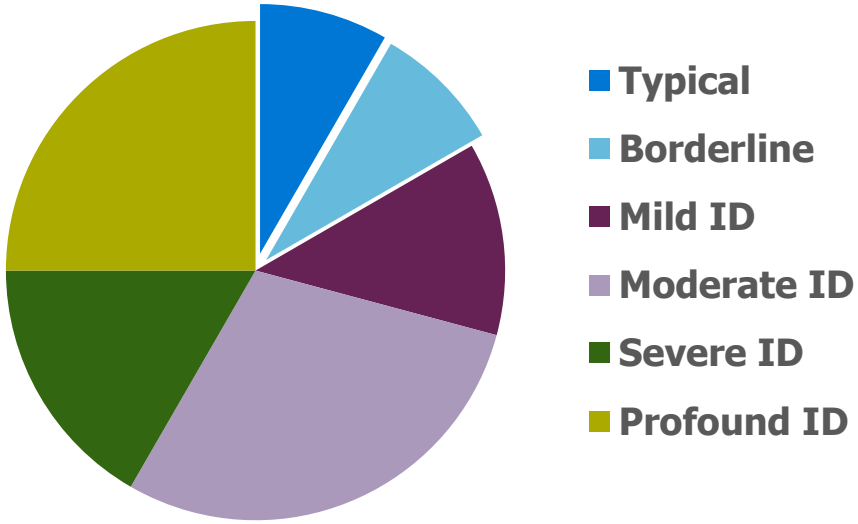
- 24% **≥10 times**

Parent-driven natural history survey

Beyond Seizures

- LGS is associated with moderate to profound disabilities

Intellectual disability



Rantala & Putkonen, Epilepsia 1999

>50% have Autism

- 35% diagnosed
- 23% features

>1/2

Beyond Seizures – Basic Functional Abilities

31% require a mobility device at home or school

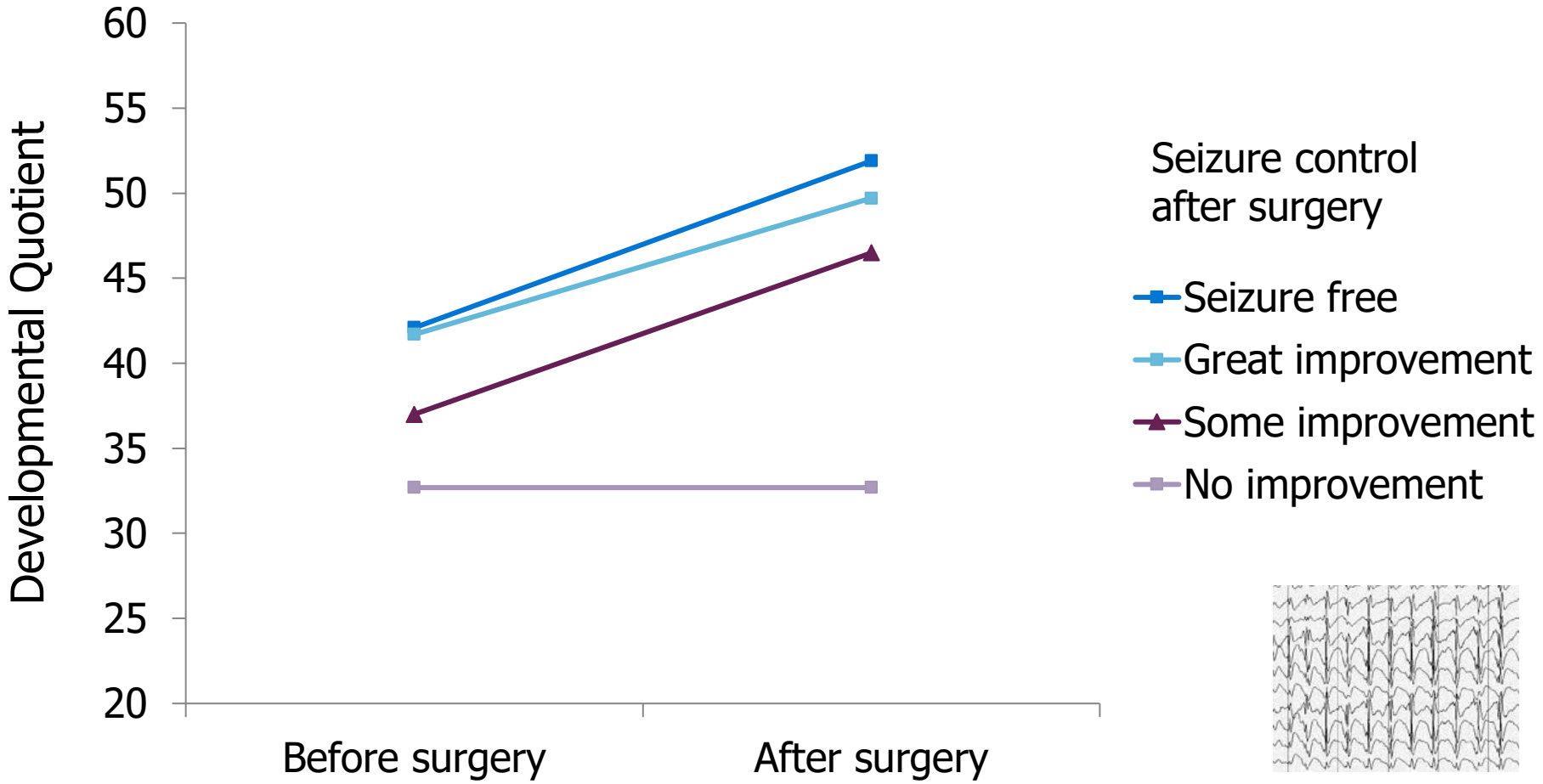
25% typically do not manipulate objects with their hands.

42% cannot feed themselves including
29% with G-tubes

50% do not use spoken language

83% dependent for toileting needs

Resective Surgery improves seizures and cognition in patients with LGS



Lee Y-J et al. Pediatrics, 2010

CRITICAL REVIEW – INVITED COMMENTARY

Epilepsia®
2021

The role of surgery in the management of Lennox–Gastaut syndrome: A systematic review and meta-analysis of the clinical evidence

Vineeth Thirunavu[1,2] | Rebecca Du[1,2] | Joyce Y. Wu[3,4] | Anne T. Berg[3,4] | Sandi K. Lam[1,2]

Non-candidates for resective surgery

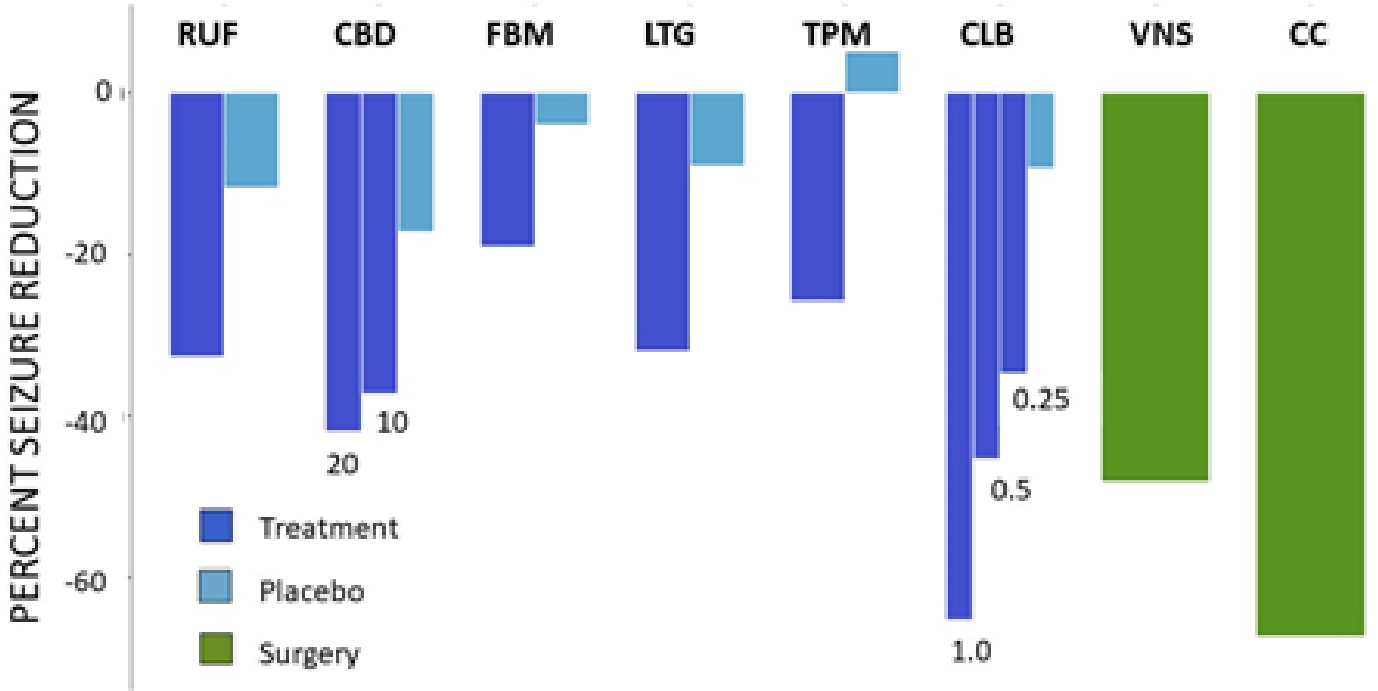
- VNS
  - Substantial reduction of atonic and tonic seizures
  - **Increased alertness**
  - No major medical complications
- Callosotomy
  - Substantial seizure reduction
  - **Some cognitive improvements**
  - Some side-effects (disconnection syndrome, transient weakness)

Non-candidates for resective surgery: Keep trying medications or try palliative surgery?

Ann & Robert H. Lurie Children's Hospital of Chicago

Published RCTs and observational studies

Figure 4: Current best evidence concerning impact of the comparators on seizure frequency ***in people with LGS.***



RUF=Rufinamide, **CBD**=Cannabidiol, **FBM**=Felbamate, **LTG**=Lamotrigine, **TPM**=Topiramate, **CLB**=Clobazam, **VNS**=Vagus Nerve Stimulator, **CC**=Corpus Callosotomy. CBD trialed at 20 and 10 mg/kg/day. CLB trials at 0.25, 0.5, 1.0 mg/kg/day

PRESERVE

Preserving Kidney Function in Children with Chronic Kidney Disease

PCORI Rare Disease Advisory Panel Meeting
May 14, 2021

Michelle Denburg – Co-PI
Aneta Jovanovska – Co-I
Becka Levondosky – Co-I
Mark Levondosky – Co-I

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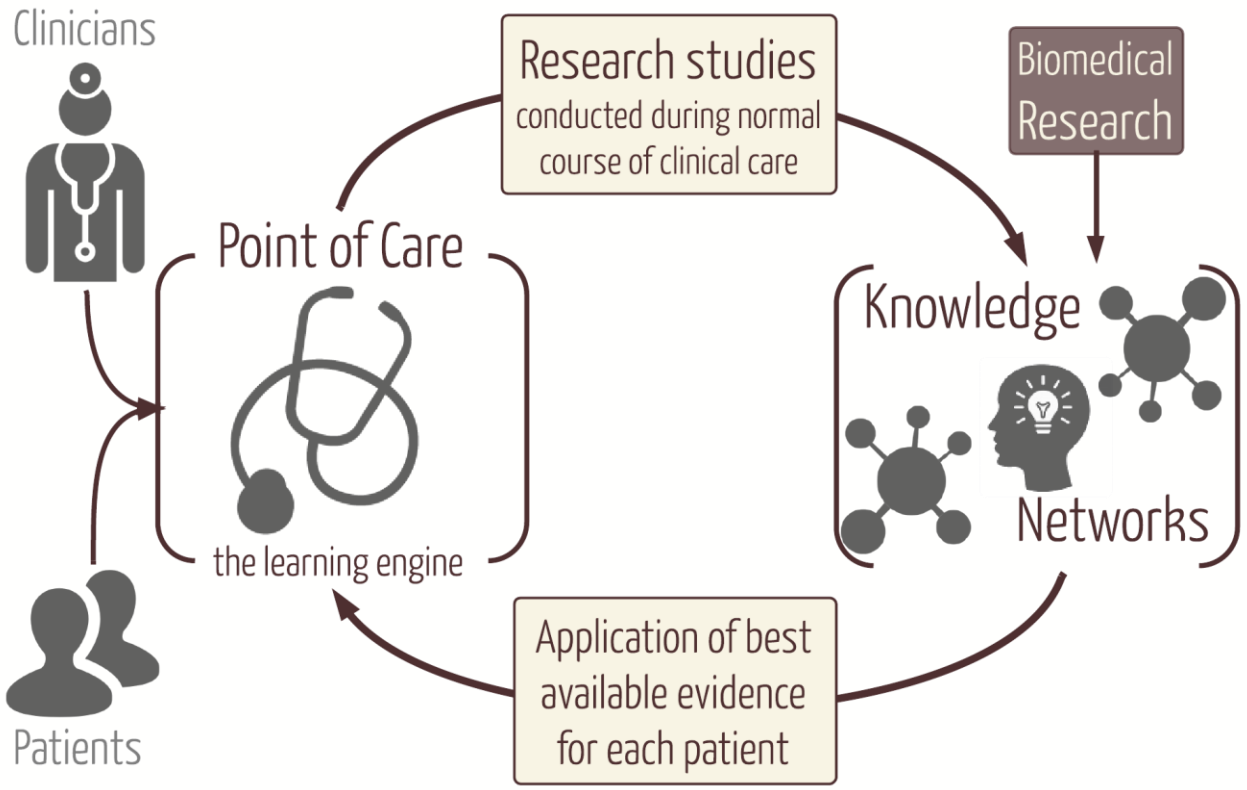
THE LEARNING CYCLE



