

PCORnet[®] Resources Are Surfacing Answers for Rare Diseases

PCORnet[®] is intended to improve the nation's capacity to efficiently conduct patient-centered health research, particularly comparative clinical effectiveness research (CER), by providing a large, highly representative network of health data, research expertise, and patient insights. PCORnet has been developed with funding from the Patient-Centered Outcomes Research Institute[®] (PCORI[®]).

SOLVING KEY CHALLENGES IN RESEARCH ON RARE DISEASES

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|  Challenge: Small, geographically dispersed study populations |  Challenge: Lack of natural history data |  Challenge: Alignment of research questions and patient priorities |
|  Solution: Real-world data from everyday healthcare encounters with more than 50 million people annually across the U.S. |  Solution: Access to standardized longitudinal data |  Solution: Patient and caregiver engagement drives meaningful research and adoption of actionable findings |

CASE STUDIES: RARE DISEASES RESEARCH POWERED BY PCORnet[®]

Cognitive Behavioral Therapy and Real-Time Pain Management Intervention for Sickle Cell via Mobile Applications (CaRISMA)

Study Design: Intervention Trial

Funder: Patient-Centered Outcomes Research Institute[®] (PCORI[®])

Principal Investigator: Charles Jonassaint, University of Pittsburgh

Research Question

Does mobile phone-delivered cognitive behavioral therapy (CBT) provide a greater benefit for daily pain and depression in adults with sickle cell disease (SCD) than digital education?

Using PCORnet, the study team

- Analyzed retrospective and prospective clinical data for 350 participants across 5 sites
- Conducted the largest behavioral intervention trial in SCD to date to compare the effectiveness of digital CBT and education to improve quality of life

Dissemination

Published results in [Blood Advances](#) and the [British Journal of Haematology](#)

Comparative Effectiveness of Epilepsy Surgery Versus Additional Anti-Seizure Medications for Lennox-Gastaut Syndrome

Study Design: Retrospective Observational Study

Funder: Patient-Centered Outcomes Research Institute[®] (PCORI[®])

Principal Investigator: Sandi Lam, Lurie Children's Hospital

Research Question

Which type of treatment — more medicines or proceeding with surgery — will have the best outcomes for a child with Lennox-Gastaut Syndrome (LGS)?

Using PCORnet, the study team

- Developed computable phenotypes for LGS to analyze data from 4,000+ patients across 18 sites
- Is conducting the first study to directly compare the outcomes of using additional antiseizure medicine versus epilepsy surgery to treat LGS

Dissemination

- Published the study protocol in [Frontiers in Neurology](#)
- Published findings from surveys of caregivers in [Epilepsy and Behavior](#)

Utilizing PCORnet® to Support Transition from Pediatric to Adult Centered Care and Reduce Gaps in Recommended Care in Patients with Congenital Heart Disease ([CHI-RON](#))

Study Design: Retrospective Observational

Funder: Patient-Centered Outcomes Research Institute® (PCORI®)

Principal Investigators: Thomas Carton, Louisiana Public Health Institute, and Anitha John, Children's National Research Institute

Research Questions

- How does receiving current recommended care affect long-term outcomes and healthcare needs among the numerous rare disease subtypes of congenital heart defects?
- What factors are associated with gaps in recommended care?
- Do patients report feeling better when they remain in specialty care?

Using PCORnet, the study team

- Linked patient-reported outcome data from a patient registry and electronic health record data accessible through PCORnet to assess study outcomes
- Leveraged the PCORnet® Common Data Model to develop an algorithm to tailor recruitment

Dissemination

- Published results in [JACC Advances](#)
- Published a paper on using PCORnet and patient engagement strategies to improve recruitment of research participants in [Medical Care](#)

Preserving Kidney Function in Children with Chronic Kidney Disease ([PRESERVE](#))

Study Design: Retrospective Observational

Funder: Patient-Centered Outcomes Research Institute® (PCORI®)

Principal Investigators: Christopher Forrest and Michelle Denberg, The Children's Hospital of Philadelphia

Research Questions

- Which monitoring strategies and blood pressure medications best preserve kidney function in pediatric patients with chronic kidney disease (CKD)?
- What are the lived experiences of families and patients managing pediatric CKD?

Using PCORnet, the study team

- Included 20,100 children (ages 1–17) with CKD seen at 15 healthcare institutions between Jan. 2009-Dec. 2021
- Engaged caregivers to assess lived experiences managing blood pressure
- Expanded the PCORnet® Common Data Model for pediatric and rare kidney disease research

Dissemination

- Published findings on the comparative effectiveness of two treatments for high blood pressure in pediatric CKD in [JAMA Pediatrics](#)
- Published findings on using real-world electronic health record data to develop age-, sex-, and height-specific blood pressure percentiles for children in [eBioMedicine](#)

OTHER RARE DISEASES RESEARCH POWERED BY PCORnet®

- [Biologic Abatement and Capturing Kids' Outcomes and Flare Frequency in Juvenile Spondyloarthritis \(BACK-OFF JSpA\)](#)
- [Neuroendocrine Tumors-Patient Reported Outcomes \(NET-PRO\)](#)
- [A Clinical Trial Readiness Study of Patient Reported Outcome Measures in Thrombotic Thrombocytopenic Purpura \(THINK-TTP\)](#)
- [Predictive Analytics via Networked Distributed Algorithms for Multi-System Diseases \(PANDA-MSD\)](#)

POWER YOUR RARE DISEASE RESEARCH WITH PCORnet®

PCORnet resources are available to academic and industry researchers, patient advocacy groups, and other organizations from all funding sources and affiliations. PCORI also offers [funding](#) for broad pragmatic studies using PCORnet.

Contact the **PCORnet® Front Door** team to start a conversation about how you can use PCORnet to support your research.



PCORnet® is a national resource, funded by PCORI, that enables insights from high-quality health data, patient partnership, and research expertise to deliver fast, trustworthy answers that advance health outcomes. The network supports both observational and interventional research.