UT Southwestern
Medical Center

Department of Pediatrics

Child Neurology

2023 Annual Report

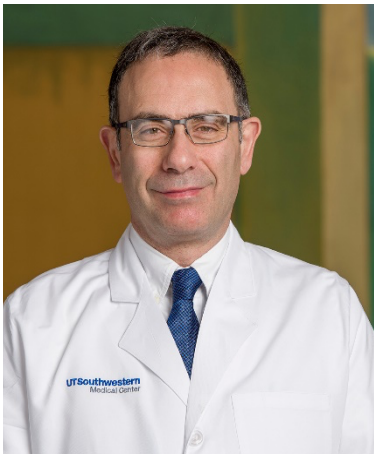
Pediatric Neurology – 2023 Annual Report

The **Division of Child Neurology** represents an academic practice with commitment to patient care, teaching, and research.

Formed in the 1980s as part of the Department of Neurology, the Division maintained a successful academic and training program through the 1990s. The Division has been part of the Department of Pediatrics since 2006, and Dr. Minassian became the Division Chief in 2016.

Under Dr. Minassian's direction, the Division provides comprehensive diagnosis and management for pediatric patients at Children's Medical Center Dallas, from newborn to late adolescence, who have disorders of the brain, spinal cord, nerves, or muscles. Faculty members specialize in providing neurological care, consultations, and second opinions for children in response to almost every neurological disorder.

Division faculty conduct a variety of neurologic bench/basic research and clinical studies, including clinical trials. Current studies are focused on the following main areas:



Berge Minassian, M.D.
Professor, Division Chief

- Gene therapy
- Movement disorders
- Metabolic disorders of the nervous system
- Neuromuscular disease clinical trials
- Anti-epileptic drug trials
- Clinical trials in pediatric stroke
- Clinical studies of the autism spectrum
- Neuroimaging studies in pediatric movement disorders
- Functional neurological disorders
- Clinical trials in pediatric migraine
- Concussion recovery utilizing cerebrovascular functioning

Numbers at a Glance

Faculty: 33 New Faculty: 4 Promotions: 3 *U.S. News & World Report*: #15	Clinical Fellows: 16 Clinical Sites: 7	Inpatient Encounters: 8,639* Outpatient Encounters: 26,892*
Research Funding Total* Grants: $5,881,138 Contracts: N/A Total # Proposals Awarded: 13 Public Federal (# K Awards): N/A Public State of Texas: N/A Other 13 Revenue from Active Clinical Trials as of 12/31/2023: $5,284,801	Active Clinical Trials: 42*	Conferences/Lectures: 71 Publications: 107* Citations: 6,019* Book Chapters: 2

*Sources: Orbit report of RHi032 Organizational Research Dashboard, UTSW Office of Research and Grants Support, Faculty Affairs, Education Alumni Affairs, Clinical Business Operations-invoice creation period for 2023, CVs

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Honors/Awards

Best Pediatric Specialists, *D Magazine*

- Michael Dowling
- Kimberly Goodspeed
- Rana Said

Texas Super Doctors, *Texas Monthly*

- Susan Iannaccone
- Jennifer Thomas (Rising Star)

Steven Gray

- Thomas Dierks Award – United MSD Foundation

Kimberly Goodspeed

- KL2 Clinical Scholar – UT Southwestern Medical Center

Rana Said

- Person of the Year – Epilepsy Foundation of Texas
- Training Director Award – Child Neurology Society and the Professors and Educators of Child Neurology

Education and Training

Rana Said, M.D., is the Director of Education for the Division and the Program Director for the Child Neurology residency. She oversees all training programs in the Division, including undergraduate medical education, the Neurodevelopmental Disabilities residency, and all fellowship programs. She also provides direct mentorship and support to the program directors and site directors for each of these programs. In her capacity as Program Director of the Child Neurology Residency Program (core program) for the past 18.5 years, she has seen the program grow considerably. Along with that growth, Lauren Sanchez, M.D., has joined as the Associate Program Director of the Child Neurology Residency Program. The program offers an Accreditation Council for Graduate Medical Education (ACGME)-accredited three-year training program, including a combined five-year "categorical" program with two years of ACGME-accredited pediatrics training and three years of ACGME-accredited child neurology training based at Children's Medical Center Dallas/UT Southwestern Medical Center.