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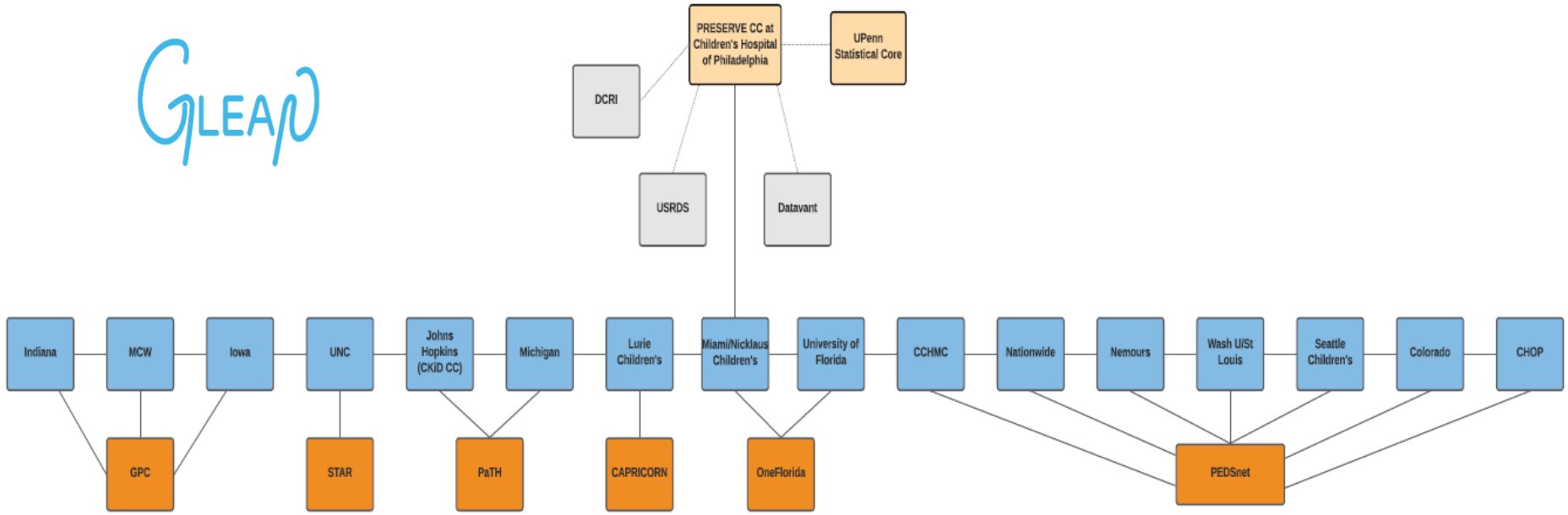
PRESERVE CONSORTIUM

pcori
PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

PEDSnet
A Pediatric Learning Health System

GLEAN

6 PCORnet networks
16 institutions
~11,000 children



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OVERVIEW

- Chronic kidney disease (CKD) in childhood is rare
  - congenital or immune-mediated conditions that lead to progressive loss of kidney function
- Once kidney function declines to end-stage kidney disease, patients require dialysis or transplantation
  - significant morbidity and shortened life expectancy
- Hypertension is a major modifiable clinical factor that contributes to kidney function decline
- Few clinical trial and large-scale observational studies evaluating alternative blood pressure (BP) management approaches
- Despite considerable focus on BP management in pediatric CKD, real-world control of hypertension remains suboptimal

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OVERVIEW

The **purpose** of **PRESERVE** is to provide new knowledge to **inform shared decision-making regarding BP management for pediatric CKD**

Specifically, this project will:

1. Expand and improve the PCORnet common data model for research in children with kidney disease
   - new pediatric- and kidney-specific variables and linking patients' EHR data to other kidney disease databases.
2. Compare the effectiveness of alternative strategies for monitoring and treating hypertension on preserving kidney function.
3. Assess the lived experiences of patients and caregivers related to BP management.

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METHODS

- The first two parts of this project will involve analyses of electronic health record (EHR) data, and the third part will involve a survey of children with CKD and their caregivers
- EHR data will be obtained from 2009 to 2022, providing a rich resource for studying patients from the time they are diagnosed with CKD to years later to determine if alternative BP clinical management strategies are associated with preservation of kidney function

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METHODS

- Study population: children with mild-moderate CKD (stages 2-3)
- Primary outcome: kidney function decline
  - 50% reduction in kidney function
  - End-stage kidney disease
- For the patient survey, we will involve 800 children with CKD and their caregivers who will respond to an electronic questionnaire

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IMPACT OF FINDINGS

- PRESERVE will be the largest study of pediatric CKD among children who have not yet reached end-stage kidney disease
- PRESERVE will address meaningful questions related to blood pressure management for youth with CKD and their clinicians
- The project will also create new PCORnet® infrastructure that can be used in future studies, such as clinical trials of BP control strategies in pediatric CKD

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THANK YOU!

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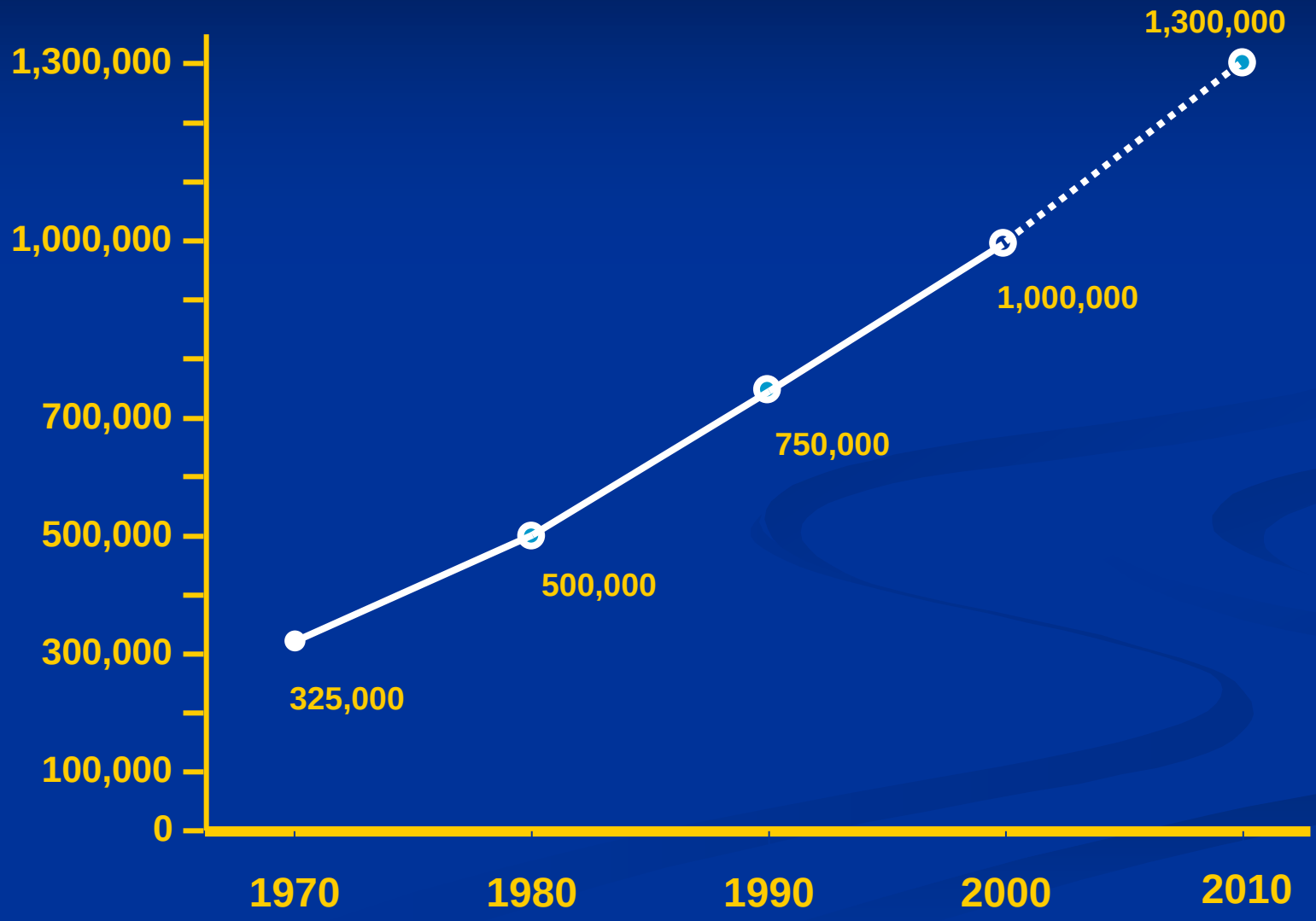
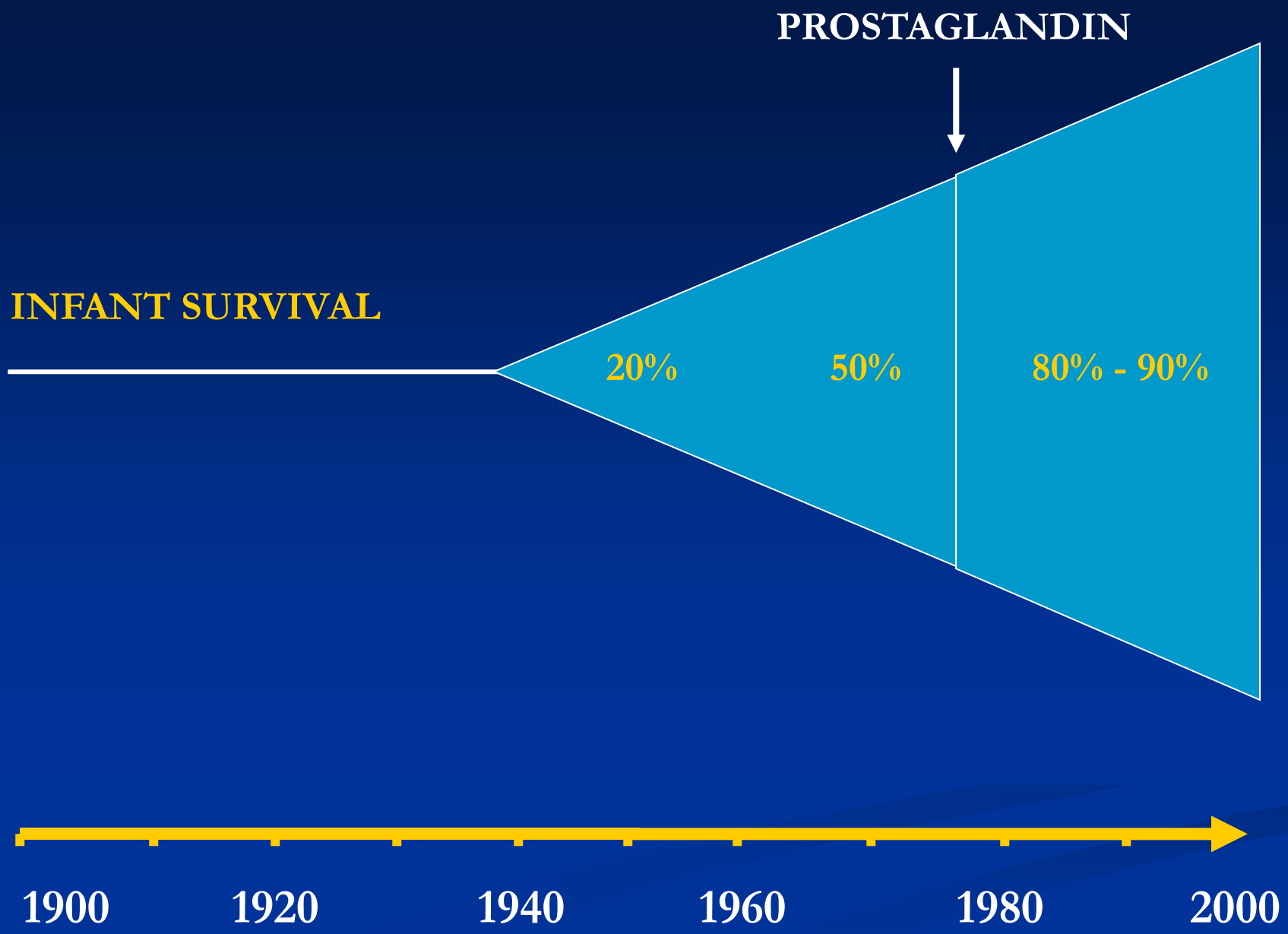
Utilizing PCORnet to support transition from pediatric to adult centered care and reduce gaps in recommended care in patients with congenital heart disease.

Dual PIs: Tom Carton (REACHnet) and
Anitha S. John (Children's National)

April 29, 2021

Number of Adults with CHD (US)




PROSTAGLANDIN
INFANT SURVIVAL
20%
50%
80% - 90%
1900
1920
1940
1960
1980
2000

Background: How did this all start?

- Congenital heart disease – most common birth defect
- Markedly increased survival – 1.5 million ACHD pts (US)
- Research challenges
  - Heterogenous set of defects (multiple rare diseases)
  - Poor rates of follow-up
  - No national registry for ACHD pts
- Heart Research Alliance/Health eHeart Study
  - CHD focused surveys (UCSF team)
  - ACHD cause group (ACHA representation)
  - Outreach to researchers (AARCC)

Children's National HeartInstitute

Project Aims/Goals

- **Specific Aim #1: Utilize PCORnet to establish an ACHD surveillance system** that will gather real-world data on healthcare use and comorbidities various CHD subtypes, and will test the hypothesis that gaps in care is associated with inappropriate healthcare use and higher comorbidities and varies with complexity of CHD subtypes.

