Rare disease

Unlocking solutions for rare metabolic diseases, including inborn errors of metabolism

Clinical trials for rare metabolic disorders and inborn errors of metabolism (IEM) pose unique challenges that demand the expertise of an experienced partner with comprehensive capabilities.

The PPD™ clinical research business of Thermo Fisher Scientific has the capabilities to overcome these challenges and drive success in clinical trials for both inborn and acquired rare metabolic disorders. With our **patient-centric approach, extensive experience, and innovative solutions**, we offer a comprehensive suite of services tailored to rare metabolic disorders.

Rare metabolic research experience

With **over 180 rare metabolic studies**, including **more than 165 IEM studies** over the past five years, we have a proven track record in tackling complex metabolic disorders. As a leader in IEM research, we leverage our deep understanding of these disorders to develop novel solutions to enhance patient outcomes. Our expertise extends to a wide range of inherited and acquired conditions.

Our expertise at a glance:

- Lysosomal storage disorders (LSD) i.e., Fabry disease, Gaucher disease
- Glycogen storage disease type I/II
- Ureal cycle disorders
- Organic acidurias
- Fatty acids oxidation disorders
- Mitochondrial disorders
- Electrolytes and mineral metabolism
- Hypoparathyroidism
- Ornithine transcarbamylase deficiency
- Phenylketonurias
- Wilson's Disease
- Congenital Adrenal Hyperplasia (CAH)

180+ rare metabolic studies

Rare disease and pediatric expertise expertise

Genome sequencing technology

Genetic therapy experience

Laboratory capabilities for biomarkers in IEM

Registries and observational studies

Consulting for IEM clinical trials and development

Patient services that drive trial success and positive health outcomes

Our team recognizes the unique challenges patients with rare metabolic disorders face in participating in clinical trials. To address this, we offer **robust patient outreach** services that enhance participant engagement and retention, providing **consistent and proactive support** that reduces the burden on patients and caregivers. Our services ensure patients can easily access and participate in metabolic disorder clinical trials, resulting in **higher retention rates and better health outcomes**.

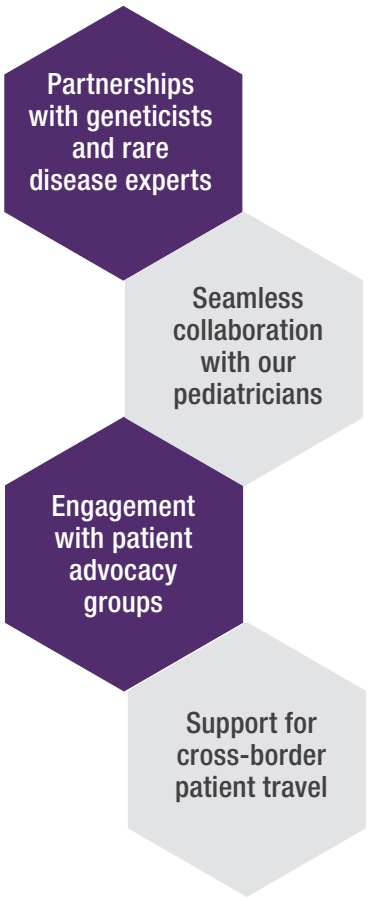
- Full-service booking and transportation coordination
- Flexible reimbursement options in 150+ currencies
- Home health care partnerships
- Flexible protocols for digital and decentralized trials

Novel solutions for site and patient access

When you partner with us, you gain access to agile solutions for site and patient access, addressing the unique challenges posed by rare metabolic disorders. Due to the fact that the majority of these disorders are expressed in early childhood, our Rare Disease and Pediatric Center of Excellence — staffed by experienced pediatricians — has conducted over 240 pediatric studies, providing seamless collaboration and specialized expertise.

We partner with geneticists and rare disease experts to understand unique medical and logistical issues related to each diagnosis and trial. We also engage with key opinion leaders in rare metabolic space, as well as community and patient advocacy groups, incorporating the patient perspective into protocols, enhancing community awareness, and connecting you with decision makers.

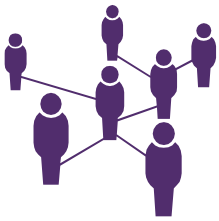
Our teams can remove participation barriers by maximizing study decentralization, utilizing remote data collection and procedures, and supporting cross-border patient travel. Moreover, we are well-versed in cutting-edge gene therapies, ensuring you have access to the latest advancements in the field.



Harnessing feasibility tools for success

We leverage powerful feasibility tools that tap into our extensive database, enabling us to design efficient and effective studies tailored to your needs. With our in-depth knowledge and comprehensive capabilities, we guide you towards success in the development of treatments for rare metabolic disorders, using data such as:

Patient locations



Number and types of providers

Analysis of the competitive landscape

Glycogen storage disease study

Our ability to navigate complex trials is exemplified by our **Phase I-II Glycogen Storage Disease Type IA trial**. This study required a meticulous schedule of assessments, including **overnight stays, home health care visits, and continuous glucose monitoring (CGM)** for patients involved in the study. Our strategic approach enhanced CGM compliance among participants, ensured trial enrollment remained on track throughout its lifecycle, and ultimately achieved 100% study retention. By leveraging our expertise and patient-focused services, we were able to effectively optimize trial outcomes to deliver high-quality data for advancing the development of life-changing treatments.

Optimizing trial outcomes

- ✓ Enhanced CGM compliance
- ✓ Maintained 100% study retention
- ✓ Kept enrollment on track
- ✓ Robust data collection

Partner with us

The complexities of rare metabolic trial logistics shouldn't hinder your progress. We offer many innovative methodological approaches in conducting clinical trials for rare metabolic disorders/IEM and strong partnerships with end-users and stakeholders involved in their development, application, and evaluation to ensure access to the many new therapies for rare diseases in the near future. Secure a partnership with us, the industry leader in rare disease research, to unlock the full potential of your clinical trials.

Learn more at **ppd.com/raredisease**