Clinical trials

# Rare diseases

ppd

# Specialized solutions for rare disease clinical research

**PPD's rare disease and pediatric center of excellence (COE)** focuses on delivering transformation science and diverse, cross-functional services to the life science leaders we work with every day. This means driving innovative and executable trial designs, operational strategies, and focusing intently on reducing the burdens of clinical research participation, all in an effort to meet the needs of patients and families with rare diseases and deliver on the promise of transformational therapies.

**Our dedicated, experienced teams** deliver cross-functional expertise providing innovative, patient-centric solutions and training to our study teams and sites to skillfully address the most complex challenges of small, globally dispersed populations — across all therapeutic areas.

# Global experience and infrastructure

PPD has extensive global experience working with drug developers in every region to design and execute successful rare disease clinical trials across all therapeutic areas.

- Neuroscience
- Hematology
- Oncology
- Musculoskeletal
- Gastrointestinal
- Dermatology
- Immunology
- Ophthalmology
- Cardiovascular
- Endocrinology/Metabolic
- Respiratory

**This proven experience means that PPD has strong, long-lasting relationships** with the investigators and sites delivering both the care and research engagement to patients and families dealing with rare diseases. These connections support improved efficiency at study start-up and increased study recruitment and overall retention.

## In the past five years, our expert development leaders and teams have partnered with clients on:

**556**
Studies
in rare disease indications

Executed in
**101+**
Countries

Leveraging study sites
**19,500**
Times

**Continuous investment in our capabilities to allows us to enhance the site, investigator, patient and caregiver experience.**

# Bringing hope to rare disease patients and caregivers

The physical and psychological toll of a rare disease on families is immeasurable. Each day a product is not on the market only increases this toll. Because every minute counts, an integrated approach to overcoming persistent development obstacles is required. These obstacles can include a lack of consensus on endpoints, inexperienced research sites, a shortage of regulatory precedents, geographically dispersed patient populations, and access to enough patients to participate in a trial. Competitive landscapes can also be a hurdle — for example, in ALS and DMD development.

At PPD, part of Thermo Fisher Scientific, we leverage our medical, operational, and regulatory expertise — combined with our real-world experience — to design and operationalize studies with a customized approach for each indication. Our teams plan for natural history studies that can run in parallel with therapeutic studies or be used as feeder studies into your clinical research program.

### These studies:

- Generate the real-world evidence required by the FDA and other regulatory bodies to better understand target patient populations.
- Can support guide the design of rare disease clinical trials by helping to describe disease frequency, features and evolution.
- Explore the use of novel trials designs in rare disease, including basket and platform trials, along with adaptive mechanisms when suitable.

# Allieviating burdens with decentralized approaches

Our aim is to decrease clinical trial burden and increase flexibility for rare disease patients and their loved ones. For both care and research, bringing patients into physical clinics can be challenging, and it can be even more difficult for children and their families or for those with limited mobility.

With the widespread availability of virtual and decentralized trial options, we can design hybrid approaches, including a mixture of both in-clinic and at-home visits. These options are supported by bespoke digital platforms and novel trial design solutions.

Decentralized options enable us to optimize the patient and caregiver experience with minimal inconvenience, leading to positive impacts on recruitment and retention.

Innovative, flexible protocols — combined with digital and decentralized trial technologies — offer new ways to reach, recruit and retain the small, globally dispersed populations of rare disease patients. With trials being reimagined to bring clinical research to the patient, we are able to meet them where they are and simplify participation with tools such as mobile sites, wearables, telehealth and e-consent.

## Patient identification and access

Our comprehensive approach includes:

- Patient pathway mapping to understand the patient diagnostic and care journey.
- Multichannel patient identification to find the patients meeting study criteria.
- Patient registry and LTFU study design and execution.

## Strategic relationships

We actively build strategic relationships with patient advocacy groups, academic institutions, health care providers and pharma industry groups.

This comprehensive engagement supports recruitment, alleviates clinical trial burdens, and assures compassionate insight and consideration of the patient journey.

## Patient concierge

PPD's patient concierge services are built to reduce trials burdens for patients and families. We offer various bespoke services along with options including a dedicated PPD resource who manages logistics and guides patients every step of the way.

# Expertise in gene therapy

As with many areas of truly advanced therapeutics, gene therapy is bringing new hope for patients with rare diseases. However, the many unknowns in gene therapy understanding, related trial execution, and critical sample/IP handling needs further complicate the existing challenges of drug development. Our guidance begins at the pre-clinical stage to maximize resources throughout the development process.

## Addressing unique challenges

Our extensive experience in gene therapy studies in rare disease patient populations — and across various therapeutic areas — supports our ability to expertly plan and operationalize studies with unique challenges.

**For example:**

- Training and monitoring sites for complex medicinal product administration.
- Oversight of logistics needed for just-in-time gene therapy delivery and appropriate site preparation/handling of valuable investigational product.
- Support-focused recruitment, including the inclusion of traveling patients, providing for cross-border participation, and reducing the trial burdens to improve the overall patient experience, seeking to increase the ability to participate.

## Operational expertise

PPD's deep medical and operational gene therapy research expertise extends to running trials across multiple phases (First in Human, Phase I-IV) as well as providing expertise for IP requiring various routes of administration. This enables us to partner with you to, if desired, support your protocol design to optimize dose and minimize potential toxicity. In addition to trial work, the complex manufacturing required can be supported by a number of gene therapy experts in our broader Thermo Fisher Scientific family.

## PPD™ Laboratory services

Our lab services combine high-quality scientific expertise with industry-leading technologies supported by a commitment to exceptional quality and a proven history of success.

We offer a range of high-value, advanced testing services that support gene therapy development, including bioanalytical, biomarkers, central lab, GMP, potency assays and vaccine sciences.

## Regulatory approval

Most gene therapy programs will require the support of natural history studies and/or registry information, as well as long-term follow-up of all dosed patients for up to 15 years. A thoughtful evidence strategy from early development is critical for both regulatory success and efficiency across your program.

## Payor strategy

The high cost of gene therapy is something that most payors are still not prepared for. We have experienced teams who can work with you to build your overall value case and identify innovative payment methods and evidence gathering solutions to ensure the patients you seek to treat have ongoing access to your approved product.

# Unmatched pediatric knowledge and experience

Children comprise over half of the rare disease population, and their unique needs and challenges must be considered when planning and executing clinical trials. PPD has broad experience working with diverse pediatric populations. In the past five years, we've executed more than 240 pediatric studies and 97 in rare pediatric indications. We provide expert guidance on:

- Consent, assent, product formulation and safety.
- Protocol development, feasibility, study design and study-related issues based on knowledge of current standard of care and regulatory trends in the pediatric space.
- Product development pathways and pediatric investigational plans.
- Appropriate regulatory structures and processes and access to investigators with relevant pediatric expertise.

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**We are committed to helping you effectively and efficiently navigate the complex development journey with a shared goal in mind: to bring independence and freedom to patients with rare diseases.**

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Learn more at **ppd.com/therapeutic-expertise/rare-diseases**

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