External Meeting Summary

Advisory Panel on Rare Disease

May 18, 2022

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Overview

The Rare Diseases Advisory Panel (RDAP) Spring 2022 meeting involved updates on the PCORI Strategic Plan, a discussion on the uptake of rare disease research results from the PCORI portfolio, and a robust discussion on healthcare cost and value. The meeting agenda included the following:

- Introduction of RDAP PCORI Staff & RDAP Program Staff Update
- Intellectual and Developmental Disabilities (IDD) Update
- Building Capacity for Cross-Cutting Rare Disease Research
- Healthcare Costs and Value
- PCORI's Strategic Plan
- Acknowledgments and Recap

Introduction of RDAP PCORI Staff & RDAP Program Staff Update

- Carly Khan, PhD, MPH, RN, Associate Director, Healthcare Delivery and Disparities Research, PCORI
- Nora McGhee, PhD, Senior Program Officer, Clinical Effectiveness and Decision Science, PCORI
- Fatou Ceesay, MPH, Program Officer, Engagement Awards, PCORI
- Meghan Berman, MPH, Program Associate II, Public and Patient Engagement, PCORI
- Rohini Mohanraj, MHA, Program Associate, Research Infrastructure, PCORI

Fatou Ceesay and Meghan Berman discussed two new Eugene Washington PCORI Engagement Awards: The Science of Engagement Initiative and the Building Capacity for Small Organizations to Engage in Patient-Centered Outcomes Research/Comparative Clinical Effectiveness Research (PCOR/CER) award. The Science of Engagement Initiative will enable PCORI to fund research studies aimed at answering some of the key engagement questions facing the field. The Building Capacity for Small Organizations to Engage in PCOR/CER award will enable small organizations and their communities to build capacity to participate in the PCOR/CER process.

Nora McGhee provided an update on upcoming PCORI activities in the fall, including the PCORI Annual Meeting (October 26-27, 2022), the next RDAP meeting (October 25, 2022), and the review of applications for new Advisory Panel members joining in Fall 2022. Finally, McGhee acknowledged that input from the RDAP at the Winter 2021 meeting resulted in changes to PCORI’s Strategic Plan that will be discussed in the presentation by Laura Rodriguez.

Intellectual and Developmental Disabilities (IDD) Update

- Carly Khan, PhD, MPH, RN, Associate Director, Healthcare Delivery and Disparities Research, PCORI

Carly Khan reminded panelists that when Congress reauthorized PCORI in 2019, they reemphasized IDD as a national priority area for PCORI. This reinforced PCORI’s long-term commitment to and legislative mandate for this research priority, guided by PCORI’s IDD Workgroup. The Workgroup is charged with developing and implementing a research agenda to ensure generation of evidence that helps neurodiverse individuals access quality care.

Khan explained that in the last two years, PCORI has focused on funding high-priority research and building capacity for PCOR. Additionally, PCORI has emphasized dissemination and implementation activities, as well as efforts to enhance research design and efficiency. Khan reviewed past and current research questions addressed via PCORI's different funding mechanisms. She then facilitated a discussion on the following questions:

Are there other topical areas of focus relevant to both IDD and rare disease?

- Caregivers play a significant role in both IDD and rare disease populations—especially when both conditions are involved.
- Noting the lack of structure for people with disabilities once they leave the education system, panelists listed patients' transitions from school into community as a key research topic.
- Research on the effects of a rare disease and associated treatments, especially related to brain development in children, is a priority for this group.
- The diagnostic odyssey and the mental health of caregivers during this process is a priority across rare diseases and is of great importance to panel members, who noted that research on siblings (in caregiver roles and in general) is also lacking.

Are there methodological considerations relevant to rare disease research that may be relevant to IDD research?

