

Advisory Panel on Rare Disease (RDAP) Virtual Meeting: Spring 2020

June 22, 2020

Scott Berns, MD, MPH
Chair, RDAP

Cindy Luxhoj, MUP
Co-Chair, RDAP

Nora McGhee, PhD
Senior Program Officer, HDDR
Staff Co-Chair, RDAP

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

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



RDAP Chairs



Scott Berns, MD, MPH

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



Cindy Luxhoj, MUP

Co-Chair, Advisory Panel on Rare Disease
Founder and former Executive Director,
Alagille Syndrome Alliance

Housekeeping

- Please note that today's webinar is being recorded for posting on PCORI's website. The recording will be available to the public after the event.
- 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 2020 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 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.

6/22 Virtual Meeting Agenda

Start Time	Agenda Items	Discussion Leader
1:00 PM	Welcome and Setting the Stage, RDAP Panel Introductions	Scott Berns Cindy Luxhoj
1:20 PM	Introduction of RDAP PCORI Members and PCORI Update	Carly Khan Nora McGhee
1:25 PM	Introduction to Dr. Cook	Nakela Cook
2:10 PM	BREAK (10 minutes)	
2:20 PM	National Priorities and Research Agenda	Michele Orza Greg Martin
3:05 PM	Overview of Reauthorization and Cost Data Provision	Jean Slutsky Andrew Hu
3:55 PM	BREAK (10 minutes)	

6/22 Virtual Meeting Agenda (continued)

Start Time	Agenda Items	Discussion Leader
4:05 PM	Maternal Mortality, Intellectual Disabilities, and Balancing Short- and Long-Term Priorities	Jean Slutsky Kristin Carman Elisabeth Houtsmuller Stanley Ip Caitlin McCormick
4:40 PM	Recognition of Departing Panel Members	Scott Berns
4:50 PM	Acknowledgments and Recap	Scott Berns, Cindy Luxhoj
5:00 PM	Adjourn	

RDAP Panelist Introductions

Introductions

Please briefly state the following:

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

RDAP Members



CLINICIANS

[Scott Berns](#) (Chair)

- CEO, National Institute for Children's Health Quality

[Sherene Shalhub](#) (University of Washington)

RESEARCHERS

[Roxanna Bendixen](#) (University of Pittsburgh)

[Stephen Mathai](#) (Johns Hopkins University)

POLICY MAKERS

[Saira Sultan](#) (CEO, Connect4Strategies)

INDUSTRY

[Marcia Rupnow](#) (GSK)

PATIENTS, CAREGIVERS, AND PATIENT ADVOCATES

[Cindy Luxhoj](#) (Co-Chair)

- Founder & Former Executive Director, Alagille Syndrome Alliance

[Julie Abramson](#) (Hennepin County)

[Vanessa Boulanger](#) (NORD)

[Julie Gortze](#) (Rare New England)

[Doug Lindsay](#) (Personal Medical Consultant)

[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

RDAP Program Staff Update

PCORnet® Limited Competition Funding Announcement: Rare Disease Research



- RDAP members engaged in discussion at the September 2019 meeting about the potential for rare disease research using PCORnet®
- Feedback was incorporated into the Cycle 2 2020 Funding Announcement: Conducting Rare Disease Research Using PCORnet®
- PCORI has received the Letters of Intent for this call and full applications will be invited by the end of June
 - Applications are due September 2020
 - Awards announced March 2021

Upcoming Activities

- PCORI staff will present at the National Organization for Rare Disorders (NORD) Living Rare, Living Stronger Forum (virtual July 18-19) during a session titled, "Current Trends & Opportunities in Rare Disease Research."
- The PCORI Annual Meeting will feature a breakout session focused on Rare Disease research

Introduction to Dr. Nakela Cook

Dr. Nakela Cook, PCORI Executive Director



The background image shows two silhouetted figures walking across a series of dark, flat stepping stones in a calm body of water. The sun is low on the horizon, creating a bright orange and yellow glow that reflects on the water's surface. The overall mood is peaceful and hopeful, symbolizing a journey forward.

Driving Toward Health Impact: Our Journey and Path Forward

Rare Diseases Advisory Panel (RDAP)
June 22, 2020

Nakela Cook, MD, MPH
Executive Director
Patient-Centered Outcomes
Research Institute

Our Discussion Today

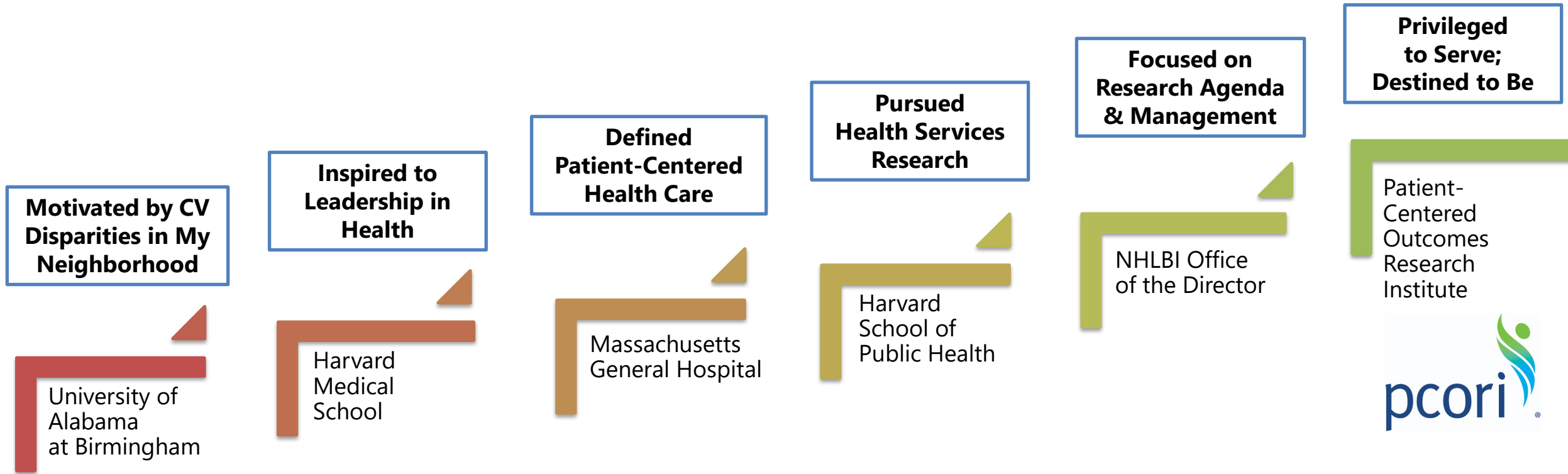
- My journey to PCORI
- Evolving healthcare landscape
- Health disparities and COVID-19
- PCORI 2.0



My Journey to PCORI



Fulfilling a Life's Purpose



At the Heart of PCORI

- Dedicated and mission-driven Staff, Board of Governors, Committees, Panels
- Committed stakeholders: Patients, Caregivers, Healthcare Community



Rare Diseases Advisory Panel Accomplishments

- Developed Rare Disease Research Guide for Merit Reviewers
- Provided input on PCORI's Guidance on Research in Rare Diseases
- Issued recommendations on developing core outcome set for Pediatric Rare Disease research
- Developed priority areas for conducting Rare Disease Research using PCORnet



Rare Diseases Advisory Panel Accomplishments

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Overcoming Challenges



“Courage is simply doing whatever is needed in pursuit of the vision.”