- **Specific Aim #2: Determine factors associated with gaps in recommended care.** We hypothesize that various patient-level factors (such as severity of illness, patient demographics, place of residence, insurance status) might influence patients' likelihood of receiving recommended care. This data will characterize the factors associated with gaps in care that includes the vulnerable and disadvantaged segments of the US population.

- **Specific Aim #3: Determine the impact of gaps in care on patient reported outcomes.** By linkage of CHI Registry to PCORnet, the association of gaps in care and patient prioritized outcomes can be assessed over time for various types of CHD subtypes. We hypothesize that gaps in care is associated with lower quality of life and other outcome measures across all CHD subtypes.

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What are the eventual goals?

- Better assess needs and outcomes and potential risk factors to generate information on CHD patients who have been lost to cardiology follow-up
- Assess the impact of loss of follow-up on PROs
- Generate a registry platform that links EMR data from PCORNet to PRO platform
  - Goal: Create infrastructure for future CER in CHD

Children's National HeartInstitute


The journey of a thousand miles begins with a single step.
Lao Tzu

CER-NET Comparative Effectiveness Research for Neuroendocrine Tumors

Principal Investigator: Michael O'Rorke michael-ororke@uiowa.edu
Patient Advocate Lead: Josh Mailman josh@norcalcarcinet.org

Outline

1. **CER-NET: Background & significance**
2. **CER-NET: Overview**

1

CER-NET
The so what?

Background & significance

Why CER-NET?

- NETs relatively uncommon

- Predominantly small bowel, pancreas and lung

- Typically **slow-growing**, **prolonged survival**

  (>40% have metasatic disease at Δ)

- Incidence and prevelance are both increasing
- Ax of **HRQoL** is important but ***scarce!***
- Optimum **Rx sequencing unknown**
- Lung NETs & G3 NETs have limited datasets

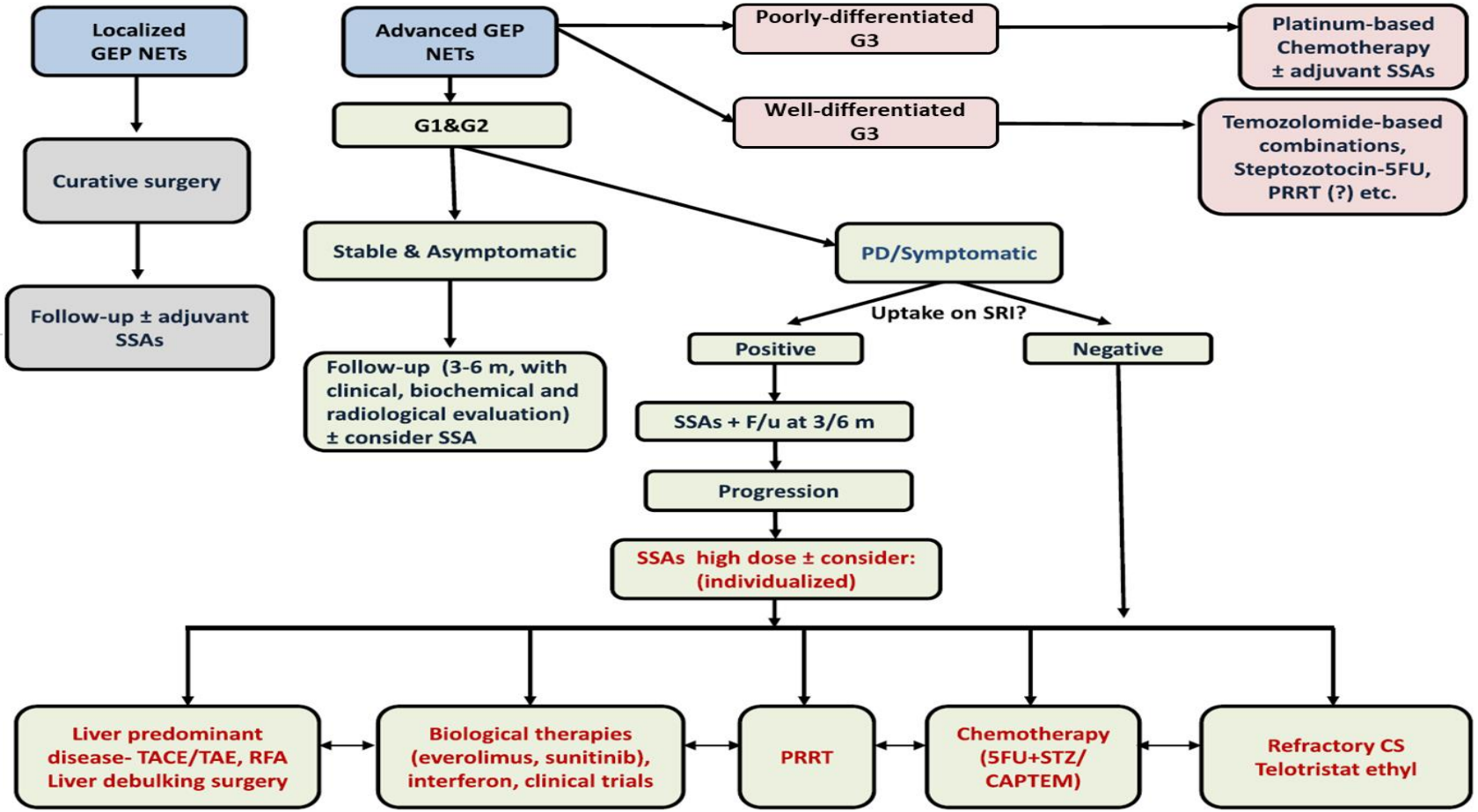
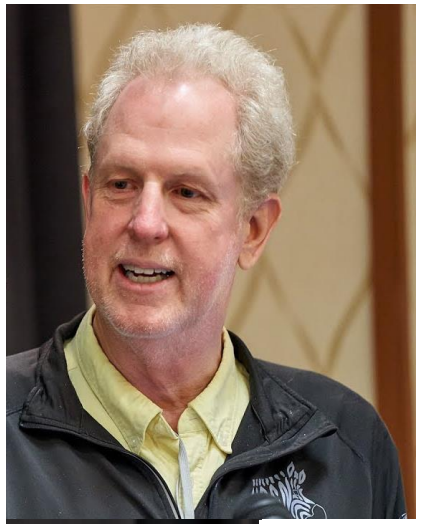


Figure 1: Overview of treatment algorithm for GEP-NETs

Somatostatin Analogues (SSAs), Peptide receptor radionuclide therapy (PRRT), 5-fluorouracil (5FU), Streptozocin-based chemotherapy (STZ), capecitabine with temozolomide (CAPTEM), somatostatin receptor imaging (SRI), Trans-arterial embolization (TAE), Trans-arterial chemoembolization (TACE), refractory carcinoid syndrome (CS), radiofrequency ablation (RFA).

‘What therapy would be best to try next?’

‘If I were to take this option now, what therapies will be cut off to me in the future?’



NorCal CarciNET
Strength in Community

2

Study Overview

CER-NET

CER-NET Specific Aims

Aim 1

Describing the frequency and sequencing of treatments & association with patient reported outcomes (PROs)

Aim 2

Whether patient, clinical and tumor characteristics impact on the choice of treatment – Assoc. with survival and disease progression

Aim 3

Effectiveness of PRRT (and Rx combinations) on outcomes of renal toxicity, disease progression and HRQoL

Aim 4

Sharing of the PCORnet infrastructure this study will generate (i.e.: NET phenotype, data curation scripts etc.)

CER-NET Study Synopsis

PROSPECTIVE Cohort Study

- Incident GEP/Lung NETs
- Δ 01/01/19 – 12/31/23
- 3,010 patients (215/site)

Study duration: 07/01/21 – 06/30/24 (36 mths study)

Max 60mths follow-up

Key Outcomes

- HRQoL
- Response rate (RECIST criteria)
- PFS/OS
- Creatinine Clearance loss per year

PaTH Network patient empowered research

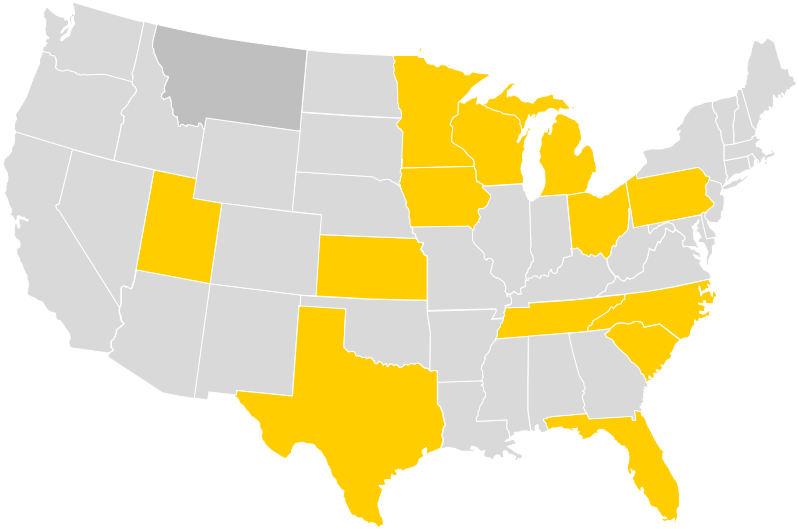
University of Michigan

PaTH Network patient empowered research

The Ohio State University

PaTH Network patient empowered research

UPMC University of Pittsburgh Medical Center



University of Iowa Coordinating Centre (UICC)

The University of Iowa | *Clinical Site*

OneFlorida Clinical Research Consortium

OneFlorida Clinical Research Consortium

Orlando Health

The University of North Carolina at Chapel Hill

STAR Stakeholders, Technology and Research CRN

UF University of Florida

STAR Stakeholders, Technology, and Research CRN

MUSC Medical University of South Carolina

UT Southwestern Medical Center

GPC Greater Plains Collaborative

KU Medical Center The University of Kansas

Vanderbilt University Medical Center

GPC Greater Plains Collaborative

The University of Utah

Medical College of Wisconsin

Allina Health

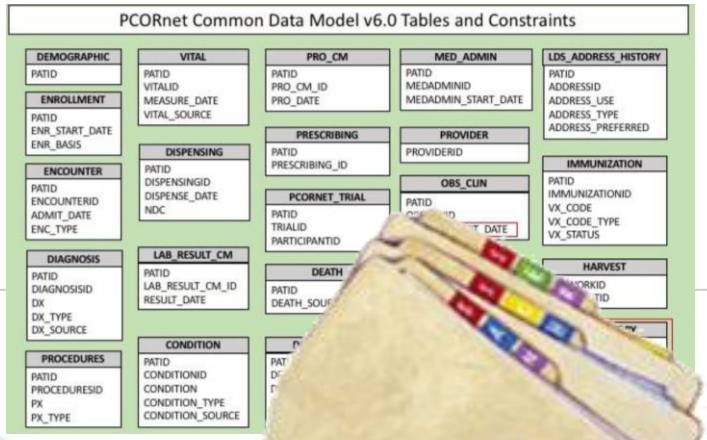
STAR Stakeholders, Technology and Research CRN

GPC Greater Plains Collaborative

GPC Greater Plains Collaborative

GPC Greater Plains Collaborative

CER-NET: Overview



EMR/
Chart review

Aims 1-3

ANALYSIS

1

NET Computable Phenotype (SAS query distribution)

Electronic Identification

2

NET Patients

1st eligibility screen
Initial Contact
Enrollment

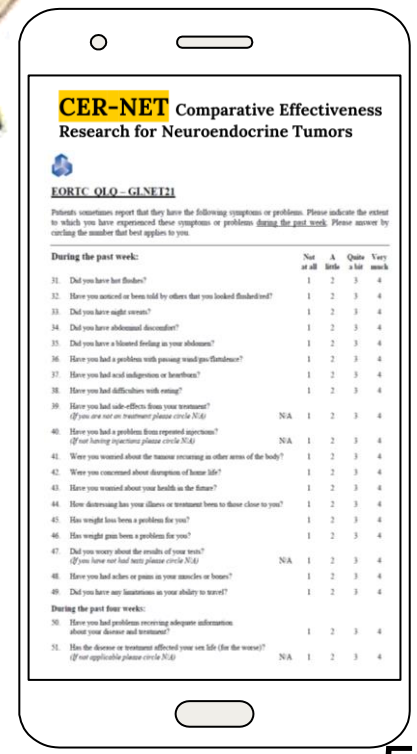
Clinical Sites

University of Iowa Coordinating Center (UICC)

THE UNIVERSITY OF IOWA

3

PHR
Patient Health Record



4

2nd eligibility screen
Onboarding
Consent
Baseline Survey ($)
Follow-up surveys (6, 12,18 mths)

NorCal CarciNET
Strength in Community

NET Foundation
Neuroendocrine Tumor Research Foundation

NANETS
Neuroendocrine Cancer Awareness Network

Healing net Foundation

Thanks!

Questions?

