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

December 16, 2020

Scott Berns, MD, MPH, FAAP
Chair, RDAP

Doug Lindsay, BS
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 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 Winter 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 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
2:00 PM	Welcome, Introductions, and Setting the Stage	Scott Berns, Doug Lindsay
2:30 PM	Introduction of RDAP PCORI Members, RDAP Program Staff Update, and Upcoming Activities	Carly Khan, Nora McGhee
2:40 PM	Engagement Awards and Rare Disease Organizations: Supporting Engagement in PCOR/CER	Karen Martin, Scott Berns
3:25 PM	**BREAK** (10 minutes)	
3:35 PM	Maternal Mortality and Intellectual Disabilities: Overview and Discussion of Issues related to Rare Diseases	Els Houtsmuller, Kelly Dunham, Scott Berns
4:15 PM	**BREAK** (5 minutes)	
4:20 PM	Identifying our National Priorities: Relevance for Rare Disease Populations	Laura Lyman Rodriguez, Jean Slutsky, Doug Lindsay
4:50 PM	Acknowledgments and Recap	Scott Berns, 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 (NORD)

Danielle Boyce (Johns Hopkins University)

Julie Gortze (Rare New England)

Mathew 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

- Conducting Rare Disease Research Using PCORnet® — PCORI Board of Governors will vote in March 2021
  - Must answer an important question about care of patients with rare disease **AND**
  - Build infrastructure of PCORnet to enhance capabilities for multi-site rare disease research
  - Studies can be either CER **OR** descriptive in nature that lead to future CER
- The 2020 PCORI Annual Meeting featured a session focused on rare disease research: sessions are available for viewing on-demand via this link
- PCORI Advisory Panel Applications are open through March 2021: please share this opportunity!
- Resources: Research Fundamentals

  Building Effective Multi Stakeholder Research Teams (Coming Soon)

Engagement Awards and Rare Disease Organizations: Supporting Engagement in PCOR/CER

Karen Martin, Program Director

December 16, 2020

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Meet Our Team

Karen Martin, MIA
Director

Alicia Thomas, DrPH, MHS
Associate Director

Ross Schwarzber, MA
Program Manager

Courtney Clyatt, MPH
Senior Program Officer

Kate Boyd, MPH, DrPHc
Program Officer

Ivey Wohlfeld
Program Manager

Rachel Mosbacher, MPA
Program Officer

Sangeeta Suku, MPH, MBA
Program Associate

Fatou Ceesay, MPH
Program Officer

Objectives

- Provide an overview of the Engagement Award Program
- Discuss how the Engagement Awards can support rare disease organizations in building capacity for stakeholder engagement in PCOR/CER
- Identify specific needs, activities, tools and assistance rare disease organizations need to successfully engage in PCOR/CER

Eugene Washington PCORI Engagement Award Program

- Support projects to build a community of patients and other stakeholders equipped to participate as partners in PCOR/CER, as well as serve as channels to disseminate PCORI-funded study results
- Funding to support engagement in, and with, research, not to conduct research

Develop Community Skilled in PCOR

Engage Community in Research Processes

Involve Community in Dissemination

Engagement Award Funding Opportunities

$250,000
Up to 2 years

Engagement Award: Capacity Building

Objective: Prepare patients and stakeholders to participate as partners in PCOR/CER and/or develop partnerships and infrastructure to disseminate and implement PCORI-funded research findings

$250,000
Up to 2 years

Engagement Award: Dissemination Initiative

Objective: Support communities and organizations to actively disseminate PCORI-funded research findings

$100,000
Up to 1 year

Engagement Award: Stakeholder Convening Support

Objective: Convene stakeholders to explore critical issues related to PCOR/CER and/or communicate PCORI-funded research findings to targeted end-users

What Engagement Awards Don’t Fund

- Out of Scope for an Engagement Award Project:
  - Projects solely intended to increase patient engagement in health care or healthcare systems rather than healthcare research
  - Projects to design or test healthcare interventions
  - Activities that involve the use of a drug or medical device
  - Development of clinical practice guidelines, care protocols, or decision support tools
  - Development of coverage, payment, or policy recommendations or guidelines
  - Projects related to quality measures, or engagement around quality measures
  - Projects to recruit and enroll patients for clinical trials
  - Projects that only involve patients as subjects (individuals enrolled into a study as participants)
  - Research studies including randomized controlled trials, observational studies, and pragmatic clinical studies

What Engagement Awards Don’t Fund (continued)

