Advisory Panel on Rare Disease Meeting Summary

April 2014

Overview

On April 30, 2014, the PCORI Advisory Panel on Rare Disease convened for the first time in Alexandria, Virginia, to begin 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, and 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.

At the meeting, PCORI staff gave several presentations to provide specific information about the existing rare disease projects in the PCORI portfolio, the rare disease roundtable, and PCORnet, which gave the panel the opportunity to start talking about how it will guide PCORI's work. The panel discussed and generated a list of topics and issues for consideration as possible priority action items. Finally, the panel went over several organizational issues and made decisions regarding meeting scheduling and frequency.

Related Information

- About This Advisory Panel
- Meeting Details and Materials
- About PCORI's Rare Disease Roundtable
- About PCORnet
- About PCORnet's Rare Diseases Task Forces
- About PCORnet's Biorepository Task Forces
- Register for the June 17 Board of Governors Meeting

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

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Introduction

The Advisory Panel on Rare Disease generated a list of topics and issues for consideration as priority action items.

Setting the Stage

Prior to their first meeting, each panelist was asked to complete the PCORI New Panelist Training, which provided fundamental information about PCORI and its processes and procedures. To kick off the meeting, Dr. Bryan Luce, PCORI's Chief Science Officer, gave an overview of the panel's charter, which highlighted the panel's purpose, its scope of work, and the role and composition of its future ad hoc panels.

Then, Greg Martin, PCORI's Deputy Director for Stakeholder Engagement, gave an overview of the Rare Disease Roundtable, which five of the new panelists had attended. This roundtable was organized by PCORI in September 2013 to seek to leverage the wisdom and experience of the rare diseases community—patients, patient advocates, industry, payers, researchers, and policymakers—to help guide development of an innovative and patient-centered research agenda to improve healthcare delivery and patient outcomes. Mr. Martin went over the meeting's objectives, discussions, and the advice and recommendations that were given to PCORI.

Following this presentation, Dr. Stanley Ip gave an overview of the rare disease projects that PCORI has already funded, and covered three selected projects in detail.

Finally, Dr. Rachael Fleurence presented on PCORnet, a large, highly representative, national network for conducting clinical outcomes research. PCORnet also has a Rare Diseases Task Force to support CDRNs and PPRNs in identifying populations, developing research priorities, as well as designing and implementing studies for rare diseases.

Foundational Issues for Priority Consideration

Throughout the day, the panel discussed the following wide range of topics and foundational issues for consideration as priority action items (these are not listed in priority order):

1. **Rare disease registries:** Several panelists identified a multitude of issues regarding rare disease registries on which the panel could provide guidance. Panelists were interested in providing guidance to those interested in starting a rare disease registry. This guidance would provide a roadmap of all the issues that would need to be tackled and recommendations for the best practices to adopt.

These include: usability, sustainability, ethical implications, ownership in the long term, governance, data linkage to other registries, legacy issues, Common Data Elements (CDEs), standardization of data, data accessibility, IRB issues around long-term registries, IRB standardization across multiple institutions, and disease definitions.
Rare disease registries was also identified as a potential topic for an ad hoc panel.