- **Principal Investigator:** Michael O'Rorke michael-ororke@uiowa.edu
- **Patient Advocate Lead:** Josh Mailman josh@norcalcarcinet.org

Improving Methods for Conducting Patient-Centered Outcomes Research

Jason Gerson, PhD
Senior Program Officer
Clinical Effectiveness and Decision Science

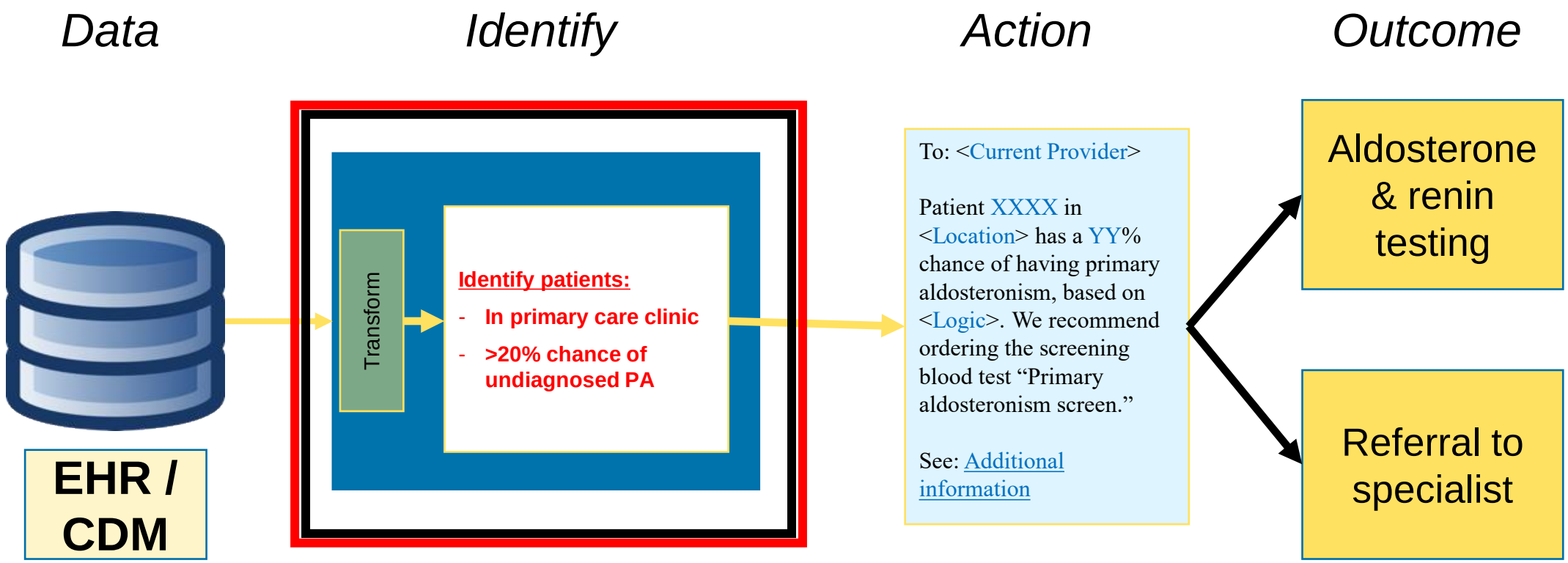
Development of methods to improve identification of patients with rare or complex diseases

Daniel S. Herman
Assistant Professor, Department of Pathology & Laboratory Medicine
Director of Endocrinology Laboratory
University of Pennsylvania

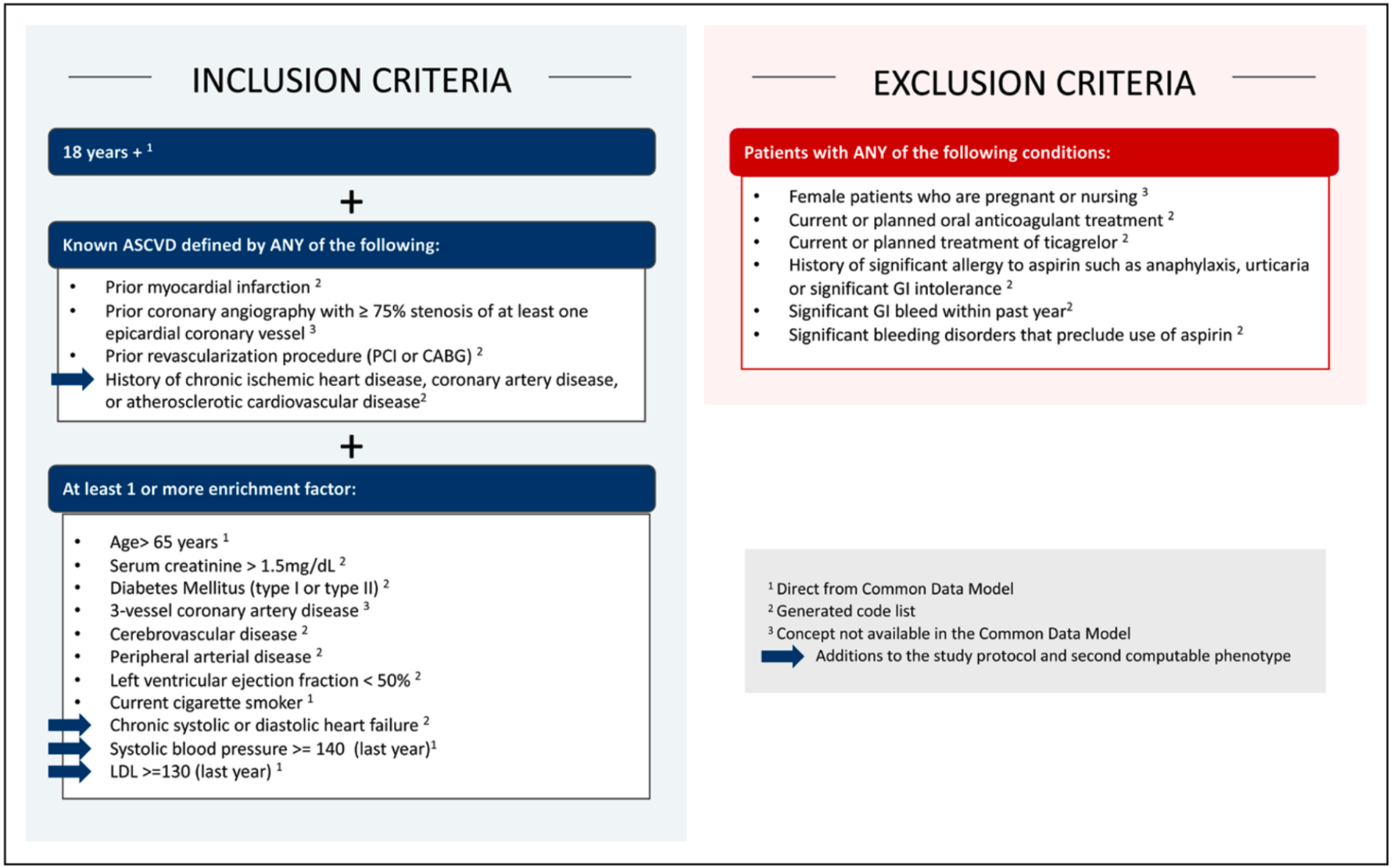
Penn Medicine

Who should we test? Who should we enroll?

Systematic targeted screening paradigm



Computable phenotype: Example



ADAPTABLE
Ahmad et al, 2020

Challenges in building computable phenotypes

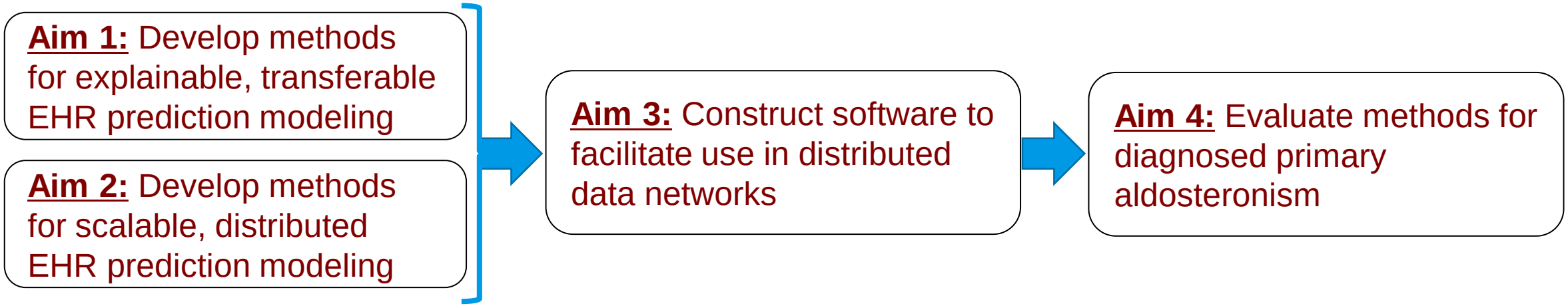
- **Labor- and time-intensive**
- **Clinical practice and documentation variability**
- **Data source variability**

Goals for methods and tools to support phenotyping:

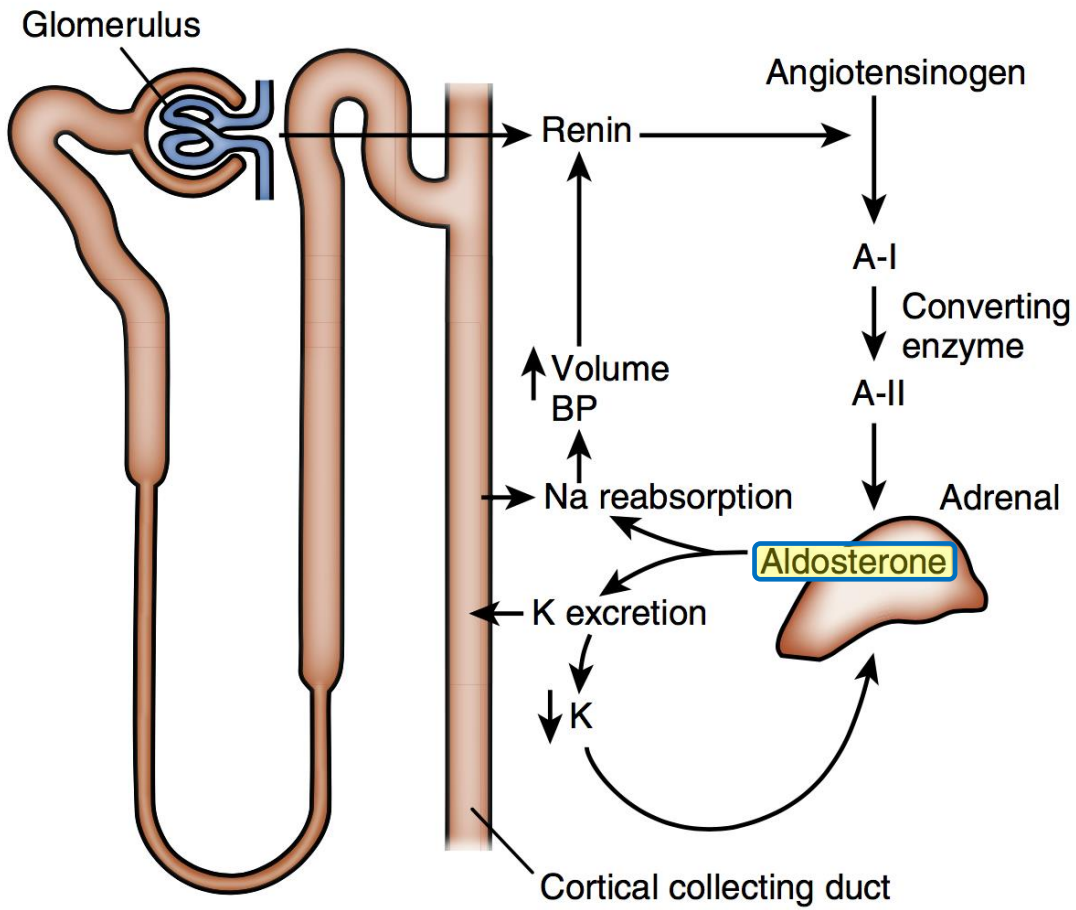
- Automation
- Accuracy
- Explainability (Interpretability)
- Site-specific flexibility
- Equity

Project objectives

Goal: Develop novel machine learning methods that leverage distributed clinical data to efficiently construct accurate, explainable, and equitable computable phenotypes



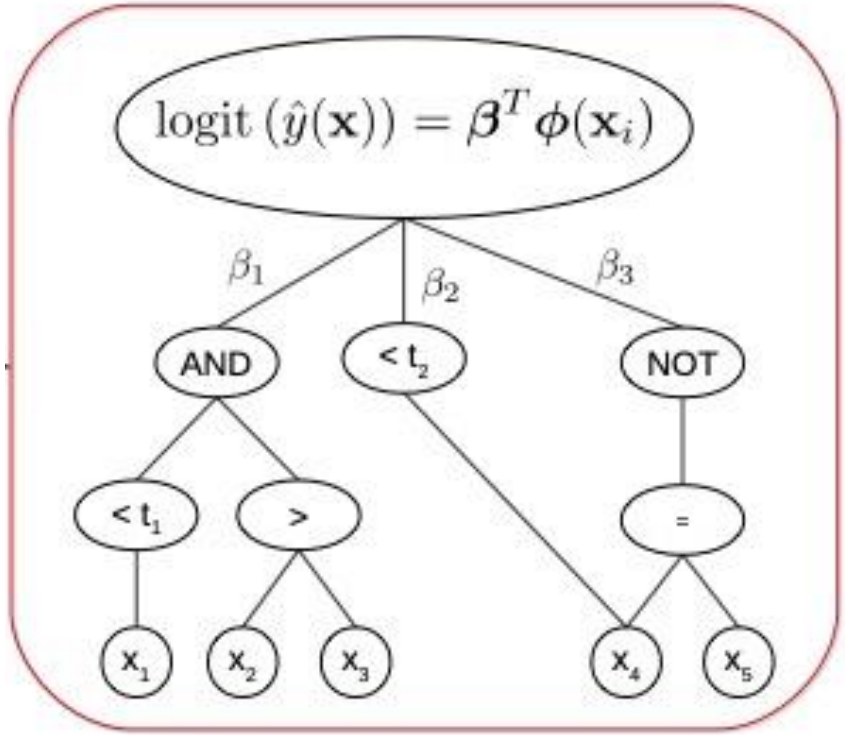
Why primary aldosteronism?