- Conditions that limit patients' communication abilities require use of innovative, accurate methods for collecting data on universal issues (e.g., pain, sleep, and mental health), especially when caregivers provide proxy measures
- Developing global outcome measures across rare diseases
- Engaging with patients in the IDD space
- Learning how to communicate effectively with patients who have communication limitations, and their caregivers
- Recognizing the lack of applicability of purpose outcomes, such as patient-reported outcomes

Are there stakeholder groups with whom PCORI should consider connecting?

- The education system, given the amount of time children with disabilities including IDDs spend in academic environments potentially until their early twenties
- DEEP Connections
- Parks and recreation departments
- Child life specialists

Uptake of Rare Disease Study Results

- Heather Edwards, PhD, MPH, MBA, Associate Director, Executive Office, Evaluation and Analysis, PCORI

Heather Edwards presented a graphic that outlines PCORI's road to impact for research studies. First, studies are completed. Next, the results are released. Then, the results are utilized and adopted. Lastly, the results have impact. Edwards noted that PCORI rare disease research projects published 169 articles prior to 2022. Other dissemination methods (e.g., social media, news stories) helped share findings and increase their uptake. Results from PCORI studies have been featured in systematic reviews, guidelines, patient-facing materials, policy documents, and UpToDate. Edwards then opened the discussion with the following questions:

What are your reactions to these findings about the Rare Disease portfolio?

- Panelists asked for a more detailed breakdown outlining the gaps in the PCORI-funded research portfolio, as well as information about PCORI's investments overall, to understand how the Rare Disease portfolio fits into PCORI's research landscape.
    - PCORI offered to share an overview of this information, including data about the investments in rare diseases research.
- Panel members encouraged PCORI to require open-access publishing.
- Panel members suggested a few dissemination strategies:
    - Designing PCORI's website to pull studies and findings like Google. For example, if a user searches for "Sickle Cell Disease," it would be helpful if all materials with tags related to that condition appear rather than just the title of a study.
    - PCORI could consider, as a cost-effective option, requiring project leads to present findings via whiteboard stories.
    - Lastly, panel members suggested funding secondary publications specifically designed for patients and the communities involved in addition to primary publications.
- Panel members recommended adding a few circles, such as "results return" and "dissemination" to the *Roadmap to Impact* diagram, which consists of the pathway from study completion to results uptake and use. One panelist suggested using the Multi-Regional Clinical Trials Center web tool to assist with returning individual research results to research participants.
    - More detailed information on PCORI's *Roadmap to Impact* can be found here.

What else would you like to learn about Results Release, Uptake, and Use in the future?

The panelists did not directly address this question during this meeting; however, it may serve as a future point of discussion.

Building Capacity for Cross-Cutting Rare Disease Research

- Mat Edick, PhD, Center for Strategic Health Partnerships, Michigan Public Health Institute (MPHI); Advisory Panel Chair

Mat Edick summarized his family's journey with a rare disease. The experiences of Edick's son, Elijah, changed Edick's mentality about research from both a participant and a researcher perspective. The MPHI manages several studies with heavy stakeholder involvement, including the Inborn Errors of Metabolism Collaborative (IBEMC). IBEMC includes several projects focused on patient-centered outcomes, such as the QUEST: IBEM with Uncertain Clinical Consequence Project and a project focused on Phenylketonuria. Edick outlined many challenges that his group faced when applying for PCORI funding due to small sample sizes and lack of evidence around rare diseases. These obstacles led his group to consider cross-cutting studies in rare diseases (e.g., mental health in rare disease) and ultimately to design the Roadmap for Rare Disease Stakeholder Engagement. Edick acknowledged the challenges and barriers to engaging stakeholders in rare disease research, such as a lack of funding and inconsistent expectations from funders.

In response to panelists' questions about convening diverse stakeholders, Edick emphasized the need to listen to every person, recognizing that all come from different perspectives. He also noted the value of skilled facilitators and the challenge of retaining engagement.

