External Meeting Summary

Advisory Panel on Rare Disease – Spring Meeting

May 14, 2021

[Additional Background on the Panel](#) | [Meeting Details and Materials](#)

Overview

On May 14, 2021, PCORI's Advisory Panel on Rare Disease (RDAP) convened virtually via GoToWebinar. The meeting started with program updates from the PCORI team, which transitioned into several presentations on active rare disease research awards. Next, there was a presentation and discussion regarding engagement awards and rare disease organizations. Following that briefing, PCORI staff facilitated a discussion of the updated draft national priorities, sought input regarding the relevant areas within these priorities for a focus on rare disease populations and provided an update on the cost-data provision. The last hour was dedicated to a conversation about COVID Connect and PCORI's investment and initiatives related to the COVID-19 pandemic, followed by acknowledgements and a farewell to departing panel members. Below, we provide a brief summary of presentations as well as key themes from each discussion throughout the agenda.

Meeting Agenda

Welcome, Introductions, and Setting the Stage

- Scott Berns, RDAP Chair
- Doug Lindsay, RDAP Co-Chair

Scott Berns welcomed the other panelists to the meeting and walked through the agenda for the day. Scott and Doug Lindsay, both introduced themselves and then facilitated full panel introductions.

Introduction of RDAP PCORI staff, RDAP Program Staff Update, Upcoming Activities

- Carly Khan, Program Officer, Healthcare Delivery and Disparities Research, PCORI
- Nora McGhee, Senior Program Officer, Clinical Effectiveness and Decision Science, PCORI
- Rohini Mohanraj, Program Associate, Research Infrastructure, PCORI

Nora McGhee provided some brief updates on PCORI's rare disease-related work, including the approval of $19 million in funding for four comparative effectiveness research (CER) studies focused on rare diseases. This work is funded through the [Limited Competition PCORI Funding Announcement: Conducting Rare Disease Research using PCORnet®.](#)

Rare Disease Research Awards: Study Highlights and Discussion

PCORI staff:

- Penny Mohr, Acting Program Director, Research Infrastructure
- Jason Gerson, Senior Program Officer, Clinical Effectiveness and Decision Science

Research teams:

- Comparative Effectiveness of Palliative Surgery vs. Additional Anti-seizure medications for Lennox-Gastaut Syndrome: Anne Berg, Tracy Dixon-Salazar, Sandi Lam
- Preserving Kidney Function in Children with Chronic Kidney Disease (PRESERVE): Michelle Denburg, Aneta Jovanovska, Becka Levondosky
- Utilizing PCORnet to support transition from pediatric to adult centered care and reduce gaps in recommended care in patients with congenital heart disease: Thomas Carton, Anitha John, Scott Leezer
- Comparative Effectiveness Research for Neuroendocrine Tumors (CER-NET): Michael O'Rorke, Josh Mailman
- Development of Methods to Improve Identification of Patients with Rare or Complex Diseases: Daniel Herman, Jinbo Chen

Penny Mohr reviewed the rationale, goals, and timeline of the Limited Competition PCORI Funding Announcement: Conducting Rare Disease Research using PCORnet®, launched in 2020 and provided a high-level overview of the four award winners and their areas of focus: 1) Lennox-Gastaut syndrome; 2) chronic kidney disease; 3) congenital heart disease and 4) neuroendocrine tumors. Jason Gerson reviewed the standing funding opportunity related to improving methods for conducting patient-centered outcomes research and introduced the recently awarded study on EHR prediction models for rare disease.

During the discussion, one panelist emphasized the importance of Lennox-Gastaut research and noted the difficulty but importance of evaluating quality of life for Lennox-Gastaut syndrome patients and caregivers. Another panelist noted the prevalence of adult congenital heart disease among pregnant women and thought there were possible opportunities to use newborn screening to track inheritability. Another panelist advised on giving providers evidence behind prediction models to encourage their application in clinical settings. Multiple panelists flagged that all research projects must consider health equity, whether that be working with a variety of patient groups or very carefully using health IT prediction models.

Because most of the projects discussed utilize PCORnet, there was a discussion about how to get support using PCORnet through the web-based access point called the PCORnet Front Door.

Engagement Awards and Rare Disease Organizations: An Update and Project Highlights

- Karen Martin, Program Director, Engagement Awards, PCORI
- Jennifer Canvasser, Founder and Director, NEC Society

Karen Martin provided an overview of the PCORI Rare Disease Engagement Portfolio. She shared that PCORI is currently funding 64 projects with a rare disease focus; among the awardees, 75% are patient-led organizations. Karen provided a few examples of collaborative projects between large institutions and patient groups.

Jennifer Canvasser presented on her experience with PCORI Engagement Awards. Her organization, the NEC Society, has received three Engagement Awards since 2017, collaborating with multiple universities. She was especially proud of her organization's capacity- building work, which was completed as a result of receiving an Engagement Award.