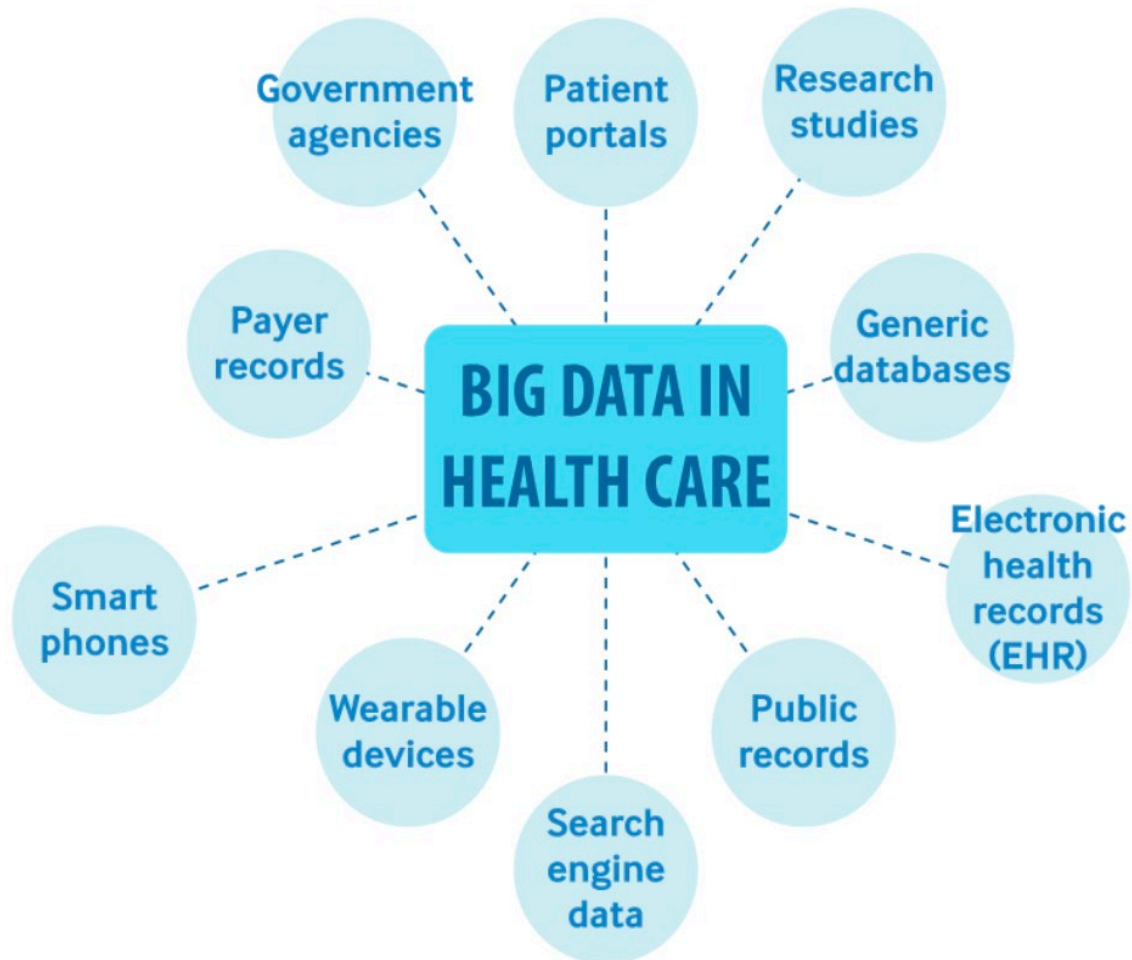
— *Peter M. Senge, The Fifth Discipline: The Art and Practice of The Learning Organization*

Evolving Healthcare Landscape



Era of Big Data

Sources of Big Data in Healthcare



Applications for Big Data in Healthcare



Leveraging Innovation to Deliver Improved Outcomes

Digital health



Big data



Precision medicine

Despite Improvements in Health, Disparities and Variation in Care Remain

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Special Communication

November 26, 2019

Life Expectancy and Mortality Rates in the United States, 1959-2017

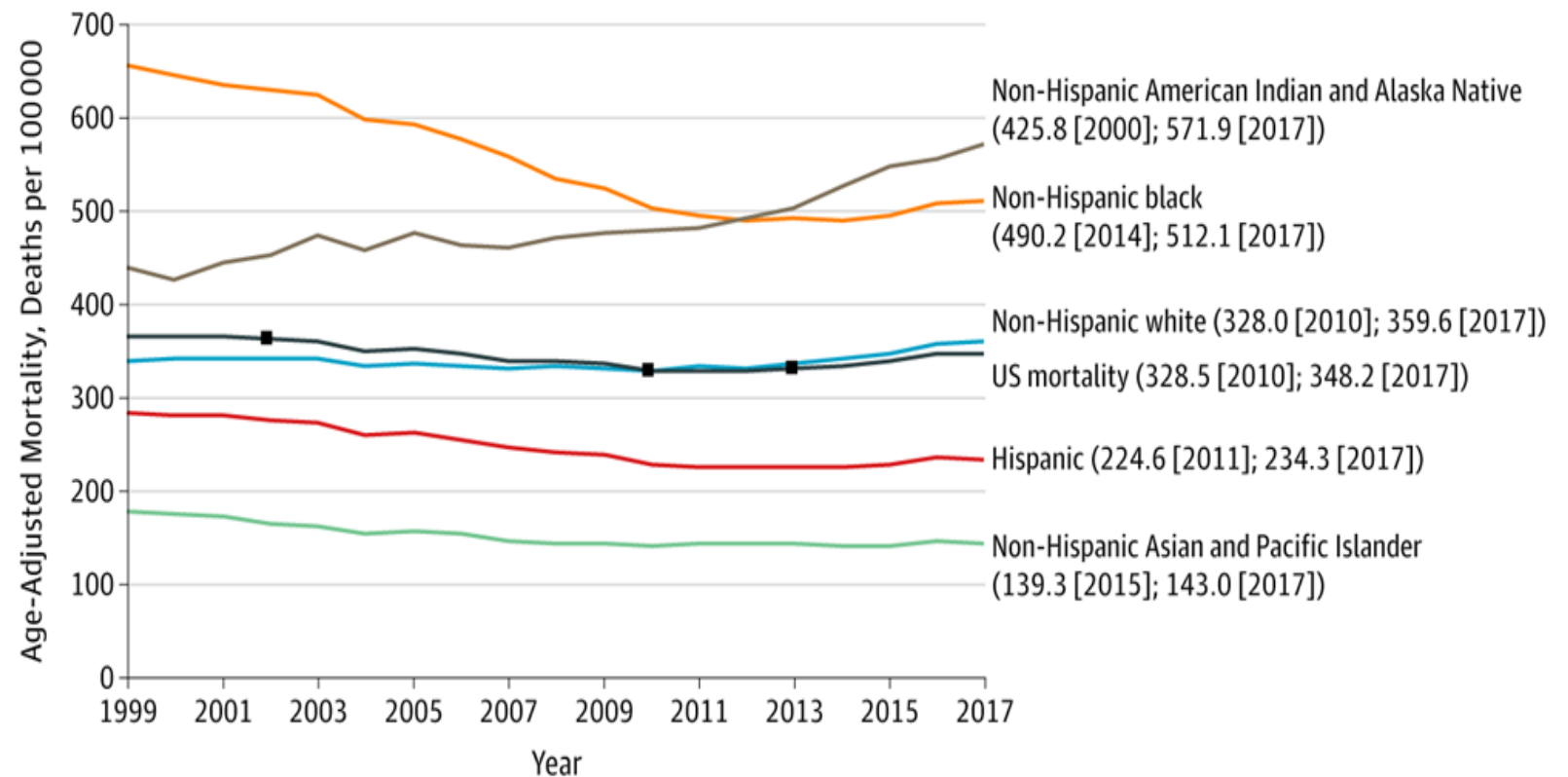
Steven H. Woolf, MD, MPH¹; Heidi Schoomaker, MAEd^{2,3}

► Author Affiliations

JAMA. 2019;322(20):1996-2016. doi:10.1001/jama.2019.16932

Black curve indicates age-adjusted mortality for all US adults aged 25 to 64 years; bolded data points indicate joinpoint years, when the linear trend (slope) changed significantly based on joinpoint analysis. The lowest mortality rates per 100 000 (and the years they were achieved) are listed first in parentheses; mortality rates for 2017 listed second.