- Development or maintenance of a registry, or recruitment to participate in a registry
- Projects designed solely to validate tools or instruments not created through a PCORI-funded project
- Writing research proposals or completing grant applications, grantmaking
- Projects focused on social determinants of health, with no focus on patient-centered outcomes research or comparative clinical effectiveness research
- Planning for dissemination or dissemination initiatives without including PCORI-funded research or related products
- Implementation of PCORI findings in a clinical practice setting (PCORI will fund dedicated implementation efforts through the Limited PCORI Funding Announcement: Implementation of PCORI-Funded Patient-Centered Outcomes Research Results)
- Projects or meetings without a clear focus on patient-centered outcomes research or comparative clinical effectiveness research

Evolution of Funding Priorities

2014-2016

- Knowledge Awards
- Training and Development Awards
- Dissemination & Implementation Awards
- Meetings and Conferences Support

October 2017-February 2018

- Engagement Award (general)
- Meetings and Conference Support

June 2018-June 2019

- Capacity Building
- Dissemination Initiative
- Conference Support

November 2019

Special Project Funding Announcements:

- Community Convening
- Accelerating the Adoption of Tools and Resources

May 2020

- Capacity Building
- Dissemination Initiative
- Stakeholder Convening
- COVID-19 Enhancements and Special Cycle

October 2020

- Capacity Building
- Dissemination Initiative
- Stakeholder Convening
- *Next Steps: Opportunities for growth under PCORI 2.0*

Feedback from RDAP Meeting – Spring 2020

- Identify ways to best support rare disease groups
- Consider targeted funding announcements – rare and ultra-rare diseases
  - Bundling rare diseases that have common symptoms
- Review language and readability of funding announcements
- Increase accessibility to rare disease community through patient outreach
- Identify essential activities, tools and skills rare disease groups need to successfully engage in PCOR/CER

Discussion Questions

- What are the needs of smaller organizations (e.g., community-based organizations, patient/caregiver/advocacy organizations, associations, etc.) with regards to building capacity for stakeholder engagement in PCOR/CER?

- What do rare disease organizations need to successfully engage in PCOR/CER?
    - PCOR/CER training for researchers or stakeholders?
        - If so, what types of activities or trainings might be helpful to organizations that aren’t familiar with PCOR/CER and/or PCORI?
    - Engagement tools?
    - Infrastructure?

- What other kinds of foundational assistance would help rare disease organizations lay the groundwork for participating in PCOR/CER?

BREAK (10 minutes)

We will return at 3:35 pm EST

PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

Maternal Mortality and Intellectual and Developmental Disabilities: Overview and Discussion of Issues Related to Rare Diseases

Els Houtsmuller, PhD

Associate Director, Healthcare Delivery and Disparities Research

Kelly Dunham, MPP

Senior Manager, Office of the Chief Science Officer

Putting our Mandate into Action

- Reauthorization language identified two research priorities:
  - Maternal morbidity and mortality (MMM)
  - Intellectual and developmental disabilities (IDD)

Hear from Dr. Nakela Cook in her recent blog

Formulating Our Approach to New Priority Research Areas

Date: September 8, 2020

Blog Topics: Executive Director's Blogs,
Funding Awards,
Research

As part of last winter's legislation that reauthorized PCORI's funding for 10 years, Congress included two new research priority areas for PCORI to address: strategies for improving maternal mortality, and improving health for individuals with intellectual and/or developmental disabilities (IDD).

These areas are of critical importance for PCORI given the long-standing health challenges faced by those affected and the opportunities that PCORI's approach to *research done differently* can contribute to meaningful health improvements. We are committed to a multipronged approach to funding research to address a variety of issues related to these two topics over the next decade.

Addressing Maternal Morbidity and Mortality

The United States consistently ranks near the bottom among high-income

Putting our Mandate into Action

Stakeholder Input; Evidence Reviews; Topic Identification, Prioritization, Development

CER AWARDS

- *Broads √*
- *Phased Large Awards √*
- *Pragmatic Clinical Studies*
- *Targeted Funding Announcements*