- **Underdiagnosed**
- **Excess morbidity & mortality**
- **Treatable** *(effectiveness data limited)*
- **Relevant data in PCORnet CDM**

Carey 2016; Fuller 2015

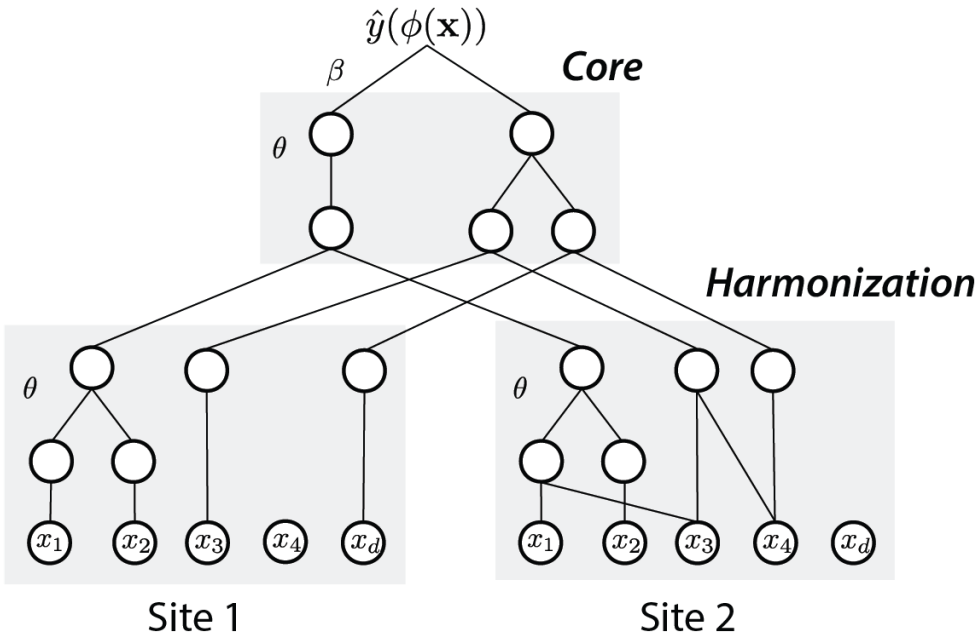
Construction of computable phenotypes with FEAT



La Cava, et. al., medRxiv, 2020

Approach for distributed learning

a



Acknowledgements

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William La Cava

Thaibinh Luong

Mike Draugelis

Jason Moore

Primary Aldo team

- Debbie Cohen
- Jordana Cohen
- Heather Wachtel
- Scott Trerotola
- Douglas Fraker
- Julia Kharlip

DORIS DUKE CHARITABLE FOUNDATION

Perelman School of Medicine University of Pennsylvania PATHOLOGY AND LABORATORY MEDICINE

Penn Medicine Predictive Healthcare

PENN CENTER for PRECISION MEDICINE

Thank you!

BREAK (10 minutes)

We will return at 2:30 pm EST

pcori®
PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

Eugene Washington PCORI Engagement Awards

Rare Disease Portfolio and Program Update

Karen Martin, Director Engagement Awards

At a glance

- **64** projects with a rare disease focus totaling **$9.7M**
  -  **26** Capacity Building
  -  **38** Stakeholder Convening Support
- **75%** patient led organizations
- **8** awardees with multiple projects

Examples of Primary Disease/Conditions

- 3q29 deletion
- Alagille Syndrome
- Alopecia Areata
- Alstrom Syndrome
- American Multiple Endocrine Neoplasia
- Aplastic Anemia & MDS
- Arterial Tortuosity Syndrome
- Beckwith-Wiedemann Spectrum
- Charcot-Marie-Tooth (CMT) & Inherited Peripheral Neuropathies (IPN)
- Cholangiocarcinoma
- Congenital Muscular Dystrophy
- Cystic Fibrosis
- Dravet Syndrome
- Ehlers-Danlos Syndrome
- Glut1 Deficiency
- Hemophilia
- Hepatoblastoma
- Hereditary Neuropathy
- Immune Thrombocytopenia
- Inborn Errors of Metabolism
- KIF1A
- Necrotizing enterocolitis
- Nemaline Myopathy
- Non-cystic fibrosis (CF) bronchiectasis
- Nontuberculous mycobacterium
- Osteogenesis Imperfecta
- Phelan-McDermid Syndrome
- Prader-Willi Syndrome
- Rare Neuromuscular Diseases
- Renovascular Hypertension
- Resective and Disconnective Pediatric Epilepsy Surgery
- Robin Sequence
- RUNX1-FPD
- Sex Chromosome Aneuploidy
- Sickle Cell Disease
- Sturge-Weber
- TANGO2-related disorder
- Turner Syndrome
- Vascular Ehlers-Danlos Syndrome
- Wilms Tumor WAGR Syndrome

Sample Project Activities and Deliverables

- Sample activities:
  - Build stakeholder networks
  - Establish PCOR/CER frameworks, including PCOR/CER agendas
  - Identify patient-driven research areas and priorities
  - Develop educational materials
- Sample deliverables:
  - PCOR/CER training tools and videos
  - Resources for successful meetings
  - Guides for developing informational materials on rare diseases
  - Roadmaps and guides for implementing and sustaining meaningful stakeholder engagement
- Full list of Engagement Award rare disease projects available here

Pediatric Renovascular Hypertension: A pRVH PCOR Collaborative

Project Objectives/Aims

- Assemble a broad collaborative of patients with and families affected by pRVH and stakeholders that are well informed; who will participate in identifying pRVH-related knowledge gaps that would benefit from PCOR and CER; and will guide the research prioritization.
- Create a pRVH PCOR Collaborative that includes an advisory board, a critical stakeholder group, and a broader virtual research network (VRN).
- Prioritize PCOR and CER priorities that consider critical diagnostic and treatment decisions.
- Conduct a pRVH PCOR Conference in 2022 that will disseminate the collaborative's interim work, share PCOR programming, and bring together stakeholders to formalize prioritized research plans.

Outcomes

- Assemble a large cohort of patients with pRVH and affected families that are well informed about pRVH and how to participate in PCOR and CER trials. The VRN will facilitate future patient activation for pragmatic clinical trials, which are very much needed for this rare disease process.

Dawn Coleman, MD
University of Michigan
Ann Arbor, MI
Project Period 2020-2022

Improving Patient-Centered Outcomes: Expanding Engagement of the Osteogenesis Imperfecta Community

Project Objectives/Aims

- Expand the osteogenesis imperfecta (OI) stakeholder community focused on performing PCOR.
- Expand existing OIF communication and education strategies; establish and extend capacity among the OI community to participate in PCOR activities.
- Develop an OI-specific PCOR toolkit and extend the OI patient-centered CER approach and products to support other rare bone disease communities.

COVID-19 Enhancement

- Ensure the OI community and the clinicians who treat them are aware of the evolving effects of COVID-19 as they seek to build capacity for PCOR/CER.
- Convene a panel of experts who will serve as a COVID-19 Task Force.
- Expand the focus of the PCOR training and toolkit to include COVID-19.

Outcomes

- Create a community of stakeholders—patients, caregivers, clinicians, researchers—trained or training in and committed to engaging in PCOR with specific attention to research topics that the OI community regards as high priority.

Tracy Hart, BS
Osteogenesis Imperfecta Foundation
Gaithersburg, MD
Project Period 2019-2021

Rare Disease and Research Engagement (RaRE)

Project Objectives

- Develop a Rare Disease Partnership Model and stakeholder-informed PCOR priorities at the intersection of rare disease and mental health.
- Build national network of diverse rare disease stakeholders interested in PCOR for rare diseases.
- Develop a comprehensive engagement model and governance structure to enhance collaborations among disparate rare disease communities for PCOR partnership.
- Identify cross-cutting patient-centered outcomes and research questions amenable to pragmatic CER studies.
- Develop a roadmap to implement and sustain robust stakeholder engagement in cross-cutting rare disease PCOR.

Outcomes

- Short-term: Establish opportunities for multi-stakeholder partnerships to support rare disease PCOR.
- Medium-term: Generate a Rare Disease Partnership Model and stakeholder-informed rare-disease PCOR priorities.
- Long-term: Researchers will implement cross-cutting rare disease PCOR with potential for broader generalizability and uptake.

Mathew Edick, PhD
Michigan Public Health Institute
Okemos, MI
Project Period 2020-2021

Awardee Feedback

"This was a transformative experience for us. The process of bringing together all stakeholders was very valuable and will help shape our efforts and activities going forward."

"PCORI Engagement Awards have been instrumental to our ability to establish a global network of families and scientists who prioritize patient-centered research. Our PCORI Engagement Awards have led to publications, quality improvement projects, webinar series, invitations to present and participate in other meetings, as well as secure funding from additional sources of support."

How PCORI has propelled the NEC Society's vision

Jennifer Canvasser, MSW

Founder, Executive Director

#preventNEC
@NECsociety
@jenncanvasser

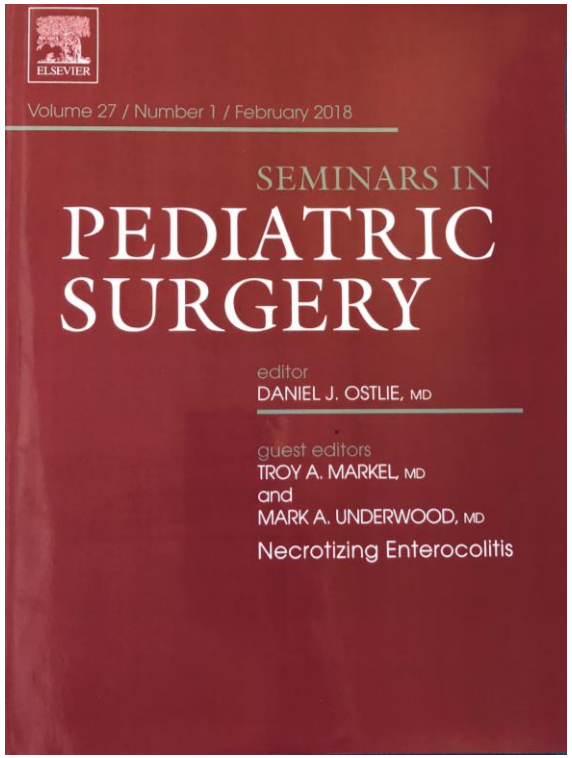
NEC SOCIETY
Building a World Without NEC

Quick intro & overview

NEC Society's Engagement Awards

- 2017: A Transdisciplinary Approach to Improve Outcomes ($50k conference support)
- 2019: Breaking Down Barriers to NEC Prevention & Treatment ($50k conference support)
- 2020: Building Capacity for PCOR/CER in the NEC Community ($250k capacity-building)

2017, in partnership with UC Davis



2019, in partnership with UMichigan

NEC family posters

Building capacity for PCOR/CER

- Skill building for patient-families & clinician-researchers
- NEC Research Priorities
- Complement CZI RAO Award
- Serving as a model for other rare neonatal disease communities

NEC SOCIETY VIRTUAL SESSIONS

MAY 19 - 21, 2021

12:00 - 1:30 EASTERN
EACH DAY

Join us for updates on the latest science, clinical practice, & connection with your NEC community!

Cincinnati Children's

NEC SOCIETY

REGISTRATION NOW OPEN!

www.NECsociety.org

PCORI projects → more opportunities!

