MEETING SUMMARY

Advisory Panel on Rare Disease Winter Meeting

December 16, 2020

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OVERVIEW

On December 16, 2020, the PCORI Advisory Panel on Rare Disease (RDAP) held its winter meeting virtually.

RDAP's 14 members include patients, caregivers, representatives of patient advocacy organizations and industry, clinicians, and researchers. The meeting was open to the public via webinar, and the agenda for the meeting was posted to the PCORI website in advance.

The meeting started with brief introductions of the panel members, the PCORI RDAP staff team, and an overview of the meeting agenda. Panel members received presentations on Engagement Awards and Rare Disease Organizations: Supporting Engagement in PCOR/CER; Maternal Mortality and Intellectual Disabilities: Overview and Discussion of Issues Related to Rare Diseases; and Identifying our National Priorities: Relevance for Rare Disease Populations.

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

Karen Martin, Program Director, Engagement Awards, started the meeting with a quick snapshot of her team and introduced the objectives of the presentation. She described the standard Engagement Award funding opportunities every regular cycle (i.e., capacity building, dissemination initiative, and stakeholder convening support), their objectives, funding limits, and durations, and funding announcements with a focus on special topics such as the one in May for COVID-19.

Martin presented a list of topics/areas that are out of scope for an Engagement Award project and described the evolution of funding opportunities in the Engagement program. She also acknowledged the feedback from the RDAP during the spring meeting and thanked them for it.
Martin presented three key questions and opened the floor to discussion:

- What are the needs of smaller organizations (e.g., community-based organizations, patient/caregiver/advocacy organizations, associations, etc.) regarding 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 are not 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?

Discussion

RDAP members raised the important concern around IRB issues in the case of Engagement Awards, the benefits of videos to help research participants understand how to be a part of a project and the different roles in research, and the fact that rare disease patient organizations have access to clinicians/researchers but have trouble with reviewing budgets and contracts.

RDAP members suggested the following ideas for PCORI to consider:

- Share information with rare disease organizations about the evolution of Engagement Awards
- Share PCORI funding opportunities with smaller organizations
- Leverage advisory panelists to disseminate information on Engagement Award opportunities and different funding mechanisms
- Understand the needs of smaller organizations and develop opportunities for them to compete for capacity building awards
- Develop a well-defined mechanism for smaller organizations to work with larger organizations with capacity
- Make the point-of-entry for applicants easy, explore an interim step of a Letter of Intent to help interested organizations think through their questions
- Develop an intermediate grant to help organizations develop infrastructure and pursue a stepwise approach to get rare disease groups ready to apply for large awards
- Evaluate the criteria both for applicants and awardees and distinguish “nice to have” from “need to have,” and articulate the value of reporting requirements
- A set of YouTube tutorial videos to walk people through awards and applications (section by section)
- Promotion/advertisement of program fit calls

RDAP members also suggested that PCORI consider the following organizations to engage in outreach efforts: NORD, Global Genes, Rare Epilepsy Network, Child Neurology Foundation, the Haystack Project, Genetic Alliance, Faster Cures, Rare Disease Legislative Advocates, and Every Life Foundation.

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

Els Houtsmuller, Associate Director of the Healthcare Delivery and Disparities Research (HDDR) program, started the presentation by providing an overview of how the two research priorities of (a) Maternal Morbidity and Mortality and (b) Intellectual and Developmental Disabilities (MMM and IDD) will be addressed. She presented the maternal mortality framework and introduced the panel to a special area of emphasis included in the Broad funding announcement—Increasing Access to and Continuity of Patient-Centered Maternal Care—and a list of categories of stakeholders that PCORI has engaged.

Kelly Dunham, Senior Manager, Strategic Initiatives, Office of the Chief Science Officer, provided background information on IDD, including its definition and an overview of PCORI’s approach to it. She presented and explained the International Classification for Functioning Disability and Health (ICF) model, a list of organizations PCORI has connected with regarding IDD, and a special area of emphasis included in the Broad Cycle 3 2020 and Cycle 1 2021: Improving Care for Individuals with Intellectual and/or Developmental Disabilities Growing into Adulthood. Dunham presented a snapshot of cross-cutting areas that have emerged for future research and asked the panel if there are other cross-cutting areas they could suggest that would benefit the rare disease community. She asked the RDAP whether there are examples from rare disease research that PCORI can learn from to support MMM and IDD research, and which other components and key features of the research plan are critical to success.

