# Advisory Panel on Rare Disease Fall 2015 Meeting: Human Subjects Breakout

## Background

Human subjects issues in the context of rare diseases vary from those encountered in more common conditions due to the small patient populations. Specific issues that arise in the area of rare disease include:

- Privacy issues
  - Rare disease research relies heavily on the creation of international registries to be able to collect sufficient data to generate reliable evidence. Rare disease patients are more vulnerable to privacy issues than others due to the smaller numbers of patients affected, and the standard privacy rules (e.g., informed consent for each data use, right to withdraw) can create barriers to collecting the necessary data.[1] The need for more widespread and less restrictive sharing of the data collected through registries makes implementing privacy protections challenging. As a result, there is a need to consider these challenges related to privacy protections, including which privacy protection measures can be implemented in this setting and how to communicate privacy risks to registry participants. Please refer to the article provided with this document for a more detailed analysis of this issue (also referenced at the bottom of this document).
- Specific informed consent issues
  - Consent processes for enrolling children often require that researchers first get consent from the child's parent and that they also attain assent from the child. However, it is often not clear whether it is necessary to re-consent that child when he or she becomes an adult.
  - Many rare diseases are hereditary, so while it is still important to obtain consent from the individual who will be enrolled in the registry, it is also important to consider the impact their participation may have on other members of their family.

## Purpose

The purpose of this workgroup is to consider how researchers and other stakeholders should handle human subject issues that arise in the context of rare diseases, in order to answer the following questions:

- *What are the most important considerations when developing consent forms for registries enrolling adults with rare diseases?*
- *What are the most important considerations when developing consent forms for registries enrolling children with rare diseases?*
- *What measures can be implemented to protect the privacy of individuals who are enrolling in a rare disease registry?*
- *What are some best practices for developing consent forms and privacy protection measures for a rare disease registry?*
- *What are some best practices to engage patients, families, and/or caregivers in the development of consent forms and privacy protection measures for a rare disease registry?*

## 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.*

## References

1. Mascalzoni, Deborah, Paradiso, Angelo, and Hansson, Matts. 2014. "Rare Disease Research: Breaking the Privacy Barrier." *Applied & Translational Genomics*. 3(2):23–29.