Not So Uncommon: Rare Brain Disorders

The Rare Brain Disorders Clinic and Laboratory brings together patients, families, physicians, and scientists to forge a common path to accurate diagnosis and better treatment for complex, underdiagnosed, or uncommon disorders.

In one sense, rare diseases are not rare. Any single disease can be uncommon, often resulting in years of frustrating misdiagnoses for patients and their families. But together, 7,000 types of rare diseases—mostly genetic—afflict 25 million people in the United States.
Dr. Pascual is one of only a few physician-scientists in the country who focus on diagnosing and treating rare or complex disorders of the central nervous system. He received his MD degree from the Universidad de Granada, Spain. He did his internship in Pediatrics at Washington University School of Medicine-St. Louis Children’s Hospital and a residency in Pediatric Neurology at the Neurological Institute of New York-Columbia University Medical Center. He received his PhD degree in Molecular Physiology and Biophysics from Baylor College of Medicine. His postdoctoral research was done at the Center for Molecular Recognition and, later, at the Colleen Giblin Research Laboratories for Pediatric Neurology at Columbia University. He is board certified in Neurology with Special Competence in Child Neurology.

The Rare Brain Disorders Clinic

The Rare Brain Disorders Clinic treats about 400 pediatric and adult patients from around the world each year. For many, this visit marks the first time they receive an accurate diagnosis of their condition. The clinic unites and supports neurologists, geneticists, pediatricians, and other professionals as they diagnose and treat patients. Almost all patients return to the clinic periodically throughout their lives, regardless of where they live or their socioeconomic status.

The Rare Brain Disorders Clinic and Laboratory provides a unique mix of comprehensive clinical care, research, and education, focused exclusively on rare neurological disorders.

Clinical Care

The Rare Brain Disorders Clinic:

- Treating more than 400 pediatric and adult patients from around the world each year
- Providing care in the first time patients receive an accurate diagnosis
- Offering comprehensive care to patients regardless of where they live or their socioeconomic status

Research

UT Southwestern aims to foster translational research that makes a real difference in clinical care by moving discoveries “from bench to bedside.” The Rare Brain Disorders Laboratory embodies this call and plays a key role in translational research efforts, such as the Collaborative Education and Test Translation (CETT) program funded by the National Institutes of Health Office of Rare Brain Diseases Research, designed to create and implement new genetic tests.

- Conducting research in rare brain disorders
- Developing and implementing new diagnostic and treatment options

Education

The Rare Brain Disorders Clinic and Laboratory holds a commitment to nurture promising academic physicians. About 50 medical students and graduate students, as well as residents and postdoctoral researchers, are involved in the Rare Brain Disorders Clinic and Laboratory annually, ensuring that the clinic’s greatest accomplishments lie ahead. The laboratory is approved for a research residency track in pediatric neurology by the American Board of Psychiatry and Neurology.

Outreach and Awareness

The laboratory’s educational mission doesn’t end with future physicians—the patients, their families, and the public add to the collaborative effort. It takes many hands working together to carry improved care from lab to bedside.

- Through invited lectures and public speaking, our faculty members increase awareness of rare brain disorders, available medical services, new scientific discoveries, and opportunities to participate in clinical trials.
- In addition, the Rare Brain Disorders Clinic and Laboratory partners with The Child Brain Foundation every year to hold The Child Brain Conference on topics of brain disease and biology of interest to the lay public.