We also offer a "categorical" child neurology basic neuroscience research pathway (five-year program, with a one-year preliminary UT Southwestern Pediatrics residency, one year of American Board of Psychiatry and Neurology (ABPN)-approved research, followed by three years of a Child Neurology residency), and for appropriate residency candidates, the Physician Scientist Training Program in Pediatrics or the Department of Neurology's R25 Research Program are available. The program was approved by the ACGME in December 2020 to increase the complement to five Child Neurology residents each year, making it one of the largest child neurology training programs in the nation. The program has continued to

receive "commended" status from the ACGME for the past review cycles. Graduates are eligible for the American Board of Pediatrics and the ABPN, with special qualifications in child neurology.

Patricia Evans, M.D., Ph.D., is the Director of the Neurodevelopmental Disabilities (NDD) clinical and residency programs. The NDD residency is ACGME-accredited, highly competitive, and one of only eight programs nationally. The NDD training program includes a combined six-year "categorical" program with two years of ACGME-accredited pediatrics training and four years of ACGME-accredited neurodevelopmental disabilities training based at Children's Medical Center Dallas/ UT Southwestern Medical Center. Graduates are eligible for three boards – specifically, the American Board of Pediatrics; the ABPN, with special qualifications in child neurology; and the ABPN in neurodevelopmental disabilities. NDD trainees at UTSW have protected time, especially in the sixth year of training, to pursue research projects with both didactic and bench training for translational research.

Alison Dolce, M.D., is the Director of the Pediatric Clinical Neurophysiology Fellowship Program. Graduates are eligible for the ABPN in clinical neurophysiology.

Deepa Sirsi, M.D., is the Director of the Pediatric Epilepsy Fellowship Program. This ACGME-accredited program accepts two fellows per year. Graduates are board-eligible in epilepsy.

Jennifer Thomas, M.D., is the Director of the Fetal and Neonatal Neurology Fellowship Program. This is a Texas Medical Board (TMB)-approved program that accepts one fellow per year who has completed prior training in either child neurology or neonatal-perinatal medicine.

Mathew Stokes, M.D., is the Director of the Pediatric Headache Fellowship Program. This program is TMB-approved and a United Council for Neurologic Subspecialties-accredited program. This program accepts one fellow per year, and graduates are eligible for United Council for Neurologic Subspecialties board certification in headache medicine. There is a sports neurology track for interested candidates.

Kimberly Goodspeed, M.D., is the Associate Program Director of the Neurodevelopmental Disabilities Residency Program and serves on the Resident Recruitment Committee, contributing to curriculum development, mentoring, and selecting future training classes. She works with the NDD trainees on a two-year NDD board preparation curriculum, quality-improvement projects in the NDD continuity clinic, and as a mentor on trainee-initiated scholarly projects. Dr. Goodspeed also manages a shared educational resource of clinical research methodologies for faculty and trainees to foster collaborations and training on protocol development and clinical research design, as well as scientific and grant writing resources.

Michael Dowling, M.D., Ph.D., is the Associate Program Director for the Vascular Neurology Fellowship Program. This is an ACGME-accredited program that accepts one pediatric-track fellow per year. Graduates are eligible for the ABPN in vascular neurology.

The Neuromuscular Medicine Fellowship Program is an ACGME-accredited program that accepts one pediatric-track fellow per year. Pediatric Neuromuscular Medicine faculty members serve in a Site Director capacity for the pediatric subspecialty fellow. Graduates are eligible for the ABPN in neuromuscular medicine.

Fellowship Program in Development:

- Pediatric Movement Disorders: This is a TMB-approved program that will accept one pediatric subspecialty fellow per year. Jeff L. Waugh, M.D., Ph.D., will be the Site Director for the pediatric subspecialty fellow.