Age-Adjusted Mortality Rates, US Adults Aged 25-64 Years, by Race/Ethnicity, 1999-2017



Source: CDC WONDER.

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<https://jamanetwork.com/journals/jama/article-abstract/2756187>

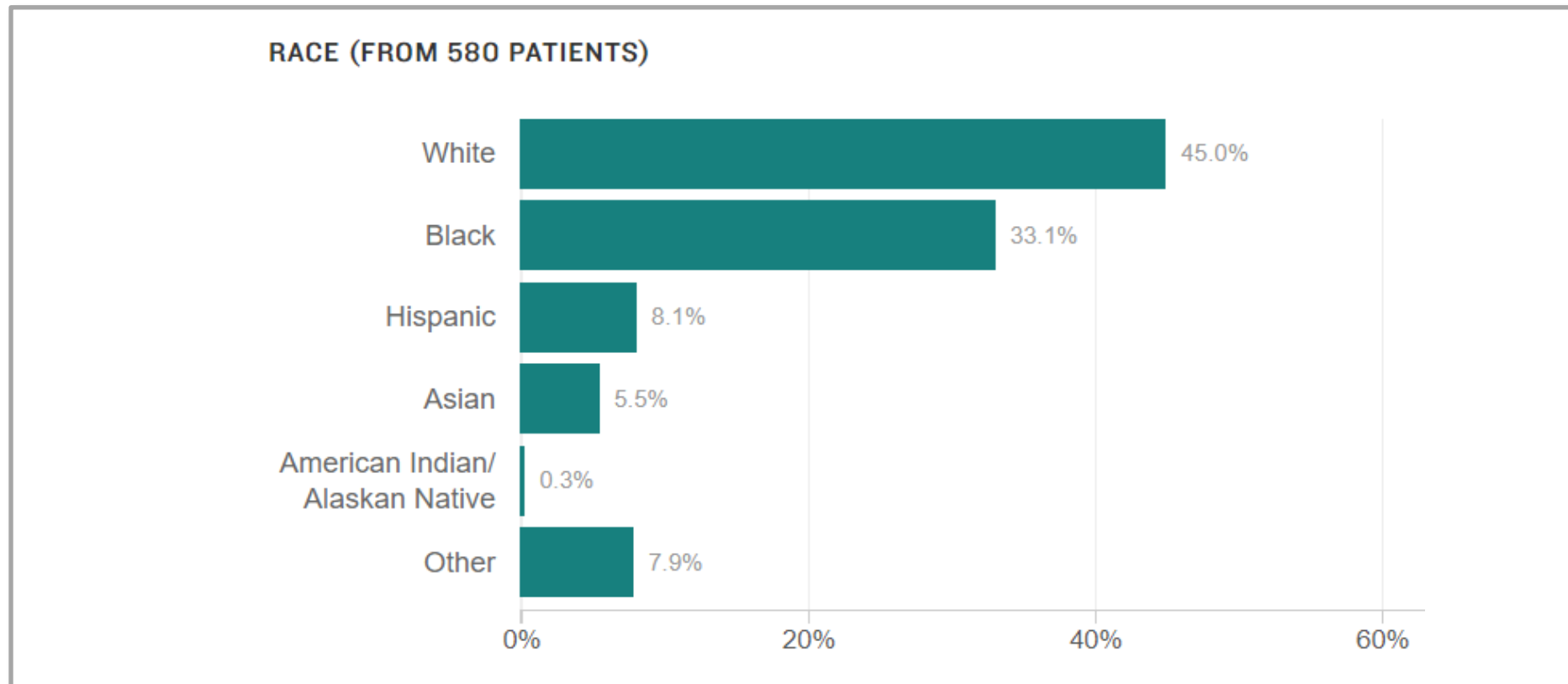


CORONAVIRUS USA

**Health disparities
and COVID-19**

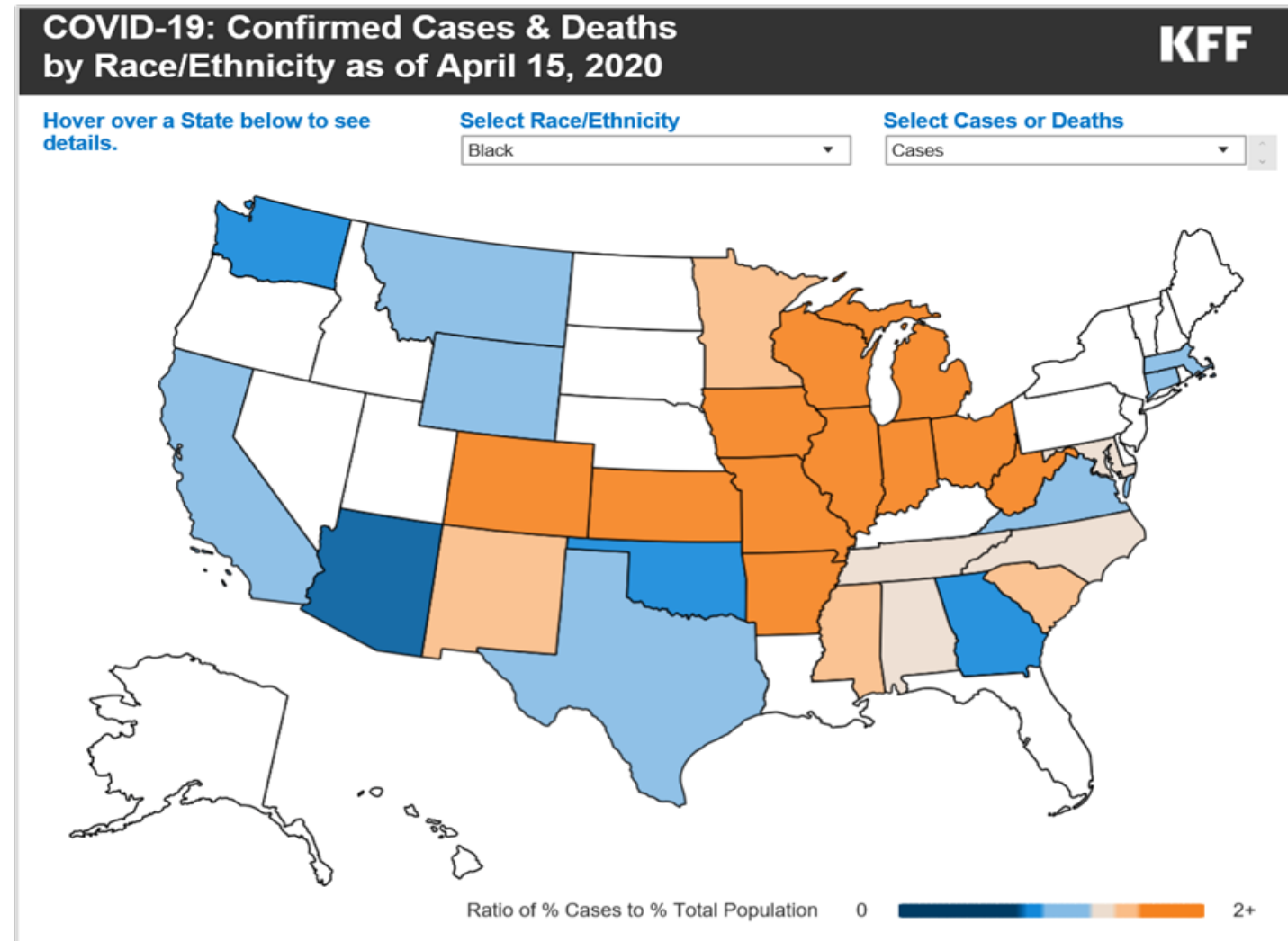
COVID-19 Outcomes Elucidate the Challenge of Health Disparities