EVIDENCE SYNTHESIS

- *Systematic Reviews*
- *Rapid Reviews*
- *Evidence maps/visualizations*

ENGAGEMENT AWARDS

- *Capacity Building √*
- *Stakeholder Convening Support √*
- *Dissemination*

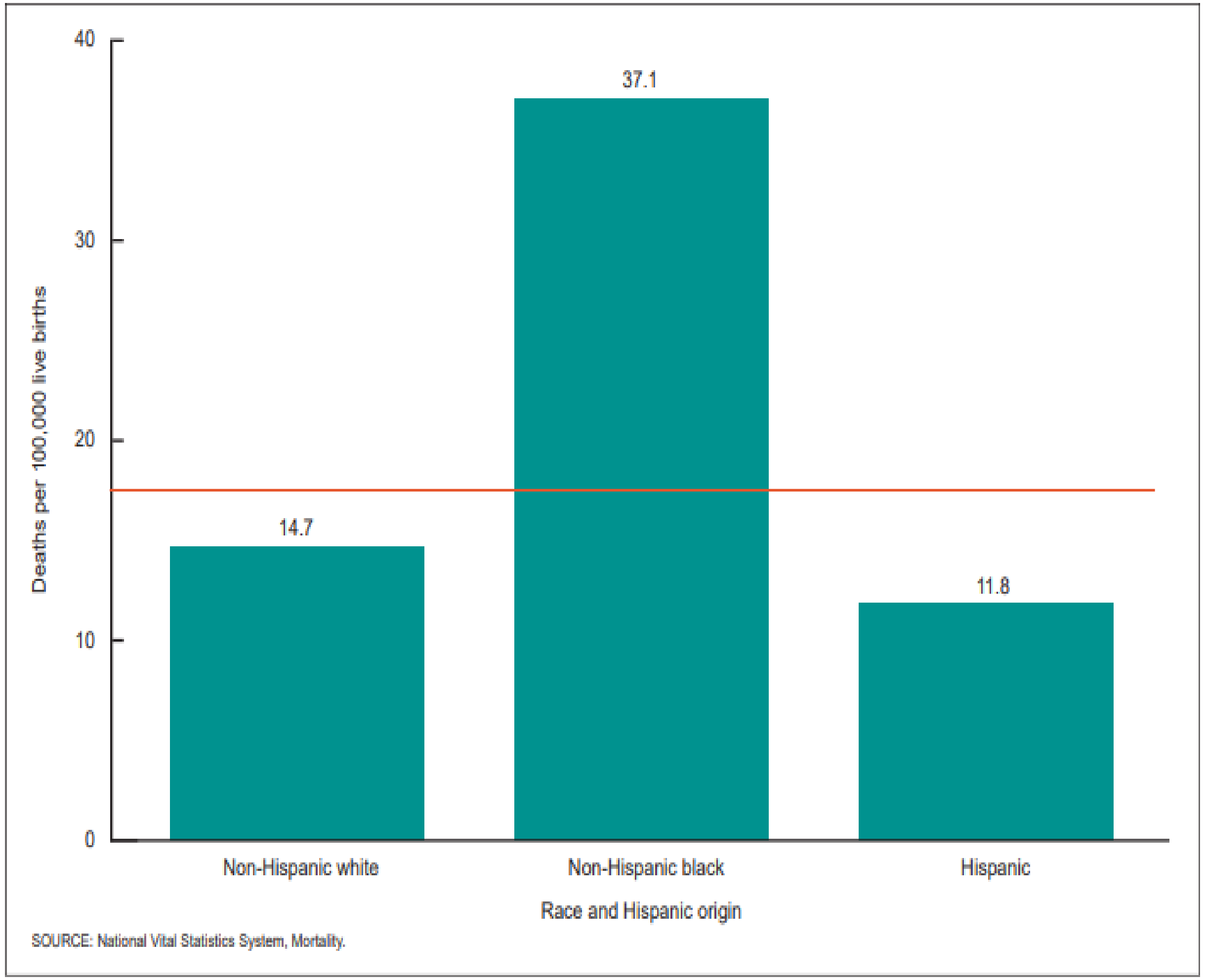
Maternal Mortality and Morbidity

Maternal Mortality: US Rates and Disparities

2017 Maternal Mortality Rates per 100,000 live births by Country

Norway	2
Italy	2
Finland	3
Greece	3
Denmark	4
Spain	4
Sweden	4
Iceland	4
Austria	5
Netherlands	5
Japan	5
Switzerland	5
Germany	7
US	19