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Research Activities

Division faculty conduct a variety of neurologic bench/basic research and clinical studies, including clinical trials. Current studies are focused on the following areas:

- Gene therapy
- Movement disorders
- Metabolic disorders of the nervous system
- Neuromuscular disease clinical trials
- Anti-epileptic drug trials
- Clinical trials in pediatric stroke
- Clinical studies in the autism spectrum
- Concussion recovery utilizing cerebrovascular functioning

Faculty research projects:

- **Dr. Minassian** is an international authority on Lafora disease, a glycogen storage disorder of the brain with intractable and fatal epilepsy. His laboratory has uncovered large amounts of knowledge on the metabolism of glycogen in the brain. In addition, he is building the premier national Gene Therapy Center with his colleague Steven J. Gray, Ph.D., and others on the faculty. The goal is a complete transformation of pediatric neurology whereby children with single gene defect brain diseases, as appropriate, would receive treatment in the form of replacing the missing gene.
- **Dallas Armstrong, M.D.,** is involved in several projects related to Rett syndrome. She is the Medical Director of the Rett Syndrome and Related Disorders Clinic, which received a Center of Excellence (CoE) designation in 2021 from the International Rett Syndrome Foundation (IRSF), and participates in an ongoing patient registry study, for which the clinic received a grant in 2021. Dr. Armstrong works with Dr. Hoang Nguyen, in the Division of Pediatric Cardiology, as well as neurology trainees on projects related to cardiac electrophysiology in girls and women with Rett syndrome and with medical students on the caregiver burden in boys with severe neonatal encephalopathy related to MECP2. She is working with several colleagues at Rett CoEs across the country on an original research project investigating anti-seizure medication use and efficacy in the Natural History Database, as well as on a revision of the clinical practice guidelines in Rett syndrome. Dr. Armstrong serves on the IRSF medical advisory board and clinical trial committee. She also works with Dr. Sirsi and resident trainees on projects related to TBC1D24 epileptic encephalopathy. Lastly, Dr. Armstrong is working with a neurology trainee as well as colleagues across institutions on a seizure semiology smartphone application.
- **Xin Chen, M.D.,** is the Primary Investigator (PI) of three research projects (DDX3X, GNAO1, and CMT4A) initially funded by Taysha Gene Therapies and two new research projects (PDHA1 and LNPK) funded by primary foundations. He is also the Co-Investigator of multiple research projects funded by private foundations, industry, and the National Institutes of Health (NIH) under Dr. Gray. Dr. Chen's other research areas are aspartylglucosaminuria (AGU), CLN7 Batten disease, SPG50 disease, SLC52A2 deficiency disease, and SRD5A3 deficiency disease. In 2021, an investigational new drug (IND) application to initiate a Phase I intrathecal gene transfer trial for AAV9/MFSD8 was approved by the U.S. Food and Drug Administration (FDA), and the trial is ongoing at Children's Medical Center Dallas, in collaboration with UTSW (clinicaltrials.gov NCT04737460). In 2022, the IND application to initiate a Phase I intrathecal gene transfer trial for AAV9/AP4M1 was approved by the FDA, and the trial is enrolling SPG50 patients at Children's Medical Center Dallas, in collaboration with UTSW (clinicaltrials.gov NCT05518188). He is currently working to facilitate the initiation of gene therapy clinical trials for AGU gene therapy. Dr. Chen is also helping to develop novel adeno-associated virus (AAV) capsids with superior efficacy, higher specificity, and less toxicity; supporting several projects in other laboratories and institutions; and mentoring laboratory personnel.
- **Dr. Sanchez** is involved in clinical research in the areas of rare neurogenetic disorders, neurometabolic disorders, and neuro-oncology.
- **Dr. Dowling** is involved in clinical research in the areas of stroke in children, Sturge-Weber syndrome, and neurologic complications of sickle cell disease.