Differences in COVID-19-related hospitalization by race/ethnicity



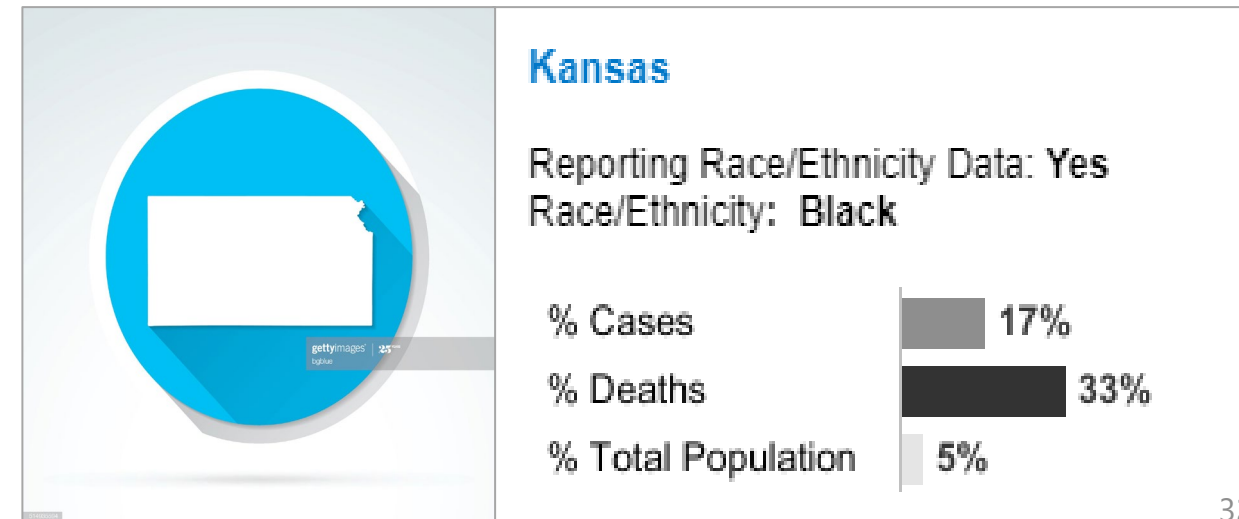
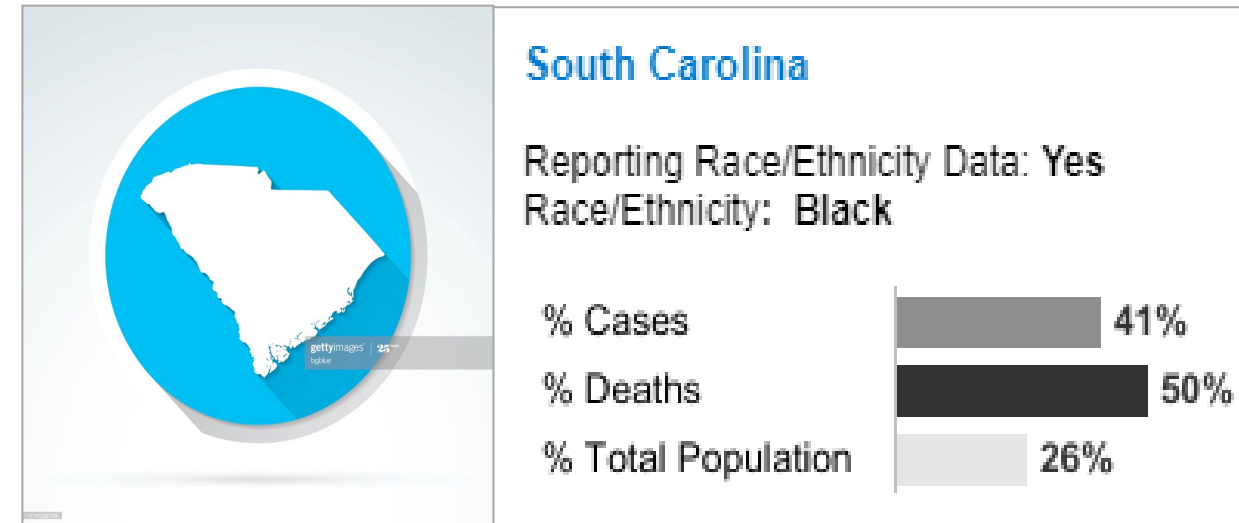
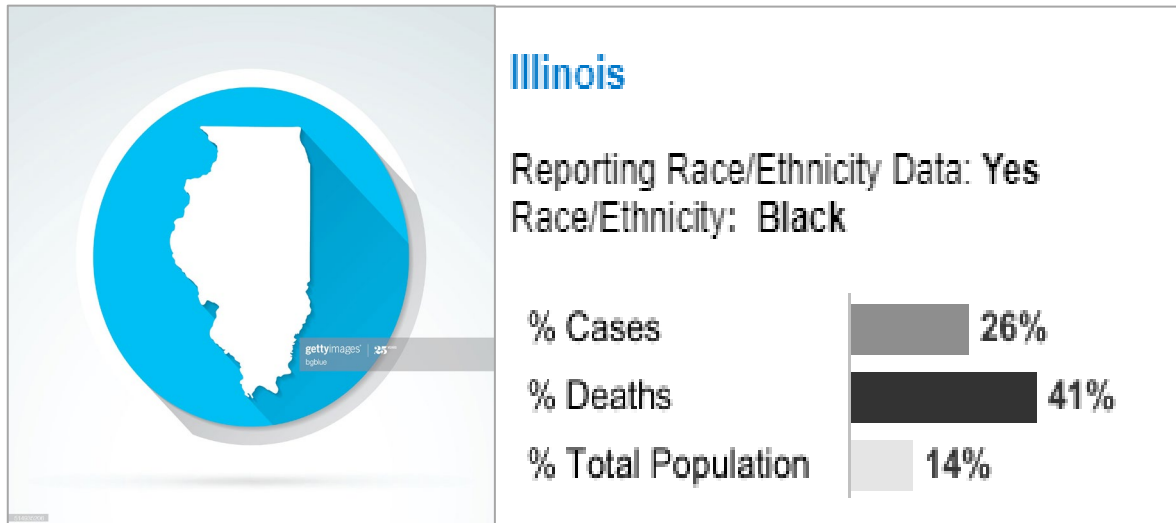
Source: Aubrey A, Neel J. CDC Hospital Data Point To Racial Disparity In COVID-19 Cases. April 8, 2020. ([link](#))
Data from Centers for Disease Control and Prevention ([link](#)); Figure credit: Stephanie Adeline/NPR

Unmasking the Interplay Between Race/Ethnicity and COVID-19



Source: Kaiser Family Foundation. COVID-19: Confirmed Cases & Deaths by Race/Ethnicity as of April 15, 2020. Distribution of Cases/Deaths by Race/Ethnicity based on KFF analysis of publicly available state websites. Total State Population Distribution by Race/Ethnicity based on KFF analysis of 2018 American Community Survey. Accessed 4/28/20

Unmasking the Interplay Between Race/Ethnicity and COVID-19



Source: Kaiser Family Foundation. [COVID-19: Confirmed Cases & Deaths by Race/Ethnicity as of April 15, 2020](#). Distribution of Cases/Deaths by Race/Ethnicity based on KFF analysis of publicly available state websites. Total State Population Distribution by Race/Ethnicity based on KFF analysis of 2018 American Community Survey. Accessed 4/28/20