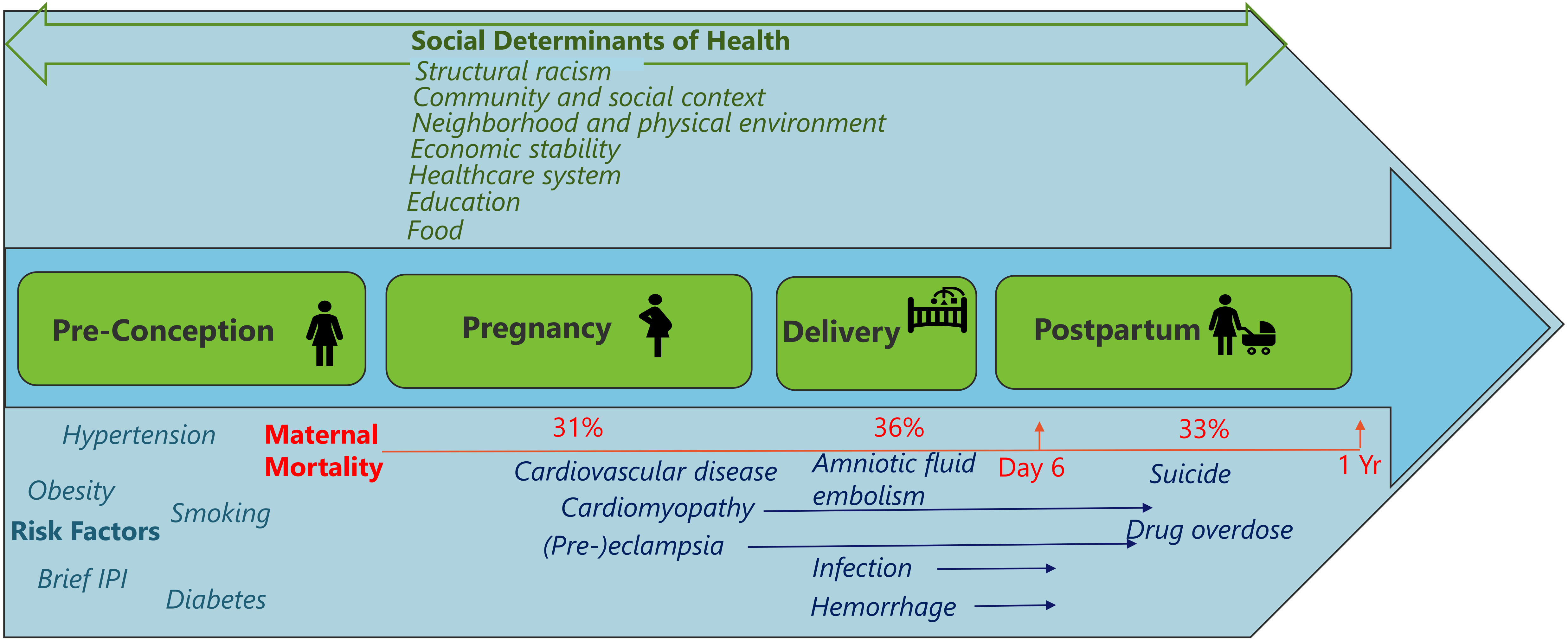
The World Factbook, 2020



SOURCE: National Vital Statistics System, Mortality.

Maternal mortality rates, by single race and Hispanic origin: United States, 2018

Maternal Mortality Framework:
More than just pregnancy and delivery



Social Determinants of Health

Structural racism
Community and social context
Neighborhood and physical environment
Economic stability
Healthcare system
Education
Food

Pre-Conception

Pregnancy

Delivery

Postpartum

Risk Factors

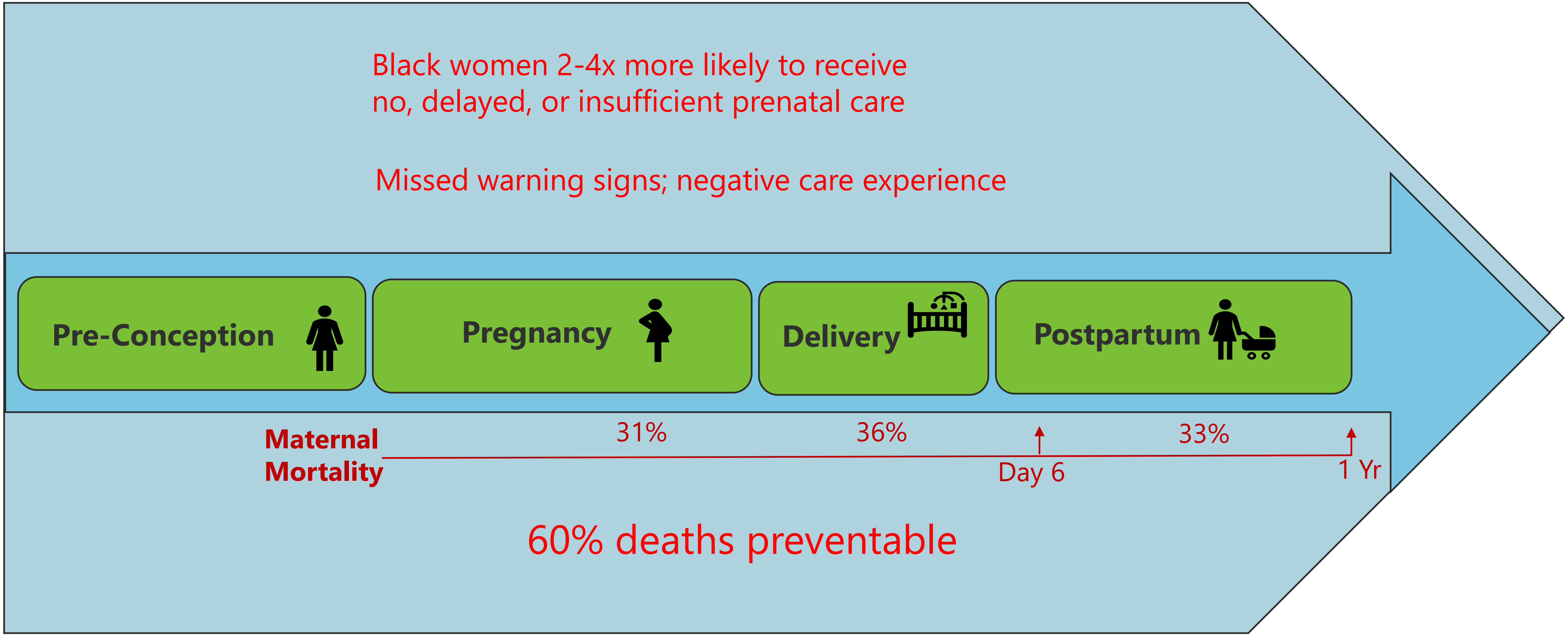
Hypertension
Obesity
Smoking
Brief IPI
Diabetes