- **Wilmot Bonnet, M.D.,** is involved in clinical research in the areas of ischemic and hemorrhagic stroke in children, acute interventions for ischemic stroke, and recovery in children with acquired brain injury.
- **Dr. Evans** is an active participant in translational research, both for autism spectrum disorders as well as fragile X syndrome. She participates in a range of studies, including the genetics of autism, spectrum disorders, and the mechanisms of fragile X syndrome cognitive deficits.
- **Dr. Goodspeed** is involved in the clinical research of rare neurogenetic and neurodevelopmental disorders and directs the Rare Genetic Neurodevelopmental Disabilities Specialty Clinic to serve these patients. She has expertise in a number of genetic conditions, including Pitt-Hopkins syndrome, SLC6A1-related neurodevelopmental disorder, SLC13A5 deficiency disorder, AGU, and Phelan-McDermid syndrome. Dr. Goodspeed is also heavily involved in the development of the UTSW Gene Therapy Program alongside Dr. Minassian and other faculty colleagues. She is the PI for several gene therapy disease programs, including clinical trial readiness studies for AGU, SLC6A1-related neurodevelopmental disorder, SLC13A5 deficiency disorder, and SRD5A3-CDG. Dr. Goodspeed has served as a consultant in the clinical trial design for a first-in-human gene transfer therapy study for GM2-related lysosomal storage diseases and consults for gene therapy companies, including Astellas Gene Therapies, Jaguar Gene Therapy, and Taysha Gene Therapies. She was awarded the UTSW Community Based Research Award to conduct a study of the efficacy of a group-based educational program at the Dallas Zoo on the social-emotional functioning of young children with autism spectrum disorder and a Planning Grant Award to develop a protocol for using noninvasive heart rate monitoring to predict challenging behaviors in the classroom among children with autism spectrum disorder. Dr. Goodpseed also was named a Dedman Family Scholar in Clinical Care for her efforts to establish a deep phenotyping registry for children with neurodevelopmental disorders, which is characterizing the cognitive and behavioral profiles of children with Pitt-Hopkins syndrome (SLC6A1-NDD) and Duchenne muscular dystrophy. She earned her graduate certificate in clinical sciences through the UTSW Center for Translational Medicine and completed her Master of Science in clinical sciences in spring 2022. Dr. Goodpseed is currently a KL2 Scholar and has submitted a K23 application to the NIH's National Institute of Neurological Disorders and Stroke (NINDS). Since September 2022, she has had four peer-reviewed manuscripts published and four poster presentations at national meetings.
- **Dr. Gray** is the PI on several grant-funded projects and sponsored research agreements. These include an NIH R01 grant to develop novel AAV capsids for central nervous system (CNS) gene transfer, an NIH R01 subcontract to develop a gene therapy approach for Niemann-Pick Type C disease, as well as foundation grants from the Cure SPG50 Foundation and the Sappani Foundation. He also has multiple sponsored research agreements from Taysha Gene Therapies, as well as sponsored research agreements from Roche, 3M, Millipore Sigma, Form Bio, Phoenix Nest, the Monoamine Oxidase Deficiency (MAO) Foundation, the Charcot-Marie-Tooth Association, the DDX3X Foundation (Co-PI), and Hannah's Hope Fund. Dr. Gray has two well-scored pending NIH grants that should initiate in 2023: U01 to initiate a trial for aspartylglucosaminuria and U19 for preclinical gene editing. His funded research projects include the development of gene therapy treatments for giant axonal neuropathy (GAN), CLN7 Batten disease, multiple sulfatase deficiency, Rett syndrome, Angelman syndrome, SLC6A1 deficiency, SPG50 disease, SLC52A2 disease, GNAO1, DDX3X, Charcot-Marie Tooth disease types 4A and 4J, fragile X syndrome, SRD5A3, non-ketonic hyperglycinemia, MAO deficiency, muccopolysaccharidosis type 3C, and Leigh syndrome. Dr. Gray created a core facility for research and clinical AAV manufacture and also facilitated the initiation of gene therapy clinical trials for multiple rare neurological diseases, including CLN7 Batten disease and spastic paraplegia type 50 at UTSW and the following disorders elsewhere: GAN, CLN1 Batten disease, CLN5 Batten disease, Rett syndrome, and GM2 gangliosidosis (Tay-Sachs and Sandhoff diseases). He's had seven publications since the start of 2022, including two recent ones of note in the *Journal of Clinical Investigation* (Chen X et al, 2022; Chen X et al, 2023).
- **Sharmistha Mitra, Ph.D.,** is the Co-Investigator of multiple basic science research projects funded by private foundations, industry sources, and the NIH, under Dr. Minassian. This includes the Wellstone Foundation Award and the multi-PI Chan Zuckerberg Initiative, a funded grant to investigate the molecular mechanism of Lafora disease. She is also PI of UTSW's internally (Small Animal Center Research Award) and externally foundation-supported (American Society for Investigative Pathology) projects to decipher the role of multiple E3 ubiquitin ligases in glycogen metabolism. Dr. Mitra focuses much of her research in the area of protein modification by ubiquitin and its impact on neuromuscular diseases. Her primary research interests are protein-protein interactions in glycogen metabolism and related neuromuscular diseases, the role of E3-ubiquitin ligases in polyglucosan body diseases, and targeted molecular therapy for certain pediatric neuromuscular diseases.

