



Advisory Panel on Rare Disease Fall 2015 Meeting: Challenges with Producing Reliable Evidence for Rare Diseases Breakout

Background

PCORI's mission is to fund research that will produce robust and reliable information to help patients and clinicians make informed healthcare decisions, and to improve healthcare delivery and outcomes. Developing reliable comparative effectiveness evidence for some rare diseases can be particularly challenging due to specific features of those diseases or the management options available. These features can influence which types of study designs can be feasibly implemented and, as a result, can also influence the type of evidence that can be produced to inform clinical decisions.

It is important for researchers and decision makers, who include patients and their caregivers, clinicians, payers, and policy makers, to understand how these different features of a rare disease impact study design decisions. However, we are not aware of any efforts to create a framework or typology that relates relevant rare disease characteristics to the strength of evidence that can be generated. Such a typology would therefore represent an important advancement in the field. It could be used by researchers to make informed study design decisions and could be used by decision makers to assess the quality of the evidence that exists to support current clinical practice.

Purpose

The purpose of this workgroup is to consider the range of issues associated with producing reliable evidence in the area of rare disease in order to generate a framework or typology that can help decision makers understand what evidence is feasible for specific clinical questions, how to improve and accelerate evidence development, and ultimately how to assess the quality of the evidence that is generated. Questions for this group to consider include:

- *What features of a rare disease impact the ability to generate reliable evidence about treatment options for that condition?*
- *How do each of those features impact evidence generation? How do those features impact which study designs are feasible to implement?*



- *Is it possible and would it be useful to organize those features into a framework or typology to help decision makers and researchers understand what type of study designs can be implemented and what level of evidence can be produced in different situations?*
- *How can we capture considerations of both strength of evidence and the degree of uncertainty and risk that is acceptable in various contexts?*

Breakout Session Structure

10:30 – 11:15 a.m.: Discuss the questions posed above.

11:15 – 11:45 a.m.: Define a preliminary set of objectives for the workgroup.

11:45 – 12:15 a.m.: Develop a set of next steps and consider what information and resources you need to achieve the objectives you have outlined. Specifically, consider the following:

- Are there specific types of expertise that should be represented on this workgroup that are currently missing? If so, can you recommend someone with that expertise?
- In order to inform future discussions, would a synthesis of the existing literature on this topic be useful? If so, what keywords/MeSH terms should be included in the search?
- Are there other resources that you need to carry out this work?

Timeline

November 2015 – January 2016: Refine the workgroup objectives and deliverables and develop an outline for the workgroup document. *At the January 2016 RDAP meeting, time will be reserved for workgroups to meet and review their document outlines.*

January 2016 – April 2016: Draft a document that provides guidance to the rare disease community based on the outline discussed at the January 2016 RDAP meeting. *At the April 2016 RDAP meeting, time will be reserved for the workgroups to discuss the complete draft documents.*

April 2016 – July 2016: Revise and finalize the draft document. *Time will be reserved at the July 2016 RDAP meeting for presentations of the final guidance documents. The goal is to publish the documents produced by each group on the PCORI website and in a special issue of a peer-reviewed medical journal.*