Maternal Mortality

31%
36%
33%
Day 6
1 Yr

Cardiovascular disease
Cardiomyopathy →
(Pre-)eclampsia →
Amniotic fluid embolism
Infection →
Hemorrhage →
Suicide
Drug overdose

Maternal Morbidity and Mortality



Black women 2-4x more likely to receive no, delayed, or insufficient prenatal care

Missed warning signs; negative care experience

Maternal Mortality

60% deaths preventable

Special Area of Emphasis

$30 million available – Broad Funding Announcement

Increasing Access to and Continuity of Patient-Centered Maternal Care

What is the comparative effectiveness of **multilevel, culturally adapted interventions** that address **barriers in access to and continuity** of optimal **patient-centered maternal care** for pregnant women from populations that experience significant disparities in outcomes (Black, NA/AN, and/or rural)?

- Multilevel interventions that compare varying levels of intensity for Maternal care coordination; Education or training for healthcare providers and/or patients; and/or Add-on or wraparound services

Maternal Mortality
Stakeholder Engagement to Date: A Sample

- A few organizations that we've engaged with
- Not an exhaustive list

Patients/Patient Advocates	• National Birth Equity Collaborative • National Partnership for Women and Families • National Rural Health Association
Policymakers	• Black Maternal Health Caucus Advisory Group • Association of State and Territorial Health Officials
Clinicians	• American College of Obstetricians-Gynecologists • American College of Nurse-Midwives • American Academy of Family Physicians
Hospitals/Health Systems	• Colorado Hospital Association • Mamatoto Village
Researchers/Funders	• National Institute of Child Health and Human Development • Individual researchers
Payers	• Centers for Medicare and Medicaid Services • Medicaid Medical Directors Network

