



PCORI's Guidance on Research in Rare Diseases

Background

PCORI funds patient-centered comparative effectiveness research (CER). Patient-centered CER is research that compares two or more interventions that are evidence based and/or that are often used to treat individuals with the medical condition being studied. The goal of this type of research is to answer questions that are important to patients and to provide patients with information to make informed health care decisions.

In addition to funding patient-centered CER focused on common conditions, PCORI is also interested in funding patient-centered CER that addresses outstanding questions about the prevention, diagnosis, and treatment of rare diseases. According to the Rare Diseases Act of 2002, rare diseases (sometimes called orphan diseases) are those that affect fewer than 200,000 people in the United States.

What Type of Rare Disease Research Will PCORI Fund?

Investigators applying for PCORI funding must make the case that the study addresses a realistic clinical choice and decisional dilemma faced by patients and providers. As part of the justification for the importance of the study, investigators should describe how the interventions being studied are currently used in clinical practice for the diagnosis, treatment or management of the condition, both in terms of how often they are used and any particular clinical and population considerations.

PCORI prefers studies comparing at least two interventions. If this is not possible, applicants should describe what the control group will receive and how this will be measured over the course of the study in each patient, and in multiple settings, if applicable. For example, if the control group will receive supportive services, applicants should describe those supportive services and how they will be tracked during the study and accounted for in the data analysis.

Investigators must also describe the existing evidence on the proposed interventions, even if the data are limited - a common issue for interventions for rare diseases. If the data have not been published, investigators should describe why they are not, as well as any plans for the public release of the data. In some instances, PCORI may consider applications that involve interventions with limited evidence *if* the application meets the other criterion described above.

If there are no efficacy data or the proposed interventions are not considered a clinical option for individuals with a given rare disease, the study does not qualify for funding opportunities offered by PCORI. We are interested in funding comparisons of real-world clinical options.



The table below compares PCORI’s requirements for patient-centered CER and describes the differences for common diseases versus rare diseases.

	COMMON DISEASES	RARE DISEASES
COMMONLY USED	The intervention(s) should be used by physicians and/or health care systems across the United States for treatment of individuals with the condition being studied.	The intervention(s) should be considered a realistic clinical choice for individuals with a given rare disease even if the intervention is not widely offered in health care systems across the country since individuals with a given rare disease are often treated in centers that specialize in that disease.
EVIDENCE BASED	The intervention(s) should have been previously studied in at least one adequately powered efficacy study.	The intervention(s) should have been previously studied. PCORI may consider applications that involve interventions with limited evidence if the application meets the other criterion described above.
COMPARATORS	PCORI prefers comparisons of two interventions. If this is not possible, applicants should specifically describe what the control group will receive and how this will be measured over the course of the study in each patient.	PCORI prefers comparisons of two interventions. If this is not possible, applicants should specifically describe what the control group will receive and how this will be measured over the course of the study in each patient.

Examples of Funded Studies

Funded projects focused on a rare disease can be found on the [PCORI website](#).