Dr. Mitra supervises and mentors senior research scientists and multiple technicians in Dr. Minassian's laboratory and has been a direct supervisor for summer students and STARS awardees.

- **Susan Iannaccone, M.D.,** is Director of Clinical Research for the Division of ChildNeurology. Since 2015, she has been Associate Director of the UTSW Wellstone Muscular Dystrophy Cooperative Research Center, funded by the NIH and led by Eric Olson, Ph.D., and Co-Chair of the Administrative Core for the Wellstone Center. She serves on research grant review committees as well as several planning committees and panels for patient groups and the NIH. She is Co-Investigator for the UT Southwestern Euronext grant from NINDS (2018-23) and has trained/mentored more than 20 pediatric neuromuscular fellows and medical students. Dr. Iannaccone is the site PI or site Co-PI for more than a dozen industry-sponsored clinical trials for pediatric neuromuscular and neurodegenerative diseases, including gene therapy and the first human trials; is currently working with the UTSW Gene Therapy Program as a mentor for early-stage investigators as part of the team performing investigational new drug (IND) applications; and is the PI or Co-PI for human trials, including:
    - A Phase I/II Open-Label Intrathecal Administration of MELPIDA to Determine the Safety and Efficacy for Patients with Spastic Paraplegia Type 50 (SPG50) Caused by a Mutation in the AP4M1 Gene (active)
    - A Phase III, Multinational, Randomized, Double-Blind, Placebo-Controlled Systemic Gene Transfer Therapy Study to Evaluate the Safety and Efficacy of SRP-9001 in Non-Ambulatory and Ambulatory Subjects with Duchenne Muscular Dystrophy (ENVISION) (active)
    - A Phase I Intrathecal Lumbar Administration of AAV9/CLN7 for Treatment of CLN7 Disease (active)
    - A Phase I/II Open Label Study to Evaluate the Safety, Tolerability and Efficacy of a Single Dose of AAV9/FIG4 Delivered via Lumbar Intrathecal Administration in Charcot-Marie-Tooth-4J (pending)
- **Saima Kayani, M.D.,** has been actively involved in the development of clinical aspects of gene therapy trials at UT Southwestern and serves as PI and Co-PI for NIH and industry-sponsored trials. These trials are ongoing at UT Southwestern/Children's Medical Center Dallas, with other sponsored programs upcoming.
  Dr. Kayani has carried out various research responsibilities, including handling and writing informed consent documents, writing clinical protocols for the institutional review board (IRB), and overseeing and upholding medical ethics and regulatory issues. She has acted in the capacity of Lead Investigator and Medical Adviser and developed protocols, case report forms, and data capture platforms. Dr. Kayani has also been actively involved in the development of the clinical section of INDs for the first-in-human Phase I intrathecal gene therapy trial for Batten disease type 7 (CLN7). These efforts have led to the successful FDA approval of an open IND for a Phase I trial for intrathecal gene therapy in CLN7. As the Co-PI for the CLN7 Phase I first-in-human clinical trial, she is leading the clinical team in the successful implementation of the trial and overseeing all regulatory compliance. As a key opinion leader, Dr. Kayani has provided extensive clinical insight and strategic planning input, along with proficient clinical operations, to biotech companies such as Taysha Gene Therapies in partnership with the UTSW Gene Therapy Program. This involves regular standing weekly/monthly meetings discussing disease phenotypes, clinical impressions, experimental design, disease-specific biomarkers, and data analysis. These efforts have led to the start of a prospective natural history trial for the rare brain disease SURF-1-related Leigh syndrome (starting in early 2022).
- **Dr. Stokes** is involved in clinical research studies focused on biomarkers and phenotypes of concussion. He is the Children's Medical Center Dallas site Principal Investigator for ConTex, which is a multicenter, prospective longitudinal registry for concussion established in 2015. Additionally, he is a Co-PI on a multicenter study, CARE4KIDS, looking at objective biomarkers of concussive head injury.
- **Dr. Said** is involved in multiple industry-sponsored clinical research trials. She is the PI for an investigational product (ganaxalone vs. placebo) for the treatment of status epilepticus. In addition, she is currently the Co-PI working with Dr. Sanchez, Daniel Bowers, M.D., (Neuro-oncology), Elizabeth Maher, M.D., Ph.D., (Internal Medicine), Rebekah Clarke, M.D., (Neuroradiology), and Robert Bachoo, M.D., Ph.D., (Neurology), in an IRB-approved study to assess a strict classic ketogenic diet for recurrent or progressive refractory brain tumors in children. This is a prospective study to assess not only the response (anti-tumor effect on the ketogenic diet) but also the ability to achieve and maintain ketosis with a rigidly calculated and supervised classic ketogenic diet. The study will also be assessing biomarkers of efficacy by screening for the expression of enzymes that are critical for ketone metabolism in brain tumor samples. Magnetic response spectroscopy will also be utilized to evaluate the presence of certain metabolites in tumors that may serve as important biomarkers of tumor behavior and response to therapy. Dr. Said is also the PI of a novel treatment for refractory status epilepticus and involved with several medical education studies in conjunction with other members of the Education Committee of the American