PCORI hosted a multi-stakeholder Salon in the fall of 2019

Intellectual and Developmental Disabilities

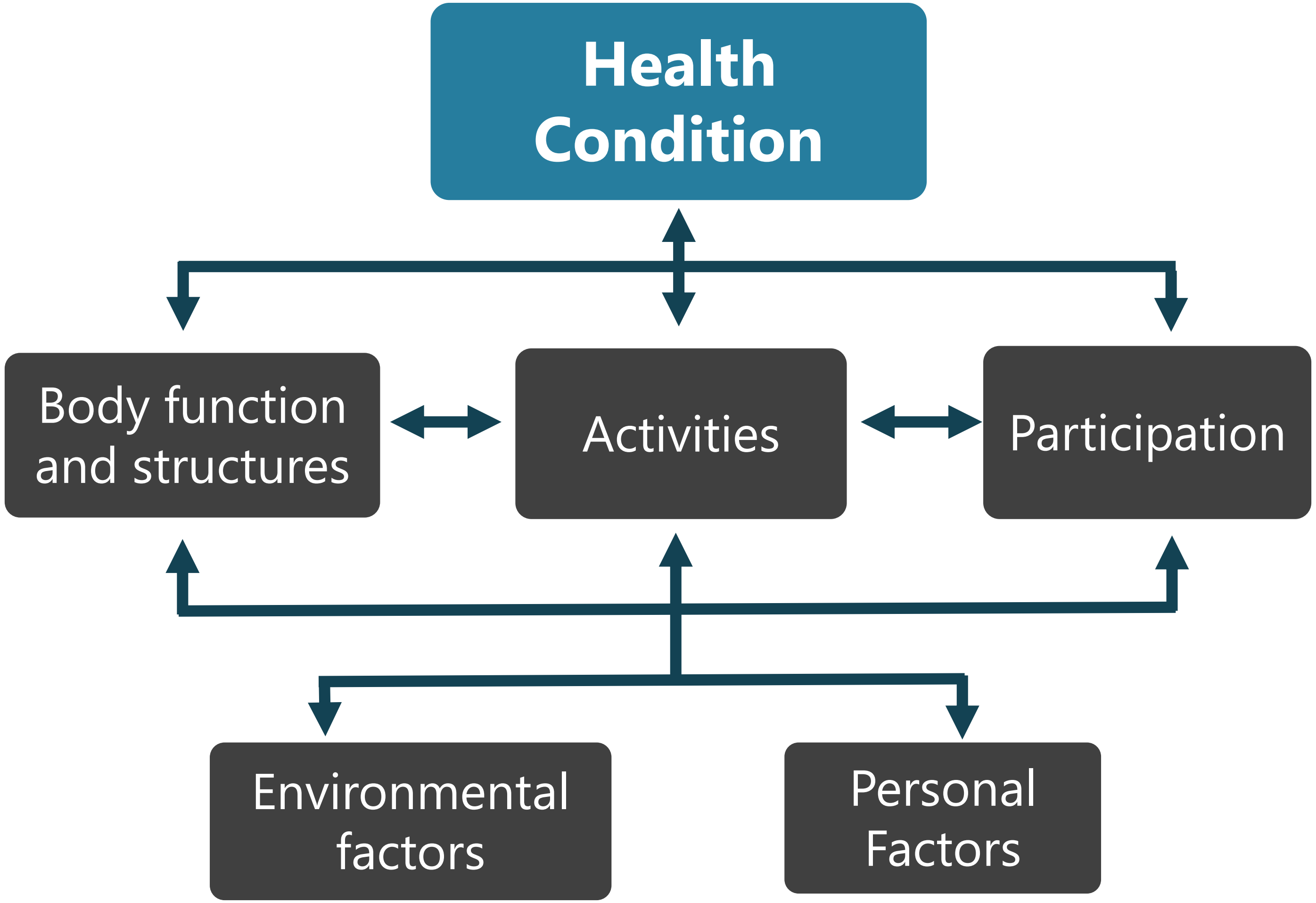
Intellectual and Developmental Disabilities: Context

- Intellectual and developmental disabilities (IDD) are disorders that are usually present at birth and that negatively affect the trajectory of the individual's physical, intellectual, and/or emotional development. [NICHD]
  - **Intellectual disabilities** are characterized by significant limitations in both intellectual functioning and adaptive behavior. [AAIDD]
  - **Developmental disabilities** are chronic and can be cognitive, physical or both. [AAIDD]
- People with IDD comprise a vulnerable population with poorer health status, shorter lifespan, and worse health care outcomes than the general population
  - Additional challenges for populations already at risk for disparities

Levers for improving health outcomes for individuals with IDD: ICF Model

Functioning and Disability

Social determinants of health (Barriers and facilitators)



Health Condition

Body function and structures

Activities

Participation

Environmental factors

Personal Factors

WHO, 2001

Intellectual and/or Developmental Disabilities
Stakeholder Engagement to Date: A Sample

- A few organizations with whom we’ve engaged
- Not an exhaustive list

Key Question for Panel

- Are there organizations we should consider?

Patients/Patient Advocates	• National Down Syndrome Society • Autism Speaks • The Arc • American Association on Intellectual and Developmental Disabilities
Policymakers	• National Association of State Directors of Developmental Disabilities Services • US DHHS/Office of Autism Research Coordination
Clinicians	• American Academy of Family Physicians • American Academy of Pediatrics
Researchers/Funders	• Association of University Centers on Disability • National Institute on Disability, Independent Living, and Rehabilitation Research • Teams conducting high-quality IDD-related PCOR (Individuals and UCEDDs)
Payers	• Medicaid Medical Directors Network

PCORI hosted a multi-stakeholder town hall at the 2020 PCORI Annual meeting.

Intellectual and/or Developmental Disabilities: Funding

- **Special Area of Emphasis, Broad Cycle 3 2020 and Cycle 1 2021:**

  Improving Care for Individuals with Intellectual and/or Developmental Disabilities Growing into Adulthood

  What is the comparative effectiveness of models of care, or of components of such models, to support the healthcare transition from childhood to adulthood and the continuation of patient-centered primary and specialty healthcare for individuals with IDD?

