Advisory Panel on Rare Disease Meeting Summary

Overview

On October 7, 2014, the PCORI Advisory Panel on Rare Disease convened for the third time to discuss PCORI's focus on rare diseases.

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

The meeting's chair, Dr. Marshall Summar, provided an update to the Panel on the collaboration with the Advisory Panel on Clinical Trials. PCORnet's Rare Disease Task Force leader, Dr. Rachel Richesson, presented on the work of her Task Force. Marshall Summar presented the first draft of the *Rare Disease Research Guide for Merit Reviewers*, and PCORI staff presented on PCORI's focus on rare diseases and cross-cutting CER topics. 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
- Advisory Panel on Rare Disease Winter 2015 Meeting

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

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Discussion and Recommendations to PCORnet's Rare Disease Task Force

Dr. Rachel Richesson presented on PCORnet's Rare Disease Task Force to include the efforts undertaken by PCORnet towards registry formation and disease definitions and the Task Force's efforts toward rare diseases. The panel developed the following recommendations for the Task Force's attention:

1. **Duplication** of registry efforts was identified as a major threat since a number of national and international organizations are working on similar goals. Panel members strongly recommended a proactive effort to use existing tools and resources and coordination with groups doing related work (e.g. NORD, NIH/NCATS GRDR[SM], EIMD, IRDiRC). They also recommended the Task Force contact and coordinate with Dr. Yaffa Rubinstein of NIH and Ms. Pam Gavin of NORD and invite them to work directly with the PCORnet Rare Disease Task Force. Members expressed the belief that collaboration could significantly accelerate the timeline around some of the PCORnet milestones and recommended that this happen as soon as possible.

2. Topics to consider developing **best practices** around were identified:

    a. Patient outreach
    These best practices should include when and how patients are identified or informed about a study, and processes to guarantee data safety and privacy protection. Panelists expressed the belief that a clear statement on privacy protections will increase participation and also pre-empt concerns. Patients should receive clear information prior to having to report any data. In addition, patients and their advocates, with better understanding of basic scientific issues (e.g. clinical trials design, biomarkers and their terminologies) can be more effective partners in the process of improving their quality of life and developing therapeutics for their disease.
    b. Electronic Health Records (EHRs) data collection and analysis
    These best practices should be especially written with respect to extending beyond financial reporting systems and their limitations (e.g., claims, ICD-9/10, HCPCS codes frequent use of 'Not otherwise specified' or 'Not otherwise classified' codes in rare diseases). They should also point to the development of structured data elements and well-defined 'computable phenotypes' for rare diseases, possibly utilizing natural language processing, machine learning, and/or computational linguistics.
    c. Screening techniques/protocols
    These best practices should cover screening techniques and protocols to identify patients and track them across studies/registries and countries (while maintaining an appropriate level of privacy)

3. The panel also encouraged the Task Force to update the panel on the progress to achieve its defined **milestones** to enhance the panel's ability to advise the Task Force.

4. Given the small size of rare disease patient populations panel members encouraged the Task Force to consider how the Rare Disease PCORnet registries and their uniformity will conform or integrate with other **global initiatives** and standards.

5. Panel members encouraged the Task Force to consider putting a process in place to **disseminate** the standards and best practices developed to researchers, patients, and other working groups.

PCORI's Focus on Rare Diseases

Dr. Summar led a discussion about the *Rare Disease Research Guide for Merit Reviewers*, and panelists suggested edits to the document. Panelists expressed that while guidelines were necessary to raise awareness of research design limitations that challenge rare disease investigators, overly prescriptive guidelines could result in further hurdles to the selection of rare disease proposals for funding.
The Panel also discussed the following ways to increase the amount and quality of applications for rare disease research awards received by PCORI:

1. Increasing the "grantsmanship" of the proposals by creating a matchmaking process to connect the rare disease community with experienced grant writers.
2. Recruiting merit reviewers with expertise in rare disease, thereby giving rare disease proposals—which are usually driven by necessity and written by people without an institutional background—a chance for funding when compared to proposals dealing with more common diseases. These reviewers should also be able to recognize ingenious and audacious proposals that don't perfectly fit the traditional and formal formats.
3. Creating materials that may provide guidance for rare disease proposal writers, including:
    a. Sample winning rare disease proposals and/or applications
    b. Rare disease proposal strategies for meeting the CER and PCOR requirements and for describing the impact of the proposed study
4. Creating a broad announcement for rare diseases with funding set aside for specific rare disease projects. This conversation was tabled until a well-crafted and thoughtful recommendation could be developed. The group will begin formulation of the recommendation when more information becomes available about the number and success of rare disease applications—information that will inform the best course of future action.
5. Distributing information packets to the target community to increase the rare disease community's awareness about PCORI and relevant and acceptable CER questions, instead of making specific calls for more proposals.

The panel then discussed cross-cutting CER topics. The panel discussed Comparative Effectiveness Research gaps and how rare diseases are framed within comparative research questions. The group generated many ideas for cross-cutting CER rare disease research topics. This list will be used in future communications to potential applicants in the rare disease research community.

Next Steps

The Panel will be further collaborating with the Advisory Panel on Clinical Trials and the PCORnet Rare Disease Task Force. They will also be meeting with the Advisory Panel on Assessment of Prevention, Diagnosis, and Treatment Options during the Winter 2015 Advisory Panel meetings. The panel will be finalizing the *Rare Disease Research Guide for Merit Reviewers*, conducting outreach to recruit reviewers and encourage the rare disease community to apply for PCORI funding. During their January meeting, on January 13, 2015, the panel will review the results from the evaluation of PCORI's merit review process in regards to rare disease proposals and discuss other opportunities within PCORI to engage the rare disease community, among other agenda items.