Academy of Neurology. She is also involved in clinical research on new-onset refractory status epilepticus, and infantile spasms.

- **Sarah Sinnett, Ph.D.,** is the PI of several projects funded by industry sources. She has also been funded by the International Rett Syndrome Foundation. Her lab focuses on developing regulated gene therapies for the treatment of rare neurodevelopmental disorders characterized by intellectual disability. Dr. Sinnett is Co-Inventor on several provisional patent applications and sole Inventor on another provisional patent application; two of these inventions have been licensed to industry. Her recent first-author publication in *Brain* has an attention score ranking it within the top 5% of nearly 23 million publications. The intellectual property described in this publication forms a broader research platform for her lab and for the industry. The ultimate goal of this intellectual property is to streamline and accelerate gene research and development for multiple disease applications. Dr. Sinnett has been PI of the Rett syndrome gene therapy project at UTSW since 2020.
- **Dr. Sirsi** is involved in a range of clinical research studies concerning electroencephalograms (EEG) as a predictor of neuroimaging and outcome in pediatric ECMO and pediatric nonaccidental head trauma; EEG biomarkers in single gene disorders such as GLUT1 deficiency syndrome; and SLC6A1 syndrome. She is the site PI for industry-funded anti-seizure medication trials. Dr. Sirsi collaborates with other clinicians and basic scientists in research involving treatment of the genetic causes of epilepsy and neurodevelopmental disorders. She participates in multicenter research studies on infantile spasms conducted by the Pediatric Epilepsy Research Consortium.
- **Dr. Dolce** is involved in a range of clinical research studies, including those in the areas of neonatal neurology, pediatric epilepsy, and Lafora disease. She is currently the site Co-PI for the Lafora Natural History Study.
- **Peter Tsai, M.D., Ph.D.,** directs the Cerebellar Neurodevelopmental Disorders Clinic and is performing both pre-clinical and translational research for autism and neurodevelopmental disorders, including fragile X syndrome and tuberous sclerosis. He is the PI on numerous federally funded preclinical and translational research projects. These include NIH R01 and Department of Defense (DOD) grants to delineate circuit mechanisms underlying autism-relevant behaviors in tuberous sclerosis, an NIH RO1 grant to explore the circuits contributing to abnormal behaviors in fragile X syndrome, and a DOD-funded grant examining brain mechanisms and the potential benefits of noninvasive brain stimulation for children with autism. He is actively working to develop therapeutic targets and examine the therapeutic efficacy of brain stimulation for children with neurodevelopmental disorders.
- **Cynthia Wang, M.D.,** is involved in clinical research characterizing demyelinating and other immune-mediated brain conditions such as multiple sclerosis, neuromyelitis optica spectrum disorder, and autoimmune encephalitis in the pediatric population. She is the PI on two upcoming clinical trial studies on pediatric multiple sclerosis and neuromyelitis optica spectrum disorder. Dr. Wang has published original research and case reports/case series on unique patient presentations in the field of rare neuroimmunological disorders.