- CZI Rare As One Network
- NICHD R13 Award
- Industry support
- International community

What's been helpful…

- Awareness
- Centering patient-families
- Recognition
- Community
- **Impact!**

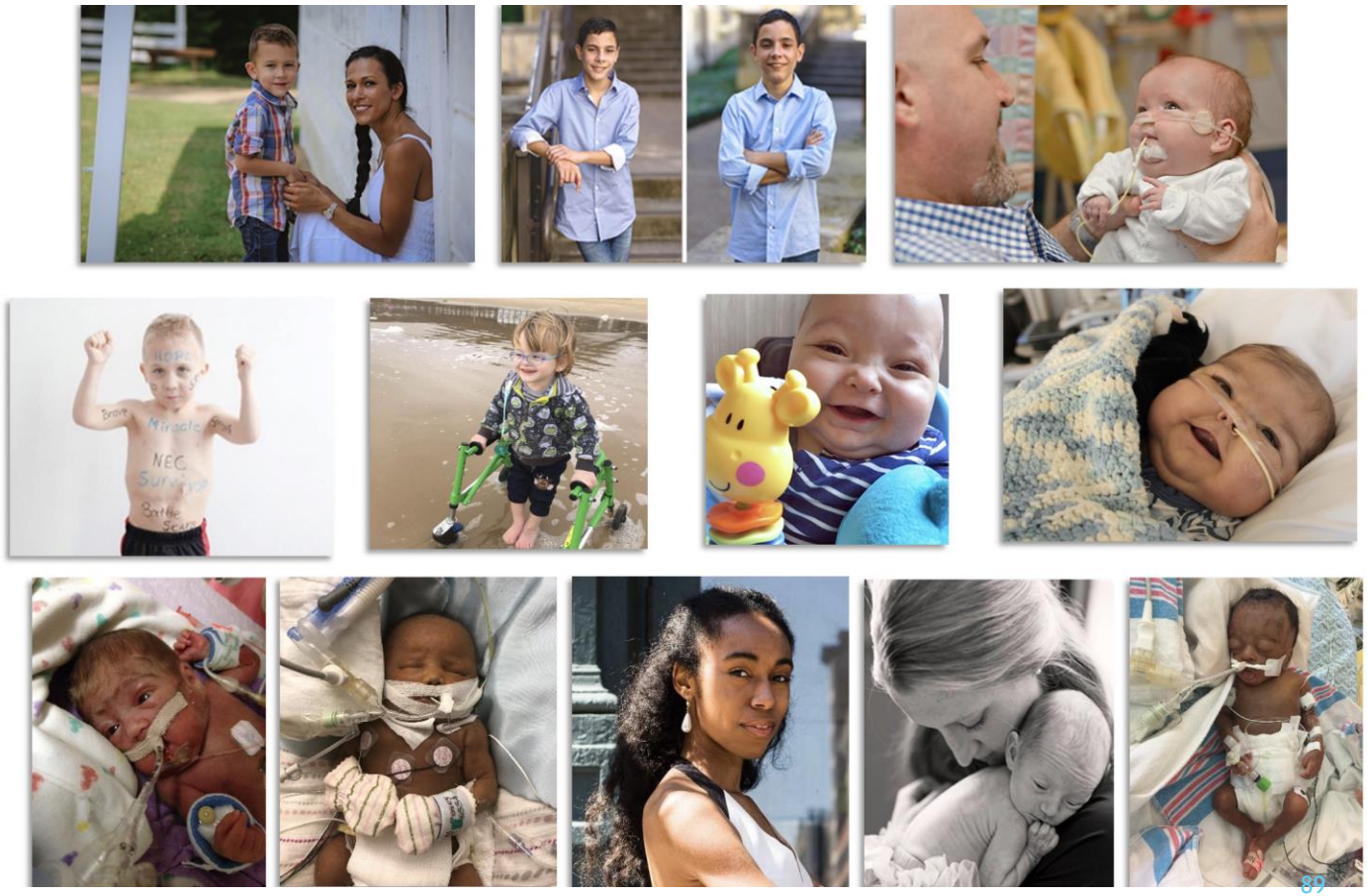
What's been challenging…

- Inadequate infrastructure
- Lack of long-term funding source
- Small team
- Finding the right fit
  (DEI values, $, time, commitment to excellence)

What's coming next…

- Established infrastructure
- Long-term funding
- Expanding & diversifying our team
- Patient-led research network
- Prepared to apply for a PCORI Research Award
- Progress towards **a world without NEC**!

Thank you, PCORI!



#preventNEC
@NECsociety
@jenncanvasser

NEC SOCIETY
Building a World Without NEC

Engagement Awards Program Updates

- **Stakeholder Interviews**
  - Elicit input from stakeholders with previous experience related to the PCORI Engagement Awards application and award lifecycle.
  - Assess challenges and other critical barriers that may have experienced during that process.
  - Identify potential solutions to ensure that the PCORI Engagement Awards are equitably accessible to all, including small organizations and investigators who are new to PCOR/CER.
- **Public Panel**
  - Broad themes from these conversations will be analyzed and shared on the PCORI web site for continued discussion among interviewees at a PCORI-hosted virtual public panel.

Identifying National Priorities for Health: Relevance for Rare Disease Populations

Laura Lyman Rodriguez, PhD.

Interim Chief Program Support Officer, Senior Advisor to the Executive Director

Reminder About Revised Strategic Framework
Evolving to National Priorites for Health

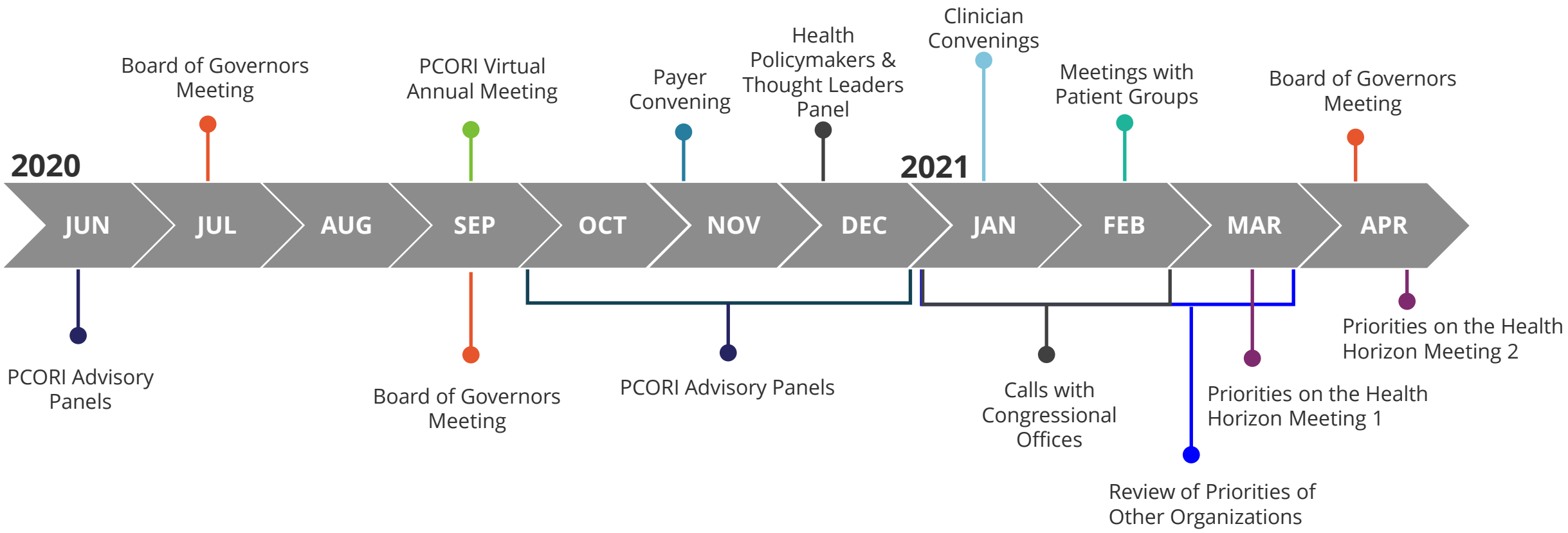
To inform the National Priorities for Health, PCORI sought input from a variety of stakeholders including this Advisory Panel. We'll take a look at the multifaceted inputs shortly.

Picking Up From When We Last Spoke

- This panel considered the revised framework and what the reframing meant for National Priorities.
- What we heard from RDAP members during the December meeting:
    - Refocusing from reducing disparities to health equity
    - Reflecting COVID-19 implications for health research (telehealth, mental and behavioral health)
    - Continuing to get research results and evidence to people who need it the most
    - Telehealth as a new, consistent option for routine and specialized care
- This was reinforced and complemented by input from other convenings, meetings, and discussions.

Input Gathering Process and Discussions Thus Far

- From June 2020 – April 2021, PCORI received input on the National Priorities from multi-stakeholder groups in addition to holding discussion at Board of Governors meetings and several other activities to help identify priorities



Resulting Themes from Input and Support by Board of Governors

- The themes below resulted from across the inputs
- At its April 2021, meeting, PCORI Board of Governors supported developing and further shaping these themes for National Priorities for Health

Health Equity

Emerging Innovations

Learning Health System

Communication, Dissemination, Implementation

Infrastructure & Workforce

Resulting Themes from Across Inputs and Summary Points for Importance

In the coming questions, we welcome your input on any of the themes and especially on the **Emerging Innovations, Learning Health System,** and **Infrastructure & Workforce** themes

Health Equity

- **Addressing disparities** is more important than ever
- **Systematic inequities** appear across health, healthcare, and health research (structural racism, implicit bias, lack of data representativeness)

Emerging Innovations

- Application of **new technologies** and **systems interventions** will be important for future of health; need to address evidence gaps
- Support **time sensitive decision-making** needs in evidence vacuum
- Inform **new delivery innovations** focused on patient-centered outcomes

Learning Health System

- Reframing transitional care from care settings to transition between health states to better **reflect patient perspective**
- Support health systems that enable **coordinated care, easy navigation** and **utilization** for patients

Communication, Dissemination, Implementation

- Importance of **doing communication and dissemination**, not just research on how to do it
- Get **right information to right people at the right time** to make informed decisions (e.g., patients, providers, health systems)

Infrastructure & Workforce

- **Workforce development** and capacity is needed to strengthen and expand the healthcare system
- Need for building **capacity for patient-centered outcomes research** (data, systems, researchers, patient partners)

We'd Like To Hear From You on How to Further Shape These

- Imagine 5 years from now, what will health, health care, and the research ecosystem look like if PCORI were to make progress towards a priority for health related to each theme? Are there specific areas that PCORI could make a real impact on in this timeframe?
- What specific areas within these themes are particularly well suited to comparative clinical effectiveness research?
- How could cross-cutting issues that impact all the priorities be considered (for example, health disparities or health equity)?

Emerging Innovations

Learning Health System

Infrastructure & Workforce

An Update on the Cost-Data Provision

Andrew Hu, MPP
Director, Public Policy and Government Relations

Overview of Reauthorizing Language

- PCORI's reauthorizing legislation directs PCORI to capture, **as appropriate**, the full range of outcomes data in the course of our research studies. This includes economic and cost data related to the utilization of health care services, but also patient centric-measures of cost and burden as well. Potential burdens and economic impacts include:

    - Medical out-of-pocket costs, including health plan benefit and formulary design, non-medical costs to patients and families, including caregiving, effects on future costs of care, workplace productivity and absenteeism, and healthcare utilization.

- PCORI is **still prohibited** from developing QALY measures and conducting cost-effectiveness analysis via our authorizing legislation.

Overview of PCORI's Implementation Plan

Pillar 1

- Providing guidance to Principal Investigators in future PFAs on how they should interpret this policy and incorporate it into their research proposals.
- **Timeline:** Final Principles and Initial Guidance for Applicants **by March 2021**

Pillar 2

- Advancing scientific methods in relation to the collection and consideration of burden and economic impact data.
- **Timeline:** Approximately **12 months** from the initiation of this process

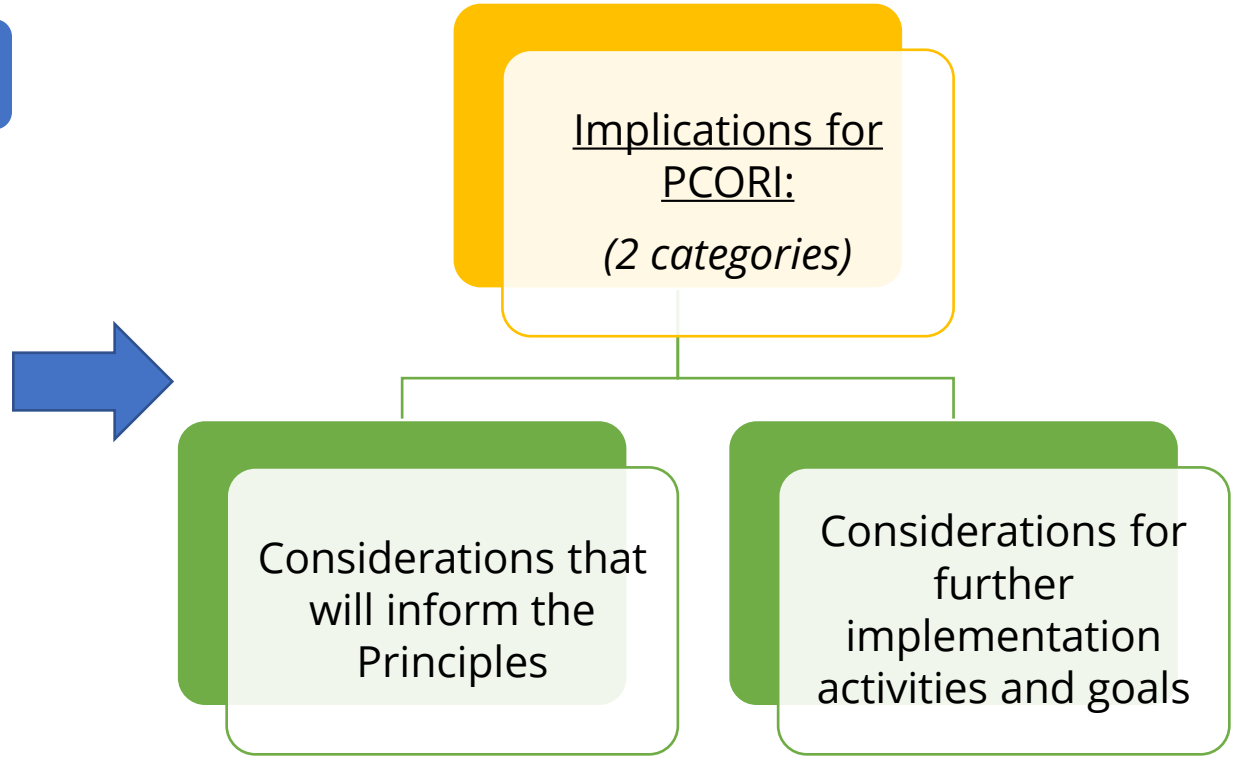
Pillar 3

- Supporting discussions related a patient-centered approach to addressing rising healthcare costs and value.
- **Timeline:** Ongoing Discussion

Overarching Themes and Potential Considerations

What PCORI Heard

- Ensure Patient-Centeredness
- Conduct Ongoing Stakeholder Engagement
- Burden and Economic Data Should Be Collected When Appropriate, But **Not** Required
- Provide Additional Guidance to Reflect the New Authority
- Identify Ways for PCORI to Inform/Advance Rigor and Standards When Considering Burden and Economic Data
- Consider Social Risk Factors



Principles for the Consideration of the Full Range of Outcomes Data in PCORI-Funded Research

Identifying Outcomes Important to Patients

- **Principle #1:** PCORI-funded research may consider the full range of outcomes *important to patients and caregivers,* including burdens and economic impacts.

