Recommendations from the PCORI Advisory Panel on Rare Disease (RDAP) to the PCORnet Rare Disease Task Force

Introduction

We were very impressed with the efforts undertaken by PCORnet toward registry formation and definition around all diseases and the task force's efforts related to rare diseases. From our subsequent conversation, we developed several specific recommendations.

Recommendations

1. **Duplication** of registry efforts was identified as a major threat of this and other efforts. A number of national and international organizations are working on similar goals. We strongly recommend 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). We recommend 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. We believe collaboration could significantly accelerate the timeline around some of the PCORnet milestones. We recommend that this happen almost immediately.

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

    a. Patient outreach, including when and how patients are identified or informed about a study, and processes to guarantee data safety and privacy protection. We believe a clear statement on privacy protections will increase participation and also preempt 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, especially 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) and into 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 to identify patients and track them across studies/registries and countries (while maintaining an appropriate level of privacy)

3. To enhance our ability to advise you, we would appreciate receiving updates from the task force on the progress of the PCORnet Rare Disease Task Force toward achieving its defined **milestones**.

4. Given the small size of rare disease patient populations, we would encourage 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. We encourage 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.