Advisory Panel on Rare Disease Meeting Summary

August 2014

Overview

On August 13, 2014, the PCORI Advisory Panel on Rare Disease convened for the second time via Web conference to continue discussing the scope of work and priority issues for the newly appointed group.

The Advisory Panel on Rare Disease is made up of 13 representatives, including rare disease patients, caregivers, patient advocacy organizations, clinicians, payers, researchers, and industry. The meeting was open to the public via webinar, and meeting materials were posted to the website in advance of the session.

The meeting was chaired by Dr. Marshall Summar, the newly appointed panel chair, who gave an update to the full panel regarding what the leadership team had discussed during two meetings following the first panel meeting in May. PCORI staff gave several presentations regarding PCORI's topic generation, research prioritization, and merit review processes; a summary of the rare disease topics that have been submitted to PCORI; and a guide to formulate CER research questions. The panel discussed outreach and other strategies to increase the number and quality of rare disease topics and research proposals.

Related Information

- About This Advisory Panel
- Meeting Details and Materials
- About PCORnet's Rare Disease Task Force

The Patient-Centered Outcomes Research Institute (PCORI) is an independent organization created to help people make informed healthcare decisions.

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Introduction and Leadership Team Updates

Dr. Summar introduced the leadership team to the panel, which includes the chair (himself), the co-chair (Vincent Del Gaizo), the ex-officio member from PCORI's Methodology Committee (Naomi Aronson), and the PCORI support staff. In the five weeks since the chair and co-chair were appointed, the team has had two meetings about what the panel should be doing to support PCORI's mission in its first year of existence. Dr. Summar presented the four priority areas for the panel as proposed by the leadership team:*

1) Analyze PCORI processes for conduciveness to rare disease research and guide PCORI on ways to improve these, including:
    a. Topic generation
    b. Research prioritization
    c. Merit review
    d. Outreach
2) Help identify priority rare disease topics
3) Help commission a landscape review on standards for rare disease research, including:
    a. Standards for rare disease registries, including data standards and bio specimens stewardship standards
    b. Rare disease research issues, including evidence evaluation and patient engagement
    c. New treatments for rare diseases and needed CER
4) Evaluate PCORI's rare disease portfolio

**This are not listed in priority order.*

Following the leadership update, PCORI staff and two panelists provided updates on three rare disease registry projects, discussing encountered issues, milestones, and process overviews. Sarita Wahba presented on PCORnet and the Rare Disease Patient-Powered Research Networks (PPRNs), Russell Teagarden presented on National Organization for Rare Diseases (NORD) efforts, and Yaffa Rubinstein presented on the Global Rare Disease Patient Registery and Data Repository (GRDR). This time was allocated to ensure appropriate collaboration and knowledge exchange between the panel and PCORnet's PPRNs, NORD, and GRDR.

PCORI's Processes and Rare Disease Topics and Proposals

PCORI staff gave several presentations on PCORI's Topic Generation and Research Prioritization (TGRP) and merit review processes. PCORI staff also presented on submitted rare disease topics and on a new guide to formulate a CER question. Following these presentations, the panelists discussed the following topics:

1) **Ad hoc advisory panels**: The panel discussed the need to place these potential ad hoc panels in the most strategic time in PCORI's merit review process. The role of the ad hoc panels will be to

improve the quality of the rare disease research PCORI funds. Input could occur before the Board meeting during which it approves the slate, and/or after the award is made as a consultation role to help implement the research plan. Selecting the point within these processes will be a very strategic decision.

2) **PCORI Funding Announcement (PFA) development:** The panel discussed providing guidelines for those applying for funding to increase the level of quality of the applications received, and to solicit targeted applications. It was also suggested that PCORI could create a different merit review track for rare disease applications, which could go through a specialized review.
3) **TGRP process improvement:** It was suggested that PCORI could require inclusion of outcomes important to rare disease patients in the Improving Healthcare Systems PFAs.
4) **Cross-cutting vs. condition-specific rare disease topics:** Panelists advised that PCORI will need to balance its portfolio between these two types of rare disease research. Three types of cross-cutting categories were identified: symptoms, care navigation, and social environments. The cross-cutting research questions will be critical to rare diseases with very small populations, as it will be methodologically complex to conduct research on these. Based on the list of submitted rare disease topics and Naomi Aronson's presentation on cross-cutting issues, panelists could find commonalities in the issues faced by rare disease patients, then suggest cross-cutting topics to include in the topic database.
5) **CER guide and outreach:** Panelists advised that this guide should be available to patients as a tool for them to submit feasible and relevant CER topics for PCORI to consider. Patients might also need help in developing these topics, and PCORI could offer to connect them with appropriate experts to develop their concerns into research questions that PCORI would be interested in funding. International outreach will be essential to ensure that rare diseases with very low prevalence will be accounted for. PCORI could also reach out to the rare disease topic submitters and invite them to reformulate their previously submitted topics into CER questions. PCORI's Engagement Awards encourage active involvement of patients, caregivers, clinicians, and other healthcare stakeholders as integral members of the research process, and it was suggested that PCORI could carry out targeted outreach to the rare disease community to apply for funding.

   It might also be helpful to analyze the type of rare disease applications PCORI has received and characterize who from the rare disease community has submitted them to evaluate if the current application process is accommodating.
6) **Challenges of PPRNs and CDRNs to meet milestones:** It was suggested that the panel should provide guidance to the PCORnet Rare Disease Task Force regarding the list of issues faced by PPRNs and CDRNs.

Next Steps

The next panel meeting will be held on October 7, in-person, in the Washington, DC, area. During this meeting, the panel will discuss and formulate cross-cutting CER questions and build strategic outreach plans. Additionally, the PCORnet Rare Disease Task Force leaders will attend to discuss collaboration opportunities.