Pandemic of Unprecedented Scale and Consequences

Cumulative
confirmed
cases of
COVID-19

**>8 million
worldwide**

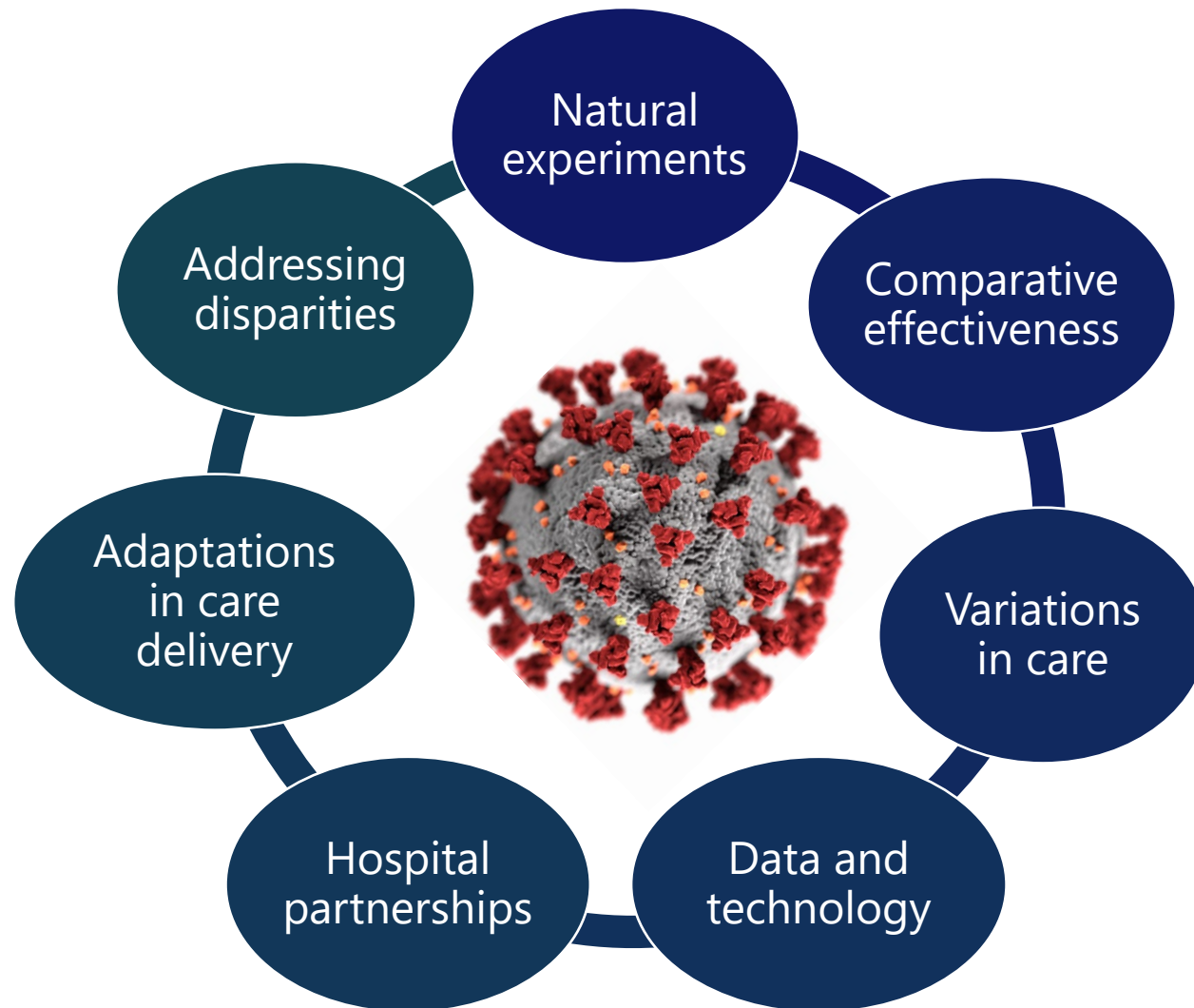


Source: COVID-19 Dashboard by the Center for Systems Science and Engineering (CSSE) at Johns Hopkins University (JHU). <https://coronavirus.jhu.edu/map.html> Accessed 4/28/20.

A Stress Test and Learning Opportunity

Advancing PCORI 2.0

A Learning
Organization
Nimbly Embracing
Opportunities in a
Crisis for Short and
Long-term Impact



PCORI's Response to the COVID-19 Health Crisis

**Healthcare
Delivery**

**Vulnerable
Populations**

**Healthcare
Workers**

**Emerging
Health Trends**

Many approaches to supporting critical work in these areas and more:

Awards

- Enhancements of existing awards
- Solicitation of new awards
- Healthcare worker registry/trial

Information Sharing

- Webinars
- Collaboration with other funders

Adapting for Awardees and Applicants

- Adaptations to existing projects
- Extending application timelines



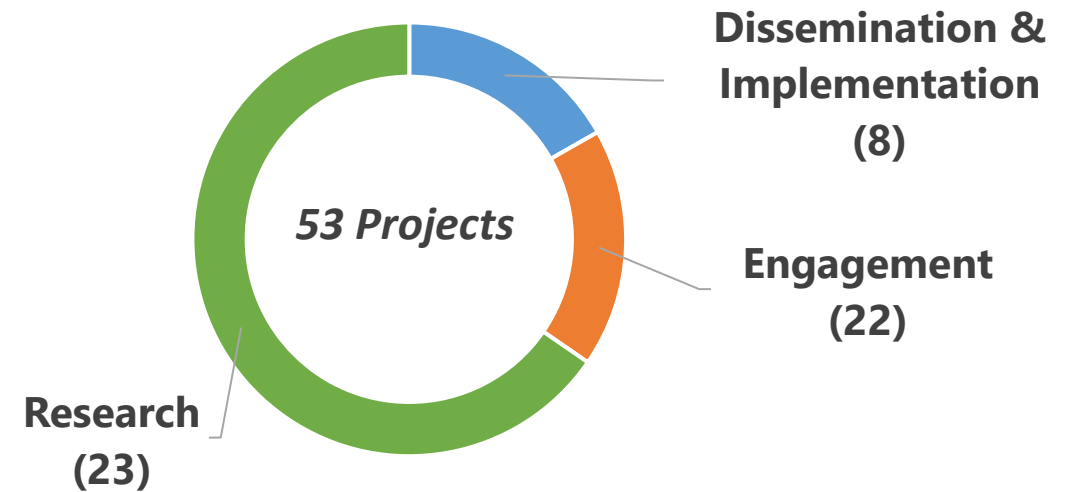
www.heroesresearch.org

COVID-19 Targeted Funding

- Adaptations to healthcare delivery
- Impact on vulnerable populations
- Healthcare workforce well-being, management and training



COVID-19 Enhancement Projects



PCORI – Always Relevant

Meaningful research questions in healthcare delivery



What is the experience of healthcare workers during COVID-19 that can inform key clinical comparative effectiveness questions?

What are the most effective practices for telehealth? Can our current natural experiment shed light on whether telehealth can deliver similar outcomes for patient care and management?

What are the predictors of poor outcomes in COVID-19 infection and can we systematize predictive algorithms to determine who is likely to need intensive care?

What approaches are effective in eliminating health and healthcare disparities in COVID outcomes and how are they generalizable to disparities more broadly?