Identifying Outcomes Important to Stakeholders

- **Principle #2:** PCORI-funded research may consider the full range of outcomes *relevant to other stakeholders,* when these outcomes have near-term or longer-term impact on patients.

Criteria for Data Collection Opportunities

- **Principle #3:** The collection of data on burdens and economic impacts of treatment options must be appropriate and relevant to the clinical aims of the study.

Consideration of Economic Analysis

- **Principle #4:** Beyond the collection of burden and economic impact data, PCORI may pursue more detailed economic analyses as part of a funded research study, to enhance the relevance and value of this information to health care decision-makers.

Additional Implementation Activities

In addition to comments on the Principles, PCORI received input outlining specific areas for the focus of the Institute's ongoing activities to implement this authority. Below are additional implementation activities PCORI can undertake.

Ensuring Patient-Centeredness	Ensuring Ongoing Patient and Stakeholder Engagement	Developing Guidance and Methodology Standards
Consider appropriate options to monitor how PCORI-funded research is used	Convenings to support direct engagement of stakeholders and researchers	Develop additional guidance to inform applicants
Build on and develop additional toolkits to support patient and caregiver engagement		Support Methodology Committee activities to advance scientific rigor
Advance and support discussions related to patient-centered approaches to rising healthcare costs		Conduct ongoing review and evaluation of Principles, guidance, and methods

BREAK (10 minutes)

We will return at 4:10 pm EST

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PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

COVID Connect:

PCORI's Response to the COVID-19 Epidemic

Claudia Grossmann, PhD

Senior Program Officer, Research Infrastructure

Co-Chair, COVID Connect

COVID Connect: Coordinating PCORI's COVID Response

Topic Identification

Stakeholder Engagement

Portfolio Analysis and Synthesis

- To develop a **mission-aligned strategy** for PCORI's rapid response to the COVID-19 pandemic informed by relevant staff, stakeholder, and portfolio input.
- To urgently **identify and develop topics**, relevant funding mechanisms, and evidence products that generate timely evidence.
- To **characterize the PCORI COVID-19 portfolio** and identify how PCORI can distinguish itself from other funders by filling gaps, expanding on existing research, enlarging outcomes measures, etc.
- To establish **relationships with other funders** to identify where PCORI can play a non-duplicative role in the funding landscape.
- To **translate knowledge and information** about PCORI's COVID-19 work to the broader public and relevant stakeholders in order to facilitate information exchange.
- To provide **cross-departmental coordination** for PCORI's COVID-19 response.

PCORI's COVID-19 Portfolio:

122 Enhancements, 9 Targeted Research Studies, and 25 Special Cycle Engagement Awards

122 Enhancements Awarded, *$34.8 million*

53
Engagement Award Enhancements
$7 million

13
D&I Enhancements
$5.8 million

47
Research Enhancements
$19.2 million

8
Methods Enhancements
$2.3 million

1
PCORnet Enhancement
$526,020

34 New Awards in Research & Engagement, *$33.5 million*

25
Engagement Award Special Cycle
$3.7 million

9
Targeted Research Studies (COVID tPFA)
$29.8 million

Information current as of April 22, 2021

COVID Enhancements to Existing Research Projects

PCORI funded **Fifty Enhancements to Research Awards** totaling $21.7 million

Focus of 41 CER enhancements

13 enhancements about COVID-19 as a condition

28 enhancements about providing care during a pandemic

Themes from 8 Methods Enhancements

2 Developing clinical prediction models

2 Informing COVID-19 care

Other themes: data visualization, machine learning

Note: studies may include more than one theme

One PCORnet Enhancement

"optimize and rigorously validate key COVID-19 data elements related to the treatment and outcomes associated with **COVID-19 coagulopathy**"

Key statistics about CER Enhancements:

Condition Categories	# of studies*
Mental & Behavioral Health	14
Nutritional & Metabolic	7
Neurological	6
Cancer	6
Cardiovascular	6

Priority Populations	# of studies*
Black, Indigenous, and People of Color	18
Low Income	12
Women	9
Older adults	9

Telehealth

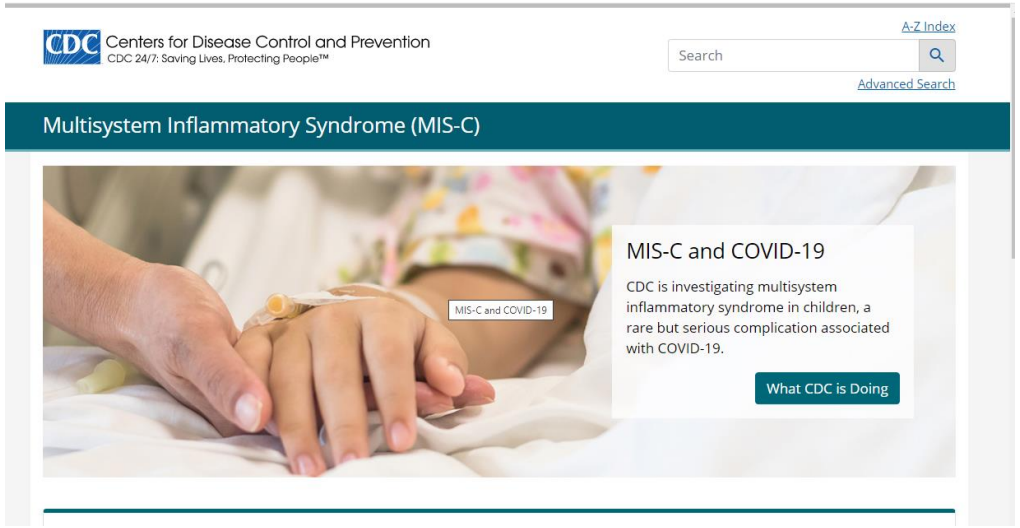
18 enhancements include telehealth components

*Studies may include more than one condition or population

Comparing Treatments for Resistant Kawasaki Disease – The KIDCARE Study

Project Summary

- PI: Jane Burns MD, University of California San Diego
- A randomized controlled trial comparing additional dose of intravenous immunoglobin (IVIG) or infliximab on cessation of fever in children with IVIG-resistant Kawasaki disease



COVID-19 Enhancement

- A new, rare health problem that can look like Kawasaki disease has appeared in children affected by COVID-19.
- With this enhancement, the team will describe the clinical features and collect blood samples from children with this new health problem and compare the clinical features to Kawasaki disease.
- To learn about children's history of exposure and initial symptoms, the parents will complete a questionnaire.

COVID-19 Targeted PFA

- PFA developed and posted on accelerated timeline in response to the urgency of the pandemic; accelerated Merit Review and programmatic review to ensure timely decision-making
- Priority Areas:
  - Adaptations to healthcare delivery
  - Impact of COVID-19 on disproportionately affected populations
  - Impact of COVID-19 on healthcare workforce well-being, management, and training
- PFA posted in May 2020; 9 awards announced in August 2020
- Studies up to 2 years in duration; actionable findings within first 12 months
- Small Studies: up to $2,500,000; Large Studies: up to $5,000,000

COVID Targeted PFA Research Projects

PCORI funded **9 Targeted COVID-19 Research Awards** totaling $29.8 million

Focus of Awards

5 awards focus on COVID-19 as a condition

4 awards focus on ways to provide care during a pandemic

Themes

3 targeted awards are relevant to nursing homes or other **congregate living settings**

Key statistics about targeted studies:

Primary Condition	# of studies*
COVID-19	5
Mental/Behavioral Health	3
Non-Disease Specific	1

*Studies may include more than one condition

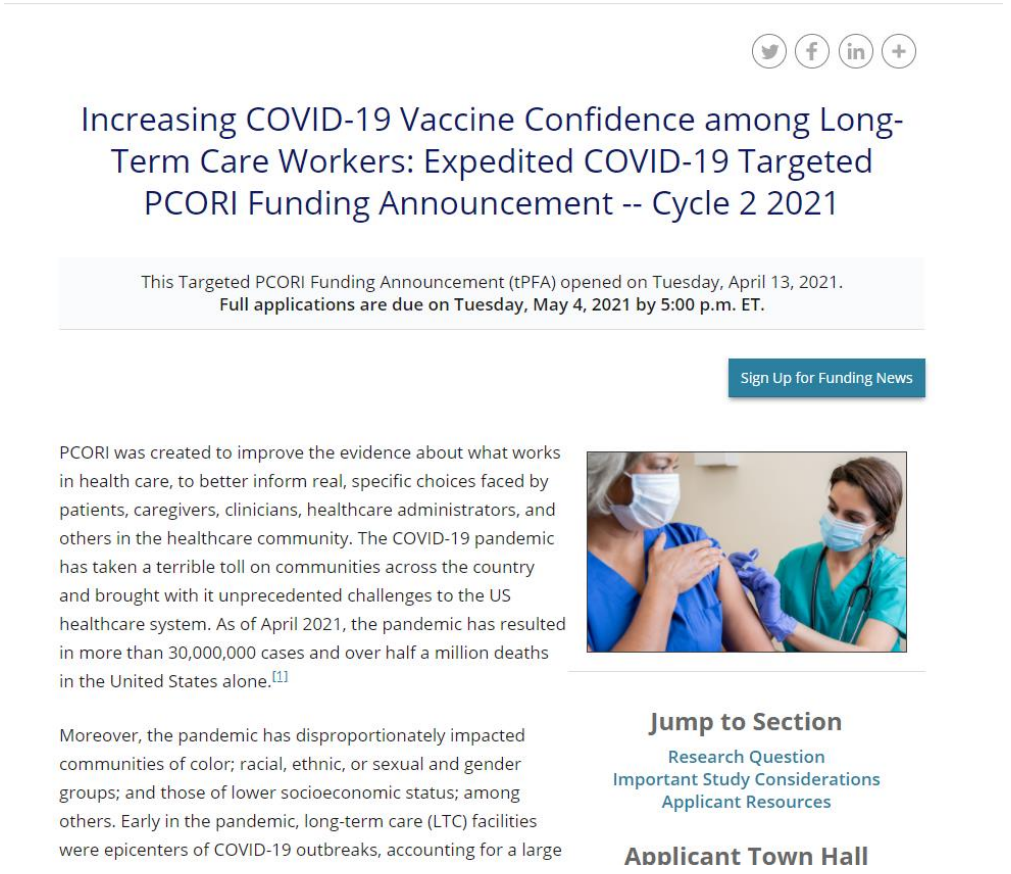
Priority Populations	# of studies*
Low Income	6
Black, Indigenous, and People of Color	5
People with MCC	3

Telehealth

5 targeted awards include telehealth components

Increasing Vaccine Confidence among Long-Term Care Workers PFA

- Using an expedited PFA mechanism; will follow more rapid review and award timeline
- PFA posted April 13, 2021; Awards Announced July 2021
- Up to 3 years
- Up to $5,000,000
- What interventions are effective in increasing COVID-19 vaccine confidence and uptake among LTC workers?



Increasing COVID-19 Vaccine Confidence among Long-Term Care Workers: Expedited COVID-19 Targeted PCORI Funding Announcement -- Cycle 2 2021

This Targeted PCORI Funding Announcement (tPFA) opened on Tuesday, April 13, 2021.
Full applications are due on Tuesday, May 4, 2021 by 5:00 p.m. ET.