  Up to $30M

- IDD identified as a **research priority in PLACER and Engagement Award PFAs**

IDD: Emerging areas for future research

- Transitions from adolescence to adulthood, and for aging individuals with IDD
- Treatments across the life course
- Challenges to delivering care for individuals with IDD (e.g., access, communication)
- Comparative effectiveness of pharmacologic and/or nonpharmacologic treatments,
- Treatment of comorbid mental health conditions in individuals with IDD
- Patient preferences and needs, autonomy
- Caregiver needs and access to wraparound support

Key Question for Panel

- Are there other research areas that you would suggest (e.g., cross-cutting topics on symptoms that might benefit the rare disease community)?

Cross-cutting issues for MMM and IDD research

Challenges of Research in Maternal Morbidity and Mortality Research

- Mortality – deaths rare event
- Patients not seeking care
- Recruitment and operations
- Pre-conception risk factors

Challenges of Research in IDD Research

- Heterogeneity of populations
- Recruitment and operations
- Accessing populations
- Caregivers as partners in data capture

- **Are there examples in rare disease that we can learn from to support MMM and IDD research?**
  - **Accessing populations that are hard to reach?**
  - **Research on mortality?**
  - **Good practices?**
  - **Which other components and key features of research plans are critical to success?**

Thank You!

BREAK (5 minutes)

We will return at 4:20 pm EST

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Identifying our National Priorities: Relevance for Rare Disease Populations

Laura Lyman Rodriguez, PhD

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

Original Strategic Framework (2013)

NATIONAL PRIORITIES for RESEARCH

Priorities that Guide Our Research

- Addressing Disparities
- Assessment of Prevention, Diagnosis, and Treatment Options
- Communication and Dissemination Research
- Improving Healthcare Systems
- Accelerating PCOR and Methodological Research

STRATEGIC IMPERATIVES

How We Create

- Engagement
- Methods
- Research
- Dissemination
- Infrastructure

OUTPUTS

What We Create

- Skilled Patient-Centered Outcomes Research Community
- Patient-Centered Outcomes Research Methods
- Portfolio of Patient-Centered Outcomes Research Studies
- Communication and Dissemination Activities
- Patient-Centered Research Networks

GOALS

What We Accomplish

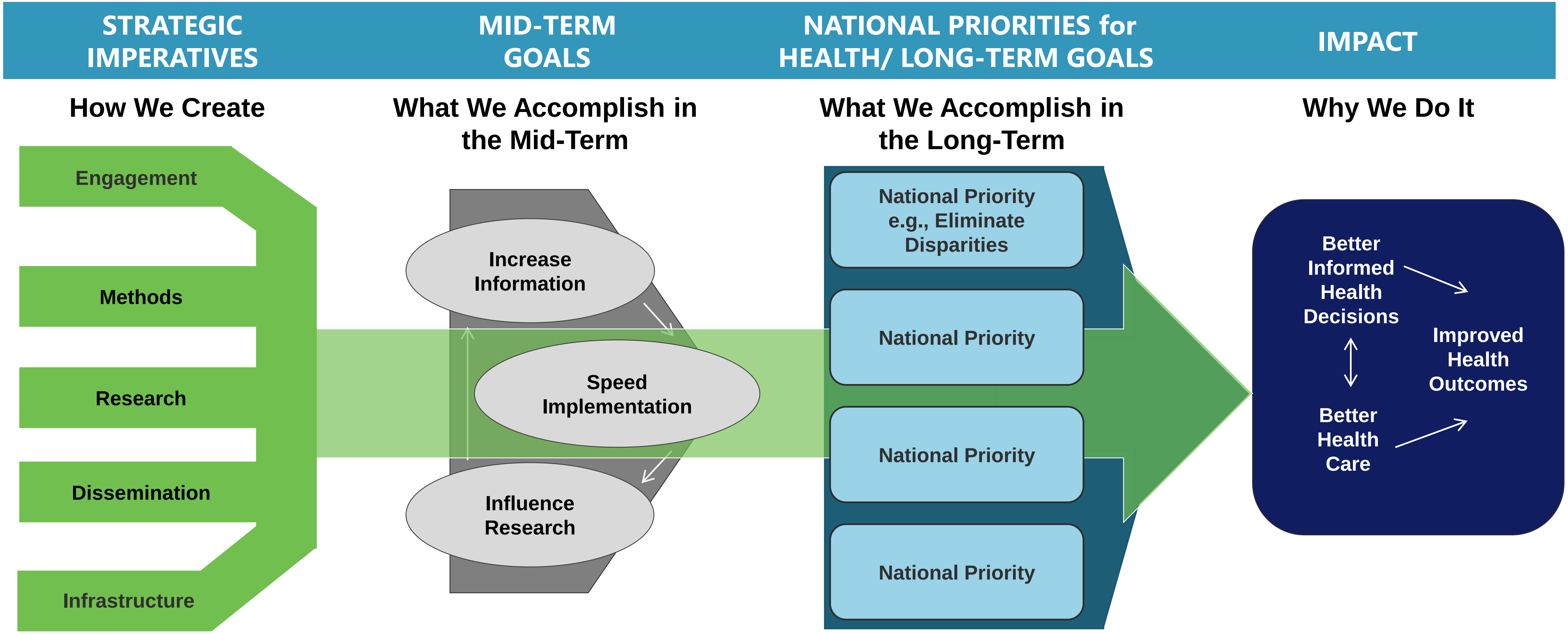
- Increase Information
- Speed Implementation
- Influence Research