Multiple panelists appreciated that the Engagement Awards have integrated previous feedback from the RDAP. Several individuals acknowledged that the latest engagement awards allow patient advocacy organizations to define research questions of interest to them, and then identify an academic partner.

A panelist whose organization had previously received an Engagement Award commented that, while these awards help small organizations to build research infrastructure, ongoing support to sustain or advance it is not available through PCORI.

Identifying Our National Priorities: Relevance for Rare Disease Populations

- Laura Lyman-Rodriguez, Interim Chief Program Support Officer and Senior Advisor to the Executive Director, PCORI

Laura Lyman-Rodriguez provided an update on the Board of Governors' progress toward developing PCORI's national priorities for the next 10 years. Following the update, multiple panelists noted the importance of PCORI applying a health equity lens to its work. Two panelists suggested that, in addition to health equity as a standalone priority, each national priority should incorporate an aspect of health equity. Another panelist noted that there is a lack of diversity and representation in evidence generated from clinical trials. In light of disparate prioritization strategies across states, one panelist suggested PCORI consider taking a role in future vaccine rollout efforts with an eye toward both health equity and representing the rare disease community; another panelist suggested PCORI provide additional oversight on use of digital therapeutics. Two panelists suggested facilitating research on more equitable genetic scoring technology within the potential national priority of emerging innovations.

An Update on the Cost-Data Provision

- Andrew Hu, Director of Public Policy and Government Relations, PCORI

Andrew Hu provided an update PCORI's implementation of the cost-data provision, which emphasizes capturing costs among the full range of outcomes measures, where appropriate. Now, in addition to clinical effectiveness outcomes and patient-centered outcomes, PCORI will allow investigators to capture economic burden and economic impact data related to the utilization of healthcare services. Mr. Hu elaborated on additional pillars that would guide implementation and stressed that PCORI would still hold patient and community feedback at the center of this work. Multiple panelists commented that any project team managing economic impact would have to be multidisciplinary.

COVID Connect: PCORI’s Response to the COVID-19 Pandemic

- Claudia Grossman, Senior Program Officer, Research Infrastructure, PCORI

Claudia Grossman introduced the panel to COVID Connect, PCORI’s interdepartmental workgroup focused on addressing the pandemic and coordinating future research efforts related to COVID-19. Additionally, she walked through PCORI’s current COVID-19 portfolio, which primarily consists of 122 COVID-19-related enhancements to existing research, and targeted funding announcements specifically for COVID-19 research. Following the presentation, multiple panelists commented that many rare disease patients were not properly prioritized for resources like ventilators or vaccines; PCORI may want to research how resources are prioritized for future reference. One panelist mentioned that people with rare diseases often have to travel to specialists that are out of network. However, during the pandemic, many rare disease patients were able to use telehealth services and avoid out-of-network fees due to government waivers. PCORI may want to research the impact of waivers on rare disease treatment options.

Acknowledgments and Recap

Scott Berns and Doug Lindsay summarized the day’s presentations and themes.

Doug Lindsay and PCORI staff acknowledged the six departing members of RDAP – Roxanna Bendixen, Vanessa Boulanger, Julie Gortze, Tilicia Mayo-Gamble, Sherene Shalhub and Scott Berns, and thanked them for their contributions.

Members of the Advisory Panel on Rare Diseases

Chair:
Scott Berns, MD, MPH, FAAP, Chief Executive Officer, National Institute for Children's Health Quality

Co-Chair:
Doug Lindsay, BS, Personal Medical Consultant and Founder, Doug Says LLC

Members:
Sarah Bacon, MS, Rare Disease Patient, Advocate, and Writer

Roxanna Bendixen, PhD, OTR/L, FAOTA, Rehabilitation Scientist, Associate Professor, and Program Director, Department of Occupational Therapy at the University of Pittsburgh

Vanessa Boulanger, MS, Director of Strategic Partnerships, Amyloidosis Research Consortium

Danielle Boyce, MPH, Senior Research Data Analyst, COPD Foundation

Mathew Edick, PhD, Director of the Center for Strategic Health Partnerships, Michigan Public Health Institute

Julie Gortze, RN, Founder, Rare New England

Salman Hussain, MPH, Consultant, Charles River Associates

Tilicia Mayo-Gamble, PhD, MPH, MA, Assistant Professor, Georgia Southern University

Nancy Rose, MD, Adjunct Professor of Obstetrics and Gynecology, University of Utah

Sherene Shalhub, MD, MPH, Vascular Surgeon and Translational Researcher; Director of the Vascular Genetic Clinic, University of Washington

Saira Sultan, JD, CEO, Connect4Strategies

Laura Tosi, MD, Founder and Director of the Bone Health Program, Children's National Hospital

Ex-Oficio Member:
Naomi Aronson, PhD, Executive Director, Clinical Evaluation, Innovation, and Policy, Blue Cross Blue Shield Association (BCBSA)