Accelerate Impact on Care and Patient Outcomes

Engagement

- New and sustained partnerships
- Diverse stakeholder inclusion
- Bipartisan congressional support

Innovation

- Patient-centered learning health care
- Rapid cycle of evidence to implementation
- Pragmatic approaches to lower disparities

Operations

- Creative funding approaches
- Efficient and effective processes

Accelerate Impact on Care and Patient Outcomes: Engagement

Engagement

Impact

- *Patients, clinicians, payers, employers, systems as partners for dissemination and uptake*
- *Evidence to implementation*
- *PCORI integral in research ecosystem*

Impact Post-COVID-19

- *Hospitals and healthcare systems working together*
- *Gaps in evidence for decision-making driving research*
- *PCORI essential*

Accelerate Impact on Care and Patient Outcomes: Innovation

Innovation

Impact

- *"Real-time" implementation of findings*
- *Uptake into guidelines and care*
- *Reduced variation in care and disparities in conditions/populations*

Impact Post-COVID-19

- *"Real-time" queries, output, and implementation guiding accelerated uptake in clinical care*
- *Acute and recommitted focus on variations in care and disparities*

Accelerate Impact on Care and Patient Outcomes: Operations

Operations

Impact

- *Nimble and adaptable methods to speed innovation to results to patients*
- *Robust pool of outstanding research (built capacity)*

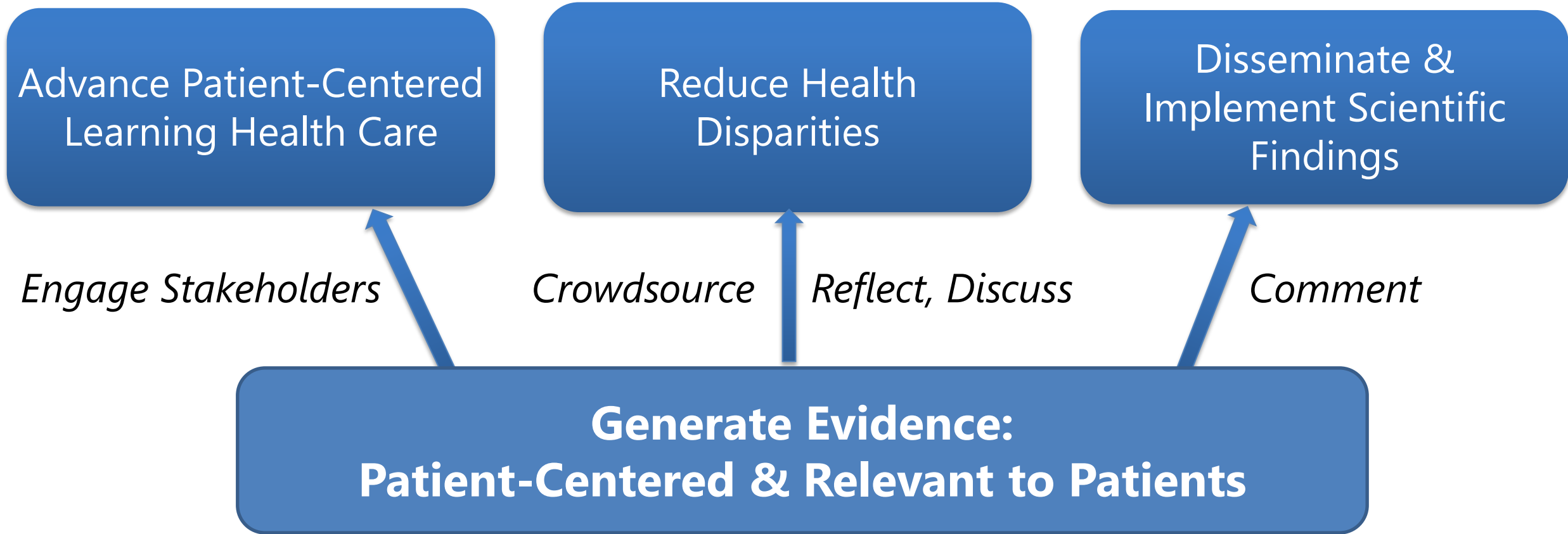
Impact Post-COVID-19

- *Understanding of strengths and limitations of agile and adaptable processes and research*
- *Clearer approach to leveraging data, technology, infrastructure*
- *Expanded research pool*



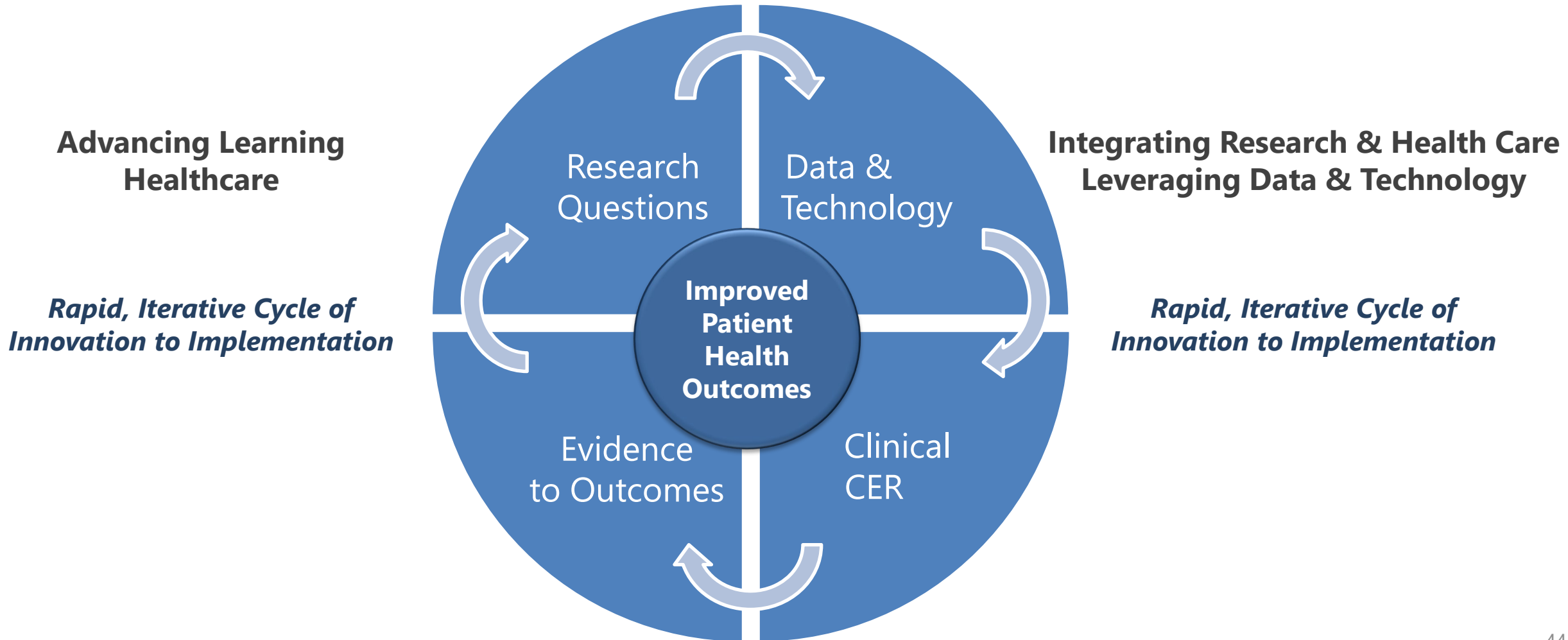
PCORI 2.0: The Journey Ahead

Envisioning PCORI 2.0



Pillars underscored by current pandemic

PCORI 2.0: A Virtuous Cycle of Evidence to Implementation



First-Year Focus

- Virtual listening tour
- PCORI's response to the COVID-19 pandemic
- National Priority setting and development of Research Agenda and strategic plan
- Other priorities stemming from legislation (e.g., maternal mortality, intellectual and developmental disabilities, cost outcomes)



Collaborative Strategy to Advance a Vision for PCORI 2.0

Refreshing National Priorities, Research Agenda, and Strategic Plan

Garner diverse input to guide scientific directions

*Patients, Clinicians, Researchers, Purchasers, Payers, Industry,
Hospitals & Health Systems, Policy Makers, Training Institutions*

Listening Tour

Conduct portfolio analyses & evaluate research programs

Refine priority research areas

Develop implementation plans

Align resources with research priorities

RDAP Advisory Panel

National Priority Setting and Research Agenda



What are the driving questions, outcomes, and measurements to fulfill PCORI's comparative clinical effectiveness research agenda in rare disease populations? What are the challenges and how do we overcome them?