2. **Minimal datasets, data standards for rare disease registries**: Provide guidance on standards of evidence and common datasets and leverage existing resources that have been validated and used nationally and internationally. Some panelists were particularly interested in quality of life outcomes. It was also suggested that standards could be established for what is most useful from different approaches, such as diagnostic or therapeutic, or for different types of data, such as symptoms or outcomes.
   These data standards would allow for cross-comparisons between rare disease registries.
   This work could involve collaboration with PCORI's Methodology Committee and Advisory Panel on Clinical Trials.
3. **Landscape review on rare disease research issues:** Commission a rare disease research landscape review, which would provide a solid starting point for the panel. This review could be a paper or a database or a combination of both. It could provide a gap analysis to shed light on what the panel should focus on.
4. **Patient proxy:** Work on the implications of the active involvement of the patient's family and other caregivers, which often takes place over the direct involvement of the patient.
5. **Biospecimens:** Address the issue of collecting biospecimens for rare diseases, to be able to connect this dataset to the patient-reported data and the clinician-reported data.
6. **New treatments:** Provide guidance on the type of evidence and standards needed when new treatments are introduced to the rare disease world. These standards should define what is useful from a diagnostic or therapeutic approach to be able to provide buy-in for third-party payers.
7. **Evaluating evidence:** Propose a new type of evidence grading system for rare disease research. This could provide credibility to different forms of evidence to improve access for patients and clinicians. This work could involve collaboration with PCORI's Methodology Committee.
8. **Cross-validation:** Provide guidance on the cross-validation of patient- and family-reported data versus investigator-reported data to test data validity. This could shed light on what patients and their families identify as an improved quality of life, and subsequently the type of trials that will yield needed results. Panelists suggested that researchers with qualitative data expertise, such as social science researchers and computational linguistics researchers, would be key in providing this kind of useful data.
9. **Missing data and dosage:** Tackle the issue of missing data to determine appropriate dosing schedules. This is often of the utmost importance to rare disease patients' quality of life. Understand that approved dosing may be able to be altered based on "actual" clinical care. Obtain evidence in support of alternative, often "real life," dosing schedules.

10. **PCORnet:** Collaborate with the task forces (in particular the Rare Diseases and the Biorepository Task Forces) and the rare disease PPRNs, and guide them on the foundational issues that the panel might work on. PCORnet was also identified as a potential topic for an ad hoc panel, which would provide an efficient exchange of information.

PCORI's Portfolio and Rare Diseases

The panel discussed possible ways that it could guide PCORI on which rare disease research to fund (these are not listed in priority order):

1. **Evaluate the topic prioritization process:** Advise PCORI on whether the current process is consistent with a partial emphasis on rare diseases.
2. **Review rare disease topics:** Consider and review rare disease topics that have been previously prioritized by the priority-setting advisory panels, or have been submitted through the various channels PCORI has in place, and advise PCORI on which of these should be pursued. Panelists also suggested that a certain number of panelists could benefit from meeting with the leadership of the priority-setting advisory panels.
3. **Identify new rare disease topics:** Put in place a process by which each panelist could use their networks to identify potential research topics or questions that the whole panel could prioritize for future vetting.
4. **Evaluate the merit review process:** Make sure that the PCORI merit review criteria are appropriate for rare disease projects to make it through the process.
5. **Provide a consultation service for funded rare disease projects**: Better define protocols and help monitor projects.
6. **Evaluate PCORI's evaluation framework:** Review how PCORI has defined usability to make sure that it will also apply to rare disease research.
7. **Advise PCORI on how to fund rare disease research:** Provide advice on the process for rare disease research—for example, by proposing a deep targeted reach mechanism by which PCORI could identify the best research team to answer a specific research question, or by identifying the best methods to conduct rare disease research.
8. **Advise PCORI on collaborating with other rare disease organizations:** Note: The panel has already provided a long list of rare disease organizations that PCORI could collaborate with.
9. **Advise PCORI on international collaboration:** Provide guidance on how best to collaborate with international research teams as rare disease research requires such partnerships.
10. **Advise PCORI on engaging and collaborating with the biotech and pharmaceutical industries.**

Organizational Issues

Dr. Bryan Luce invited each panel member to submit nominations, including self-nominations, for the panel's chair and co-chair. The panel also discussed meeting frequencies and agreed that two to four in-person meetings a year would be appropriate, with additional webinars in between the in-person meetings. It was suggested that the panel have a webinar meeting during the summer.

Next Steps

Proposed panel chairs will be presented to the Board of Governors on June 17, 2014 for approval during a public webinar. Following the meeting, the first panel leadership meeting will take place to prioritize the key ideas for possible activities that were generated during the spring meeting. The panel will then meet in person in the fall for their second meeting. The panel also will be starting to think about establishing its ad hoc panels.