



RESEARCH SPOTLIGHT ON

# Rare Diseases

Rare diseases are often chronically debilitating and sometimes life threatening. They each affect fewer than 200,000 people nationwide. Rare diseases may seem like a small corner of health care, but together, they affect nearly 10 percent of the US population. That's about equal to the number of Americans diagnosed with diabetes. Yet most rare diseases are not studied enough. As a result, patients with rare diseases often face years of testing or ineffective treatments.

Rare diseases affect about **25 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

I have lupus, which causes my immune system to attack healthy organs. My lupus led me to develop kidney disease. To prevent kidney failure, I need to choose between several medications. But several factors affect how each drug will work with my body. How can I know which of the available treatments will work best for me?



### CAREGIVER

My daughter has cerebral palsy and attends physical therapy once a week for two hours to help develop her motor skills. As she's gotten older, I've seen less progress. Which would better help her to continue to progress or even speed up her development: attending longer weekly sessions or attending more sessions per week?

## STUDY SPOTLIGHTS

### Comparing Pediatric Crohn's Disease Treatments

Crohn's disease affects about 38,000 children in the United States. This study is comparing the experience of patients who receive a treatment called anti-TNF with those who get a combination of anti-TNF and an anti-inflammatory drug called methotrexate. It assesses how well each treatment reduces or eliminates disease symptoms and how many drug-related side effects occur. Details about this project appear at [www.pcori.org/Kappelman026](http://www.pcori.org/Kappelman026).

### Comparing Treatments for Adults with Sickle Cell Disease

Around 100,000 Americans have sickle cell disease, a blood cell disorder that can cause episodes of severe pain and sometimes even lead to death. People with sickle cell disease 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 go to emergency departments versus those who visit infusion centers. Details about this study appear at [www.pcori.org/Lanzkron027](http://www.pcori.org/Lanzkron027).

## PCORI's Commitment to Rare Disease Research

PCORI is committed to changing the culture of research by bringing together a variety of stakeholders to help drive useful, relevant research about what works best for individual patients. Part of this commitment specifically focuses on funding research that addresses outstanding questions about the prevention, diagnosis, and treatment of rare diseases. Learn more about our rare disease resources at [www.pcori.org/RD-resources](http://www.pcori.org/RD-resources).

PCORI's Advisory Panel on Rare Disease seeks to bring voices from across health care and the rare disease community into our work on rare conditions. The panel provides a wide range of stakeholder guidance on everything from refining and prioritizing research questions for potential funding to engaging patients as partners in research. More details are available at [www.pcori.org/RDAP](http://www.pcori.org/RDAP).

As of June 2020, PCORI has awarded

**\$92 MILLION TO FUND 30**

comparative clinical effectiveness research studies related to rare diseases.

### BY THE NUMBER OF PROJECTS

These studies focus on 19 diseases and conditions. Learn more at [www.pcori.org/RD-projects](http://www.pcori.org/RD-projects).

- 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
- Myasthenia gravis
- 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 body systems:



CARDIOVASCULAR/  
CIRCULATORY



DIGESTIVE/  
EXCRETORY



ENDOCRINE



IMMUNE



MUSCULAR/  
SKELETAL



NERVOUS



REPRODUCTIVE



RESPIRATORY