- **Jeff Waugh, M.D., Ph.D.,** is involved in neuroimaging projects that utilize MRI to study the brain in patients with the movement disorder dystonia. In addition to these disease-related research projects, he develops novel methods for improving the accuracy of quantitative diffusion imaging and for identifying compartments within the human striatum in vivo that have previously been visualized only via post-mortem histology. Dr. Waugh collaborates with researchers and clinicians at the Harvard-MIT Martinos Center for Biomedical Imaging, Boston Children's Hospital, the Lübeck University in Germany, and the University of Santo Tomas and the University of the Philippines, both in Manila, Philippines. He has current grant funding from the NINDS, the Children's Clinical Research Advisory Committee, and the Brain and Behavior Research Foundation.
- **Felix Nitschke, Ph.D.,** is a PI investigating the metabolic pathways involved in several neurodegenerative diseases, such as Lafora disease and adult polyglucosan body disease. His work currently focuses on glycogen metabolism and the treatment of associated neurological diseases with novel gene therapy approaches. Dr. Nitschke received funding from the Orphan Disease Center at the University of Pennsylvania. In addition, he serves as Lead Investigator on a five-year grant from the NIH's NINDS and as Co-PI on a collaborative grant from the Chan-Zuckerberg Initiative.

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Clinical Activities

The Pediatric Neurology multidisciplinary teams include board-certified pediatric neurologists, pediatric nurse practitioners, physician assistants, genetic counselors, licensed pediatric nutritionists, occupational therapists, physical therapists, and speech therapists. Our teams offer accurate diagnosis and comprehensive management plans, as well as access to state-of-the-art clinical trials.

The Division's faculty treat:

- Brain tumors
- Cerebrovascular disease and stroke
- Intractable epilepsy, rare genetic epilepsy syndromes, epilepsy due to brain malformations, epilepsy surgery evaluations, ketogenic diet, and dietary treatments for epilepsy
- Developmental delay
- Neurobehavioral disorders and autism
- Neuroimmunologic disorders, such as multiple sclerosis
- Neurologic complications of sickle cell disease
- Neurometabolic, neurogenetic, and complex/rare diseases
- Neuromuscular disorders, such as muscular dystrophy and myasthenia gravis
- Movement disorders, such as dystonia, chorea, myoclonus, tremors, Parkinsonism, and tics
- Urgent epilepsy for new onset afebrile seizures
- Neonates with neurological complications
- Pediatric headache disorders, including urgent headache treatment and intracranial hypertension
- Pediatric mild traumatic brain injury, including concussion diagnosis and Return-to-Play treatment plans

Research

The Comprehensive Epilepsy Center continued to participate in clinical research, with the faculty involved in IRB-approved clinical research studies as primary investigators (industry sponsored, nonfunded) and co-investigators in NIH-funded trials.

- There were two industry clinical trials that started recruitment last year and one that closed out after successful enrollment.
- There were multiple industry-sponsored trials that were in the startup stage the previous year, with anticipated initiation in 2023.
- There are 10 nonfunded research projects that were initiated in 2022 and are in various stages at UTSW among the epilepsy team (faculty, fellows, and residents) that are producing ongoing scