



RESEARCH SPOTLIGHT ON
Rare Diseases

Rare diseases are often chronically debilitating and sometimes life-threatening conditions that each affect fewer than 200,000 people nationwide. They may seem like a small corner of health care, but together, they affect roughly **10 percent of the US population**. That's about the same as the number diagnosed with diabetes. Yet rare diseases are frequently not studied enough. Patients with rare diseases often face years of unnecessary testing or treatments because their conditions are unrecognized by clinicians.

Rare diseases affect about **30 MILLION** Americans

Source: Centers for Disease Control and Prevention

**Research Addressing
Questions That Matter**

PCORI funds **comparative clinical effectiveness research (CER)** to determine which healthcare options work best for which patients, based on their needs and preferences. CER produces evidence that helps people make better-informed healthcare choices.



PATIENT

Having lupus has damaged my kidneys. To prevent my kidney disease from advancing to kidney failure I need to choose between several medications. How can I best know which of the available drugs will work best for me?



CAREGIVER

My daughter has cerebral palsy and attends two-hour physical therapy sessions weekly to help her to develop her motor skills. As she's gotten older however, I've seen less progress. Would attending longer sessions, or attending two-hour sessions more than once a week help her to continue her progress or speed it up?

STUDY SPOTLIGHT

Comparing Pediatric Crohn's Disease Treatments

Pediatric Crohn's disease affects about 38,000 children in the United States. The disease causes abdominal pain and bloody diarrhea, which can lead to frequent hospital visits, surgery, and low quality of life. This study is comparing the experience of patients who receive a biologic drug called anti-TNF with those who get a combination of anti-TNF and an anti-inflammatory drug called methotrexate. It will assess the ability of each approach to reduce or eliminate symptoms and the occurrence of drug-related side effects. It is led by Michael Kappelman of University of North Carolina at Chapel Hill.

STUDY SPOTLIGHT

Comparing Treatments for Adults with Sickle Cell Disease

Around 100,000 individuals in the United States have sickle cell disease (SCD), a blood cell disorder that affects the body's ability to distribute oxygen. It leads to episodes of severe pain and sometimes even death. People with SCD often face negative attitudes and long delays when seeking treatment for pain crises in emergency departments. This study is comparing the timeliness and quality of care experienced by patients who visit specially designed infusion centers versus those who receive treatment in emergency departments. It is led by Sophie Lanzkron of Johns Hopkins University in Baltimore, Maryland.

The Promise of PCORnet in Improving Rare Disease Care

PCORI is investing in people and infrastructure to support high-quality patient-centered research for years to come. **PCORnet, the National Patient-Centered Clinical Research Network**, will enable us to learn from the data and experiences of more than 145 million Americans. It is building capacity for research and other studies by supporting patient-driven networks and helping them connect with researchers who are studying similar topics, including rare diseases.

Rare diseases are well represented in PCORnet's 33 partner networks. Several of the 20 Patient-Powered Research Networks, which are operated and governed by groups of patients and their partners, focus on one or more such illnesses. And each of the 13 health system-based Clinical Data Research Networks within PCORnet has a rare disease as one of its conditions of primary focus.



Bringing Patients Together to Drive Research

Patient-Powered Research Networks (PPRNs) that focus on rare diseases:

- CENA
- DuchenneConnect
- NephCure
- Rare Epilepsy Network
- PARTNERS
- Phelan-McDermid Syndrome Data Network
- PI-CONNECT
- Vasculitis PPRN

As of August 2017, PCORI has awarded

\$62 MILLION TO FUND 25

comparative clinical effectiveness research studies related to rare diseases

BY THE NUMBERS

These studies focus on 18 diseases and conditions:

- Acute Myeloid Leukemia
- Cerebral Palsy
- Chiari Type I Malformation and Syringomyelia
- Disorders of Sex Development
- Duarte Galactosemia
- Eosinophilic Esophagitis
- Hydrocephalus
- Idiopathic Subglottic Stenosis
- Kawasaki Disease
- Lupus Nephritis
- Non-CF bronchiectasis
- Pediatric Crohn's Disease
- Pediatric Transverse Myelitis
- Polyarticular Juvenile Idiopathic Arthritis
- Sickle Cell Disease
- Spinal Cord Injury and Spina Bifida
- Systemic Scleroderma
- Urea Cycle Disorders

These conditions affect the following bodily systems:



CARDIOVASCULAR/
CIRCULATORY



DIGESTIVE/
EXCRETORY



ENDOCRINE



IMMUNE



MUSCULAR/
SKELETAL



NERVOUS



REPRODUCTIVE



RESPIRATORY