Sign Up for Funding News

PCORI was created to improve the evidence about what works in health care, to better inform real, specific choices faced by patients, caregivers, clinicians, healthcare administrators, and others in the healthcare community. The COVID-19 pandemic has taken a terrible toll on communities across the country and brought with it unprecedented challenges to the US healthcare system. As of April 2021, the pandemic has resulted in more than 30,000,000 cases and over half a million deaths in the United States alone.[1]

Moreover, the pandemic has disproportionately impacted communities of color; racial, ethnic, or sexual and gender groups; and those of lower socioeconomic status; among others. Early in the pandemic, long-term care (LTC) facilities were epicenters of COVID-19 outbreaks, accounting for a large

Jump to Section
Research Question
Important Study Considerations
Applicant Resources

Applicant Town Hall

Broad PFA Special Area of Emphasis: Post-acute COVID-19

Cycle 1 2021

- 16 of 99 LOIs addressed COVID special areas of emphasis
- Management and survivorship of post-acute COVID-19
- Impact of COVID-19 on disproportionately affected populations
- Impact of COVID-19-related social isolation and loneliness on health outcomes

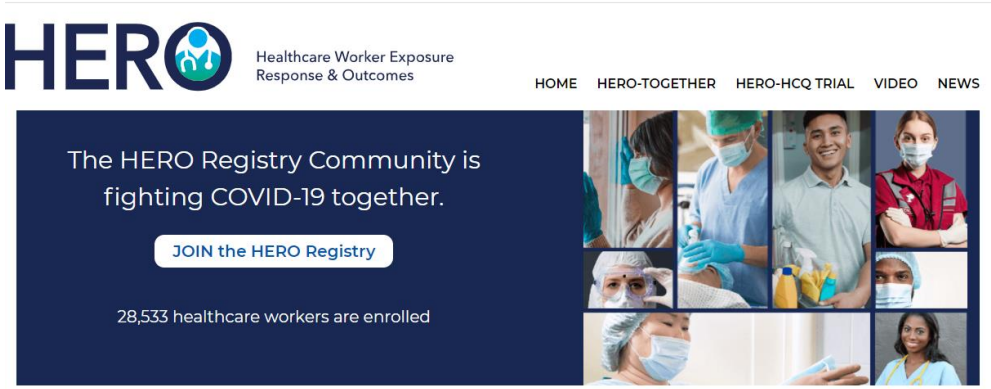
Cycle 2 2021

- LOIs due 6/1/21
- Treatment and survivorship of post-acute COVID-19
- Health system and healthcare delivery management of post-acute COVID-19
- Strategies to improve outcomes of COVID-19 for disproportionately affected populations
- Impact of COVID-19-related social isolation and loneliness on health outcomes

HERO Program

- HERO Health Care Worker Registry
  - >28,000 HCW enrolled as of April 2021
  - Addition of family members
  - HERO Together- Pfizer-funded study on long term vaccine side effects

- Hydroxychloroquine Trial
  - Completed Feb 2021
  - 1,363 enrolled
  - Expect manuscript submission shortly



ACTIV-6

- ACTIV-6 is a randomized, blinded, and placebo-controlled Phase III platform trial to test the efficacy of repurposed medications to treat COVID-19 in the outpatient setting.
  - 5-8 arms determined by existing ACTIV medication prioritization committee
  - Conducted under an IND
- PCORnet is leading ACTIV-6, serving as the Clinical Coordinating Center, Data Coordinating Center, and contributing approximately 40 vanguard sites.
  - Leverage HERO-HCQ experience
- Only ACTIV trial to include stakeholders in the trial governance
- To enroll 15,000 patients across estimated 80+ sites

Engagement Award
COVID-19 Enhancements

- Awarded on rolling basis starting in Spring 2020
- $150,000 total costs limit
- May not increase project timeline by more than 12 months
- Final Deliverables provided 6-12 months after funding

- 53 Enhancements to Engagement Awards ($7 million)

37	10	4	2
Capacity Building *$4.9 million*	Dissemination Initiative *$1.4 million*	PPRN Limited Competition *$540,625*	Conference Support *$93,515*

The STRETCH Project: To Build Capacity Advancing Patient-centered Research in Ehlers-Danlos Syndrome

Project Objectives

- Develop STRETCH Project training materials and curriculum targeted for delivery to clinician-patient research partnership sites.
- Engage individuals with the different EDS syndromes and symptoms as patient partners in order to capture the range of patients' experiences

COVID-19 Enhancement

- The enhancement leverages existing engagement infrastructure to address ways to alleviate the burden of COVID-19, as well as reducing the risk of morbidity, in the vulnerable Ehlers-Danlos Syndromes (EDS) population
- **Deliverables include reports describing research questions generated and the clinical practice implications that emerge from discussions with patients, clinicians, and other healthcare stakeholders.**

Outcomes

- The project is expected to significantly benefit people with EDS and provide a replicable model to expand capacity for patient-centered outcomes research and comparative clinical effectiveness research in other rare disease communities.

Jane Schubart PhD, MBA, MS
Pennsylvania State University Hershey Medical Center
Hershey, PA
Project Period 2018-2021

Engagement Award
COVID-19 Targeted PFAs

Summer 2020

- 25 Special Cycle COVID-19 Awards
- $3.7 million awarded June 2021

- Up to 12 months
- Up to $150,000 total costs
- Focused on methods of engagement to build capacity for PCOR/CER in the context of COVID-19

Fall 2021

- Up to 18 months
- Up to $200,000 total costs
- Focus on building capacity for stakeholder engagement in PCOR/CER specifically related to:
  - Long-term effects of post-acute COVID-19;
  - Impact of COVID-19 on disproportionately affected populations;
  - Impact of COVID-19 on social isolation and loneliness
  - Engaging, educating, and promoting informed decision making around COVID-19 vaccines

Dissemination and Implementation
COVID Enhancements

13 Enhancements to D&I Awards ($6.1 million) will adapt interventions for remote delivery, update materials, and increase reach

8 D&I enhancements have added new remote interventions – or remote delivery of interventions – including via an EHR system or patient portal.

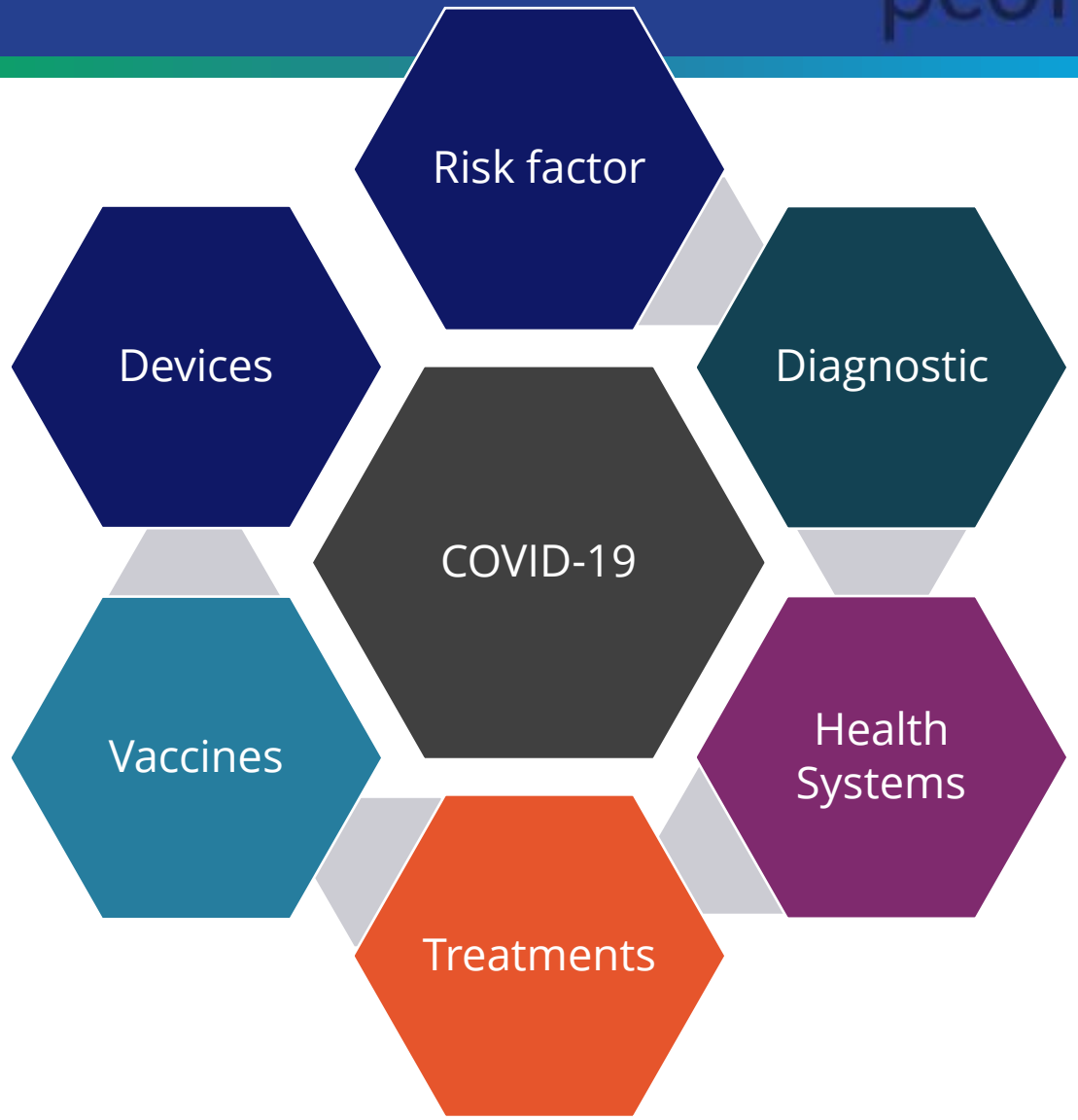
7 D&I enhancements are adding new content for patients, including educational and support materials addressing challenges associated with COVID.

>40k

9 D&I enhancements are increasing their reach, delivering interventions to more than 40,000 additional patients and caregivers.

Horizon Scanning
COVID-19 Supplement

- COVID-19 six priority issues
  - Devices
  - Prognostic/Risk factor
  - Diagnostic
  - Health Systems
  - Treatments
  - Vaccines
- Bi-weekly Scan (COVID-19 Only)
- COVID-19 Status Report
- COVID-19 High Impact Report



COVID-19 & the Future of Telehealth

PCORI Virtual Hill Briefing

- Hosted on Tuesday, March 23, 2021 to an audience of over 200 attendees from across the health policy spectrum
- Featured remarks from **U.S. Senator Tina Smith (D-MN)** and presentations from **Ray Dorsey, MD, MBA (**University of Rochester Medical Center), **Maureen Hensley-Quinn, MPA (**National Academy for State Health Policy), and **Penny Mohr, MA** (PCORI)
- Key takeaways include:
    - Telehealth can produce positive outcomes regarding quality of care, patient preference and convenience, and patient comfort with care.
    - Telehealth can expand access to care, particularly among populations experiencing limited access.
    - Telehealth can lessen or exacerbate disparities in care access and outcomes, and it depends on how it is used.
    - There is strong support for ongoing coverage of telehealth services to continue post-pandemic.

Discussion

- Are there other activities we should consider?
- Are there specific topics related to rare disease that you would consider a priority for research?

Acknowledgments and Recap

Recognition of Departing Panel Members

Thank you!

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

- Roxanna Bendixen
- Vanessa Boulanger
- Julie Gortze
- Tilicia Mayo-Gamble
- Sherene Shalhub
- Scott Berns

Panelist Recognition: Roxanna Bendixen

- Rehabilitation Scientist and Associate Professor at the University of Pittsburgh, Department of Occupational Therapy, School of Health and Rehabilitation Sciences.
- Bendixen has served on numerous rare-disease work groups, such as the American Academy of Neurology Muscular Dystrophy Measure Development Work Group and has provided expert scientific review for clinical guidelines in both Duchenne muscular dystrophy and congenital muscular dystrophies
- Contributions at the RDAP meetings: Raised the concern about transition of care at adulthood for pediatric onset diagnoses, suggested a focus on IDD for the rare disease community, highlighted the cross-cutting topic of sleep and fatigue

Panelist Recognition: Vanessa Boulanger

- Director of Strategic Partnerships at the Amyloidosis Research Consortium
- Prior to joining ARC, Boulanger was the Director of Research at the National Organization for Rare Disorders (NORD), where she led the strategic development, growth, and implementation of NORD's research and scientific activities, developing evidence-informed programs to support the rare disease community.
- Contributions at RDAP meetings: Provided input on evaluating and validating core clinical data outcome measures, shared insights on the topic of maternal health

Panelist Recognition: Julie Gortze

- Registered nurse and Founder of Rare New England, a nonprofit patient advocacy organization focused on improving the lives of those living and working with rare and complex disorders.
- A member of the Regional Genetics Network Steering Committee, Global Genes Alliance, and North Attleboro Commission on Disability.
- She is a 2017 Health Resources and Services Administration Regional Genetics Collaborative Advocate Leaders Partnership Program Recipient
- Contributions at RDAP meetings: Shared her 18-year diagnostic odyssey as a patient with a rare disease, provided input on raising awareness about PCORI

Panelist Recognition: Tilicia Mayo-Gamble

- Assistant Professor for the Department of Health Policy and Community Health at Georgia Southern University.
- She conducts research on community-engaged approaches to improving protective health behaviors in patients with one or more chronic conditions.
- Spouse to a husband with sickle cell disease. She understands the difficulties of providing input to healthcare providers and researchers about personal health.
- Contributions at RDAP meetings: Advocated for more rare disease merit review stakeholders, raised the issue of access to care for rare disease patients in remote/rural communities, emphasized the importance of engaging patients in dissemination of research findings, shared her experience working on an Engagement Award with the sickle cell community

Panelist Recognition: Sherene Shalhub

- Vascular surgeon and translational researcher at the University of Washington (UW) and the Director of its Multidisciplinary Vascular Genetics Clinic
- Her work aims to provide high-quality care to patients suffering from rare vascular conditions and to improve the success of surgical repair and long-term survival for these high-risk patients.
- Contributions at RDAP meetings: Shared her experience working on a Capacity Building award, noted the value of tele health for rare disease patients, highlighted issues with the healthcare delivery system

Panelist Recognition: Scott Berns

- Co-Chair of the PCORI Rare Disease Advisory Panel (RDAP) – 2019 to 2021
- President and CEO of NICHQ (National Institute for Children's Health Quality)
- Co-founder of The Progeria Research Foundation (PRF) and Chair of the organization's Board
- The U.S. Food and Drug Administration (FDA) approved Zokinvy™ (lonafarnib), for the treatment of Progeria and processing-deficient Progeroid Laminopathies (PL). PRF, a pioneer in the rare disease research foundation space, has led Zokinvy clinical trial research since 2007.
- At PCORI's fifth Annual Meeting in September 2019, Berns and his wife Leslie Gordon, MD, PhD, presented the Opening Keynote.
- Contributions at RDAP meetings: Recommended for more collaboration within the rare disease community, input on PCORI's national priorities, the topics of MMM and health equity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Thank You!

Adjourn