Healthcare Cost and Value

- Greg Martin, Acting Chief Engagement and Dissemination Officer, PCORI
- Kristen Giombi, PhD, Research Economist, RTI International

PCORI recently developed a project studying healthcare cost and value with the aim of supporting understandings of "patient-centered value" across stakeholder groups, in PCOR, and in policy development. After reviewing the objectives of the project, Greg Martin discussed the project's progress on conducting a landscape review of the meaning of patient-centered value in health and health care, and hosting convenings with patient and stakeholder representatives via key informant interviews, small focus groups, and large group discussions. The findings from these activities will be described in a report that will be delivered to the PCORI Board of Governors. Key themes emerged during the discussion from the following questions:

Do the directions, activities, and goals resonate with you?

The panelists did not directly address this question during this meeting; however, it may serve as a future point of discussion.

Will the activities meet the informational needs of your stakeholder community?

- Noting significant variance across different communities, panelists wondered if these activities will sufficiently meet the needs of understanding patient-centered value for all PCORI's diverse stakeholders. Martin offered to present the findings as case studies, and Giombi cited a specific conversation with the epilepsy community.

What additional considerations should be kept in mind as we proceed?

- The trust-building needed to gather information about the meaning of value from stakeholders
- Panelists also noted that the research valued by stakeholders can change based on circumstances (e.g., research on the SARS genome).

Update on Strategic Planning: PCORI's Strategic Plan

- Laura Rodriguez, PhD, Deputy Executive Director for Strategy and Planning, PCORI

Laura Rodriguez reviewed the strategic planning process, outlining the substantive components and the cornerstones of the Strategic Plan: the National Priorities for Health and the Research Agenda. She emphasized the importance of these priorities as PCORI plans its next decade of work. She acknowledged the importance of stakeholder input that informed the Research Agenda, including input provided by the RDAP at the previous advisory panel meeting. PCORI is now finalizing the full strategic plan document for review by the Board of Governors. Rodriguez also discussed accessibility and communication strategies for disseminating the strategic plan once it is final. Next steps include implementation planning and collecting input on opportunities related to the Research Agenda. A discussion followed:

What are meaningful ways to engage with stakeholder communities represented on this panel to solicit ideas and inform topic development (e.g., convenings, surveys, webinars)?

- Attend the annual meetings of organizations and professional associations working directly with rare disease communities to develop meaningful relationships and ensure PCORI's priorities align with those of the communities they serve
- Ensure accessibility in many forms (e.g., using plain language or sign language interpreters)
- Regarding dissemination, it may be helpful to think beyond journal articles or consider starting a journal for PCORI awardees.
- Add articles to PubMed, given the weight it holds with researchers
- Findings from PCORI-funded studies need to be shared with many different communities, which requires a broad dissemination strategy.
- Value key partnerships—leveraging those that already exist and intentionally creating partnerships with those not yet engaged with PCORI

What does a responsive, transparent Research Project Agenda development process look like from your perspective?

The panelists did not directly address this question during this meeting; however, it may serve as a future point of discussion.

Given the broad, integrated nature of the adopted National Priorities for Health and Research Agenda, how could PCORI demonstrate relevance to stakeholder communities represented by this panel?

The panelists did not directly address this question during this meeting; however, it may serve as a future point of discussion.

Based on this panel's expertise in CER for rare disease and engaging with the rare disease community, what are the panel's suggestions to stay up to date on emerging issues in this space?

- Given smaller populations of people with rare diseases, related work often spans countries. Noting this distinct concern for the rare disease community, PCORI will continue to consider this issue, and panelists are encouraged to continue raising questions on it.

Acknowledgments and Recap

- Mat Edick, PhD, Director of the Center for Strategic Health Partnerships, MPHI, Advisory Panel Chair
- Doug Lindsay, Founder, Doug Says LLC Advisory Panel Co-Chair

Mat Edick and Doug Lindsay recapped each topic covered during the meeting and emphasized their excitement regarding PCORI's recent announcements and new awards. They recognized Advisory Panel members ending their tenure, including member Saira Sultan and co-chair Doug Lindsay. Co-chairs thanked PCORI staff and the presenters for being true champions pushing for rare disease research.