IMPACT

Why We Do It

- Better Informed Health Decisions
- Better Health Care
- Improved Health Outcomes

Revised Strategic Framework



STRATEGIC IMPERATIVES

How We Create

- Engagement
- Methods
- Research
- Dissemination
- Infrastructure

MID-TERM GOALS

What We Accomplish in the Mid-Term

- Increase Information
- Speed Implementation
- Influence Research

NATIONAL PRIORITIES for HEALTH/ LONG-TERM GOALS

What We Accomplish in the Long-Term

- National Priority e.g., Eliminate Disparities
- National Priority
- National Priority
- National Priority

IMPACT

Why We Do It

- Better Informed Health Decisions
- Better Health Care
- Improved Health Outcomes

Existing National Priorities (Adopted in 2012)

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.

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.

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.

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.

Accelerating PCOR 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.

Legislatively-Mandated Priority Topics Cut Across Our National Priorities

Intellectual and developmental disabilities and maternal morbidity and mortality:
Priority topics for the next 10 years

Is Anything Missing

Looking at all the existing National Priorities and considering the revised framework,

- Is there an important goal for health that is not reflected in the National Priorities?
- How can PCORI best support a goal-focused strategy on health for our National Priorities that is inclusive of rare disease?

- What we heard from the RDAP members during the June meeting:
  - CER should be made more relevant and understandable to the average person
  - Leverage engagement to facilitate dissemination and implementation of CER research findings

Where Do We Go From Here
Accelerating PCOR and Methodological Research

Description

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.

What we've been hearing about this Priority

- Consider infrastructure needed to support the other priorities
- Data infrastructure ecosystem could lead to efforts that complement PCORI's work
- Emphasize human component of infrastructure
  - Capacity building to include diverse participants in research
  - Development of a research pipeline for PCOR

We want to hear from you

- What does the reframing of the National Priorities from categories of research to goals for health mean for the Methods priority?
- How should goals for health for rare disease be considered when crafting a national priority related to Methods?

Where Do We Go From Here
Communication and Dissemination Research

Description

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.

What we’ve been hearing about this Priority

- Importance of doing communication and dissemination, not just the research of it
- Community engagement can facilitate strong dissemination
- Continued need to effectively communicate research findings to those who need it

We want to hear from you

- What does the reframing of the National Priorities from categories of research to goals for health mean for the CDR priority?
- What considerations related to rare disease arise from the reframing of the CDR priority?

Where Do We Go From Here
Addressing Disparities

Description

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.

What we've been hearing about this Priority

- Remains more important than ever
- Needs to be strengthened (e.g., eliminate disparities rather than addressing)

We want to hear from you

- What does the reframing of the National Priorities from categories of research to goals for health mean for the AD priority?
- How can PCORI support a focus on health goals in populations that experience disparities?

Where Do We Go From Here

Improving Healthcare Systems

Description

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.

What we've been hearing about this Priority

- Reflect intersection of priority with broader public health ecosystem (e.g., social determinants of health)

We want to hear from you

- What does the reframing of the National Priorities from categories of research to goals for health mean for the IHS priority?
- What systems-level factors should PCORI consider related to rare disease?

Where Do We Go From Here
Assessment of Prevention, Diagnosis, and Treatment Options

Description

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.

What we've been hearing about this Priority

- Importance of prevention and its link to broader public health
- Incorporate the environmental factors and public health efforts with this priority
- Important to maintain focus on comparative trials of drugs, devices, surgical techniques, and other interventions

We want to hear from you

- What does the reframing of the National Priorities from categories of research to goals for health mean for the APDTO priority?
- When there are not treatment options for a rare disease, what other CER questions are important?

Thank You!

Acknowledgments & Recap

Adjourn