How do we identify common symptoms across rare diseases that affect burden of disease to identify effective approaches to diagnosis and treatment?

How do we advance Rare Disease Research amid the current pandemic?

What are the critical resource and workforce issues to implement these considerations for the next phase for PCORI?

Thank You



BREAK

2:10 – 2:20 PM EST

National Priorities & Research Agenda

Michele Orza, Chief of Staff

Greg Martin, Deputy, Chief Engagement and
Dissemination Officer



Agenda

1. Statutory Mandates
2. Current National Priorities
3. Looking Forward: Panelists' Perspectives on a Refresh

1.

Statutory Mandate



National Priorities

- Required under PCORI's authorizing law.
- Identification is a core duty—the first enumerated duty—of the Institute
- Intended to guide PCORI and provide transparency to the public on the Institute's investment strategy in patient-centered comparative clinical effectiveness research

Subtitle D—Patient-Centered Outcomes Research

SEC. 6301. PATIENT-CENTERED OUTCOMES RESEARCH.

(a) IN GENERAL.—Title XI of the Social Security Act (42 U.S.C. 1301 et seq.) is amended by adding at the end the following new part:

“PART D—COMPARATIVE CLINICAL EFFECTIVENESS RESEARCH

“COMPARATIVE CLINICAL EFFECTIVENESS RESEARCH

“SEC. 1181 [42 U.S.C. 1320e]. (a) DEFINITIONS.—In this section:

“(1) BOARD.—The term ‘Board’ means the Board of Governors established under subsection (f).

“(2) COMPARATIVE CLINICAL EFFECTIVENESS RESEARCH; RESEARCH.—

“(A) IN GENERAL.—The terms ‘comparative clinical effectiveness research’ and ‘research’ mean research evaluating and comparing health outcomes and the clinical effectiveness, risks, and benefits of 2 or more medical treatments, services, and items described in subparagraph (B).

“(B) MEDICAL TREATMENTS, SERVICES, AND ITEMS DESCRIBED.—The medical treatments, services, and items described in this subparagraph are health care interventions, protocols for treatment, care management, and delivery, procedures, medical devices, diagnostic tools, pharmaceuticals (including drugs and biologicals), integrative health practices, and any other strategies or items being used in the treatment, management, and diagnosis of, or prevention of illness or injury in, individuals.

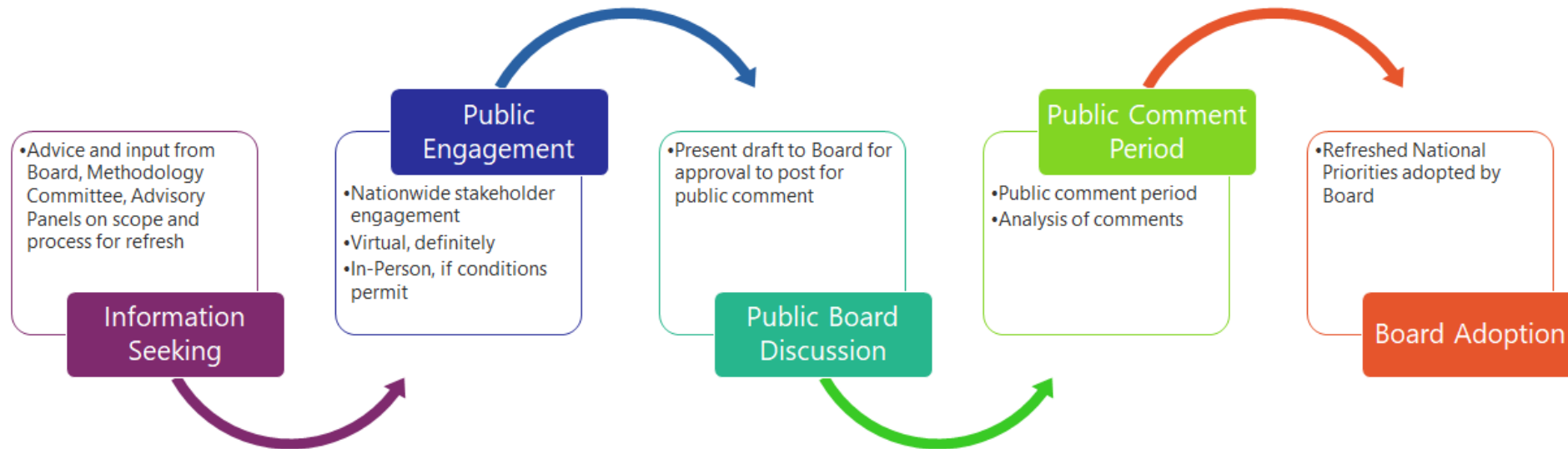
National Priorities

- The Institute shall identify national priorities for research, taking into account **factors of disease** incidence, prevalence, and burden in the United States (with emphasis on chronic conditions), **gaps in evidence** in terms of clinical outcomes, **practice variations** and **health disparities** in terms of delivery and outcomes of care, the **potential for new evidence** to improve patient health, well-being, and the quality of care, the effect on national expenditures associated with a health care treatment, strategy, or health conditions, as well as patient needs, outcomes, and preferences, the relevance to patients and clinicians in making informed health decisions...

**emphasis added*

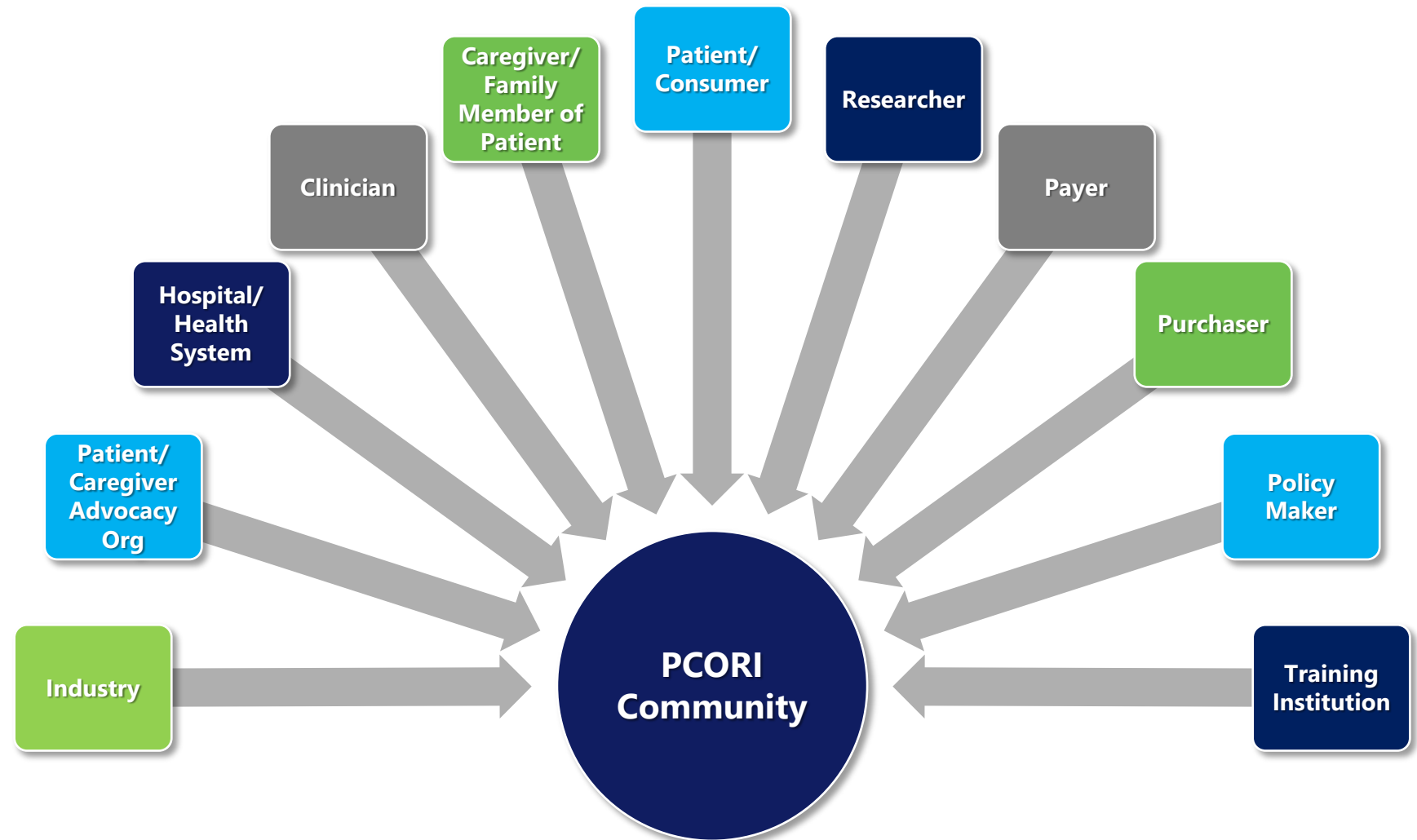
Mandated Process

- Formal Public Comment is required
 - Minimum: 45 days
 - Maximum: 60 days
- Final approval/adoption rests with PCORI Board of Governors



Who We Will Engage

- PCORI
 - Board of Governors
 - Methodology Committee
 - Advisory Panels
- Our Stakeholders across the Nation



2.

Current
National Priorities



Our Current National Priorities

Our prior approach

- Broad national priorities
- Articulation of criteria to inform the *process* for establishing a research agenda

Assessment of Prevention, Diagnosis, and Treatment Options

- Comparing the effectiveness and safety of alternative prevention, diagnosis, and treatment options to see which ones work best for different people with a particular health problem.

Improving Healthcare Systems

- Comparing health system-level approaches to improving access, supporting patient self-care, innovative use of health information technology, coordinating care for complex conditions, and deploying workforce effectively.

Communication and Dissemination Research

- Comparing approaches to providing comparative effectiveness research information, empowering people to ask for and use the information, and supporting shared decision-making between patients and their providers.

Addressing Disparities

- Identifying potential differences in prevention, diagnosis or treatment effectiveness, or preferred clinical outcomes across patient populations and the healthcare required to achieve best outcomes in each population.

Accelerating Patient-Centered Outcomes Research and Methodological Research

- Improving the nation's capacity to conduct patient-centered outcomes research, by building data infrastructure, improving analytic methods, and training researchers, patients and other stakeholders to participate in this research.

3.

Looking Forward:
Perspectives
on a Refresh



We Want to Hear from You



- Questions and Discussion

Overview of Reauthorization & Cost Data Provision

Jean Slutsky, Chief Engagement and Dissemination
Officer

Andrew Hu, Director, Public Policy and Government
Relations

Consideration of Full Range of Outcomes Data



Where appropriate, PCORI-funded studies may capture data on the **potential burdens and economic impacts** of the utilization of medical treatments, items, and services for all stakeholders. This data includes:

- Medical out-of-pocket costs, including:
 - Health plan benefit
 - Formulary design
- Healthcare utilization
- Nonmedical costs to patients and family, including:
 - Caregiving
 - Effects on future costs of care
 - Workplace productivity
 - Absenteeism

This compromise language **does not** remove statutory prohibitions on PCORI establishing cost-per quality adjusted life year (QALY) thresholds or allow PCORI to conduct cost-effectiveness analyses

Consideration of Full Range of Outcomes Congressional and Stakeholder Intent



- Congressional leaders and stakeholders across the healthcare community weighed in on whether to include provisions directing PCORI to capture additional economic/cost-related data
- The goals of this provision is to direct PCORI-funded research to better inform healthcare decisions based on the clinical effectiveness, impact on patient-reported outcomes, **and** potential economic and cost burden
- The provision is a compromise between stakeholders to ensure PCORI-funded research capture both population-level (i.e., utilization, ROI, etc.) and patient-centric (i.e., caregiver burden, impact on medical out-of-pocket costs, etc.) data
- This compromise language **does not** remove statutory prohibitions on PCORI establishing cost-per quality adjusted life year (QALY) thresholds or allow PCORI to conduct cost-effectiveness analyses

Consideration of Full Range of Outcomes Early Stakeholder Input on Implementation



Summary of the key take-aways for implementation include:

- Ensure **transparency**, notably patient engagement, throughout implementation
- Identify the **long-term goals** of capturing this data to inform implementation efforts
- Consider the **full range of treatment options** and the **investment costs** for payers and health systems to implement practice change
- Need to **develop standards** around identifying and capturing patient-centric cost data
- **Concerns around the use/misuse** of cost data that could lead to cost-effectiveness or inappropriate value assessment
- Hope to **expand beyond traditional health economic perspectives** on cost/value

Considerations of Full Range of Outcomes **Tentative** Implementation Proposal

Phase 1

- Providing guidance to PIs in future PFAs on how they should interpret this policy and incorporate it into their research proposals.
- **Timeline:** Complete Phase 1 by January 5, 2021

Phase 2

- Establishing standards and methods to inform the field on how to capture relevant data.
- **Timeline:** 12 months from the initiation of this process

Phase 3

- Convening discussions on how this information can/should be used.
- **Timeline:** Ongoing Discussion

BREAK

3:55 – 4:05 PM EST

Maternal Mortality, Intellectual Disabilities, and Balancing Short- and Long- Term Priorities

Jean Slutsky, Chief Engagement and Dissemination Officer

Kristin Carman, Director, Public and Patient Engagement

Elisabeth Houtsmuller, Associate Director, HDDR

Stanley Ip, Interim Program Director, CEDS

Caitlin McCormick, Associate Director, Public Policy



Recognition of Departing Panel Members

Thank you!

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

- Cindy Luxhoj
- Stephen Mathai
- Marcia Rupnow
- Julie Abramson

Panelist Recognition: Cindy Luxhoj, MUP



- Represented: Patients, Caregivers, and Patient Advocates
- Served as Co-Chair of the RDAP from 2018-2020
- Served as the Executive Director of Alagille Syndrome Alliance (ALGSA) and tirelessly helped families cope with the daily challenges of ALGS
- Provided the patient caregiver perspective of the challenges associated with rare diseases

Panelist Recognition: Stephen Mathai, MD



- Represented: Researchers
- Extensive experience treating patients with pulmonary arterial hypertension (PAH), a rare disease that affects 15-50 people per million in the United States
- Serves on the Scientific Leadership Council of the Pulmonary Hypertension Association
- Chair-elect of the Pulmonary Vascular Disease Network of the American College of Chest Physicians

Panelist Recognition: Marcia Rupnow, MS, PhD



- Represented: Industry
- Vice President of Value Evidence and Outcomes at GlaxoSmithKline
- Unique breadth of experience in both pharmaceutical and medical device sectors
- Co-authored over 50 publications in peer-reviewed journals and over 100 poster/podium presentations at scientific meetings
- Also served on the PCORI Advisory Panel on Assessment of Prevention, Diagnosis, and Treatment Options from 2013 to 2016

Panelist Recognition: Julie Abramson, MS



- Represented: Patients, Caregivers, and Patient Advocates
- Serves as a patient advocate and co-chair on the Steering Committee for the NephCure Kidney Network, a PCORI-funded PPRN
- Mentored patients in learning about Nephrotic syndrome and contributed to a new patient learning module with NephCure
- Diverse skillset including an understanding of the scientific process and creative methods to engage patients

Acknowledgments & Recap

Adjourn