Discussion

Scott Berns noted that determinants of health are critical in both MMM and IDD. In terms of IDD, he added that expertise in relational health (caregiver-child dyad and the interplay) is important. Berns also spoke about Title V in the maternal and child health arm at the Maternal and Child Health Bureau (HHS), which has a block grant program for states with a significant amount of funding for children and youth with special healthcare needs.

Danielle Boyce shared that her son has severe IDD secondary to epilepsy and suggested a rare epilepsy group, which represents many different developmental encephalopathic epilepsies, for PCORI to connect with. She also provided the link to a paper she wrote on transition from pediatric to adult care that involved a focus group study among caregivers of individuals with Dravet syndrome. Furthermore, Boyce noted that caregivers of children with IDDs often slip through the cracks with some prominent transition programs.

RDAP members suggested the following ideas for PCORI to consider:

- Disability Twitter or met Twitter for reaching communities with IDDs and relevant organizations
- Engage with smaller patient organizations in addition to large national organizations
- Leverage PCORI Ambassadors involved in rare disease in identifying cross-cutting issues
- The cross-cutting topic of sleep
- Engaging midwives as the network is powerful, especially in rural communities

RDAP members also suggested that PCORI explore the following organizations and initiatives:

- The National Network of Perinatal Quality Collaborative
- The Maternal and Child Health Bureau
- Health Resources and Services Administration (HRSA) Alliance for Innovation in Maternal Health (AIM)
- The National Institute of Child Health and Human Development (NICHD)and its maternal mortality initiative
- The American College of Obstetricians and Gynecologists (ACOG)
- The U.S. Department of Health and Human Services (HHS) Office on Women's Health's partnership with Premier, Inc. around the use of their data engine to do some performance improvement around MMM
- The Centering Healthcare Institute, a well-thought-out and successful community-driven model for maternal health care

Identifying our National Priorities: Relevance for Rare Disease Populations

Laura Lyman Rodriguez, Interim Chief Program Support Officer and Senior Advisor to the Executive Director, presented the original strategic framework developed by the Board of Governors, and the revised strategic framework for the next 10 years, with national priorities now focused on goals for health rather than research priorities.

Rodriguez added that she would like to hear from the RDAP regarding what PCORI should be considering as highest-priority areas for PCORI to focus on. She acknowledged the feedback received from the RDAP during the June 2020 meeting and posed the following questions to the panel:

- Are there areas that are missing from 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 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?

Discussion

RDAP members raised the following important points:

- Refocusing from disparity reduction to health equity
- Suggestions to study the impact of COVID, prioritize telehealth, and prioritize behavioral health among children
- Telehealth component of COVID-19 has afforded patients access to experts across the country for consultations and guidance on how to assemble their team. Improving access to expertise for rare disease patients has a high return on investment, even beyond COVID-19.
- Challenge in research that white women are the ones primarily engaged. It is important to educate the PCORI community on engaging other communities and bolstering efforts in education.
- Lack of involvement of patients in the dissemination of research affects patient engagement in shared decision making.
- It is also important to think of what a clinical encounter looks like and make sure dissemination of research involves different types of clinicians.
- Providers on the front lines of rare disease diagnosis are critical stakeholders to access in order for the rare disease community to be better informed and have access to useful resources. The issue of clinicians missing rare disease diagnoses early on is an area that PCORI needs to focus on in some capacity.

Acknowledgments and Recap

Scott Berns informed the panel of the post-meeting survey, upcoming meeting in Spring 2021, and thanked the PCORI staff team for putting the meeting together.

Scott Berns and Doug Lindsay provided a recap of the meeting and the topics covered.

Nora McGhee thanked everyone for joining the meeting and reminded them of the post-meeting survey. She also informed the panelists that they can email additional feedback to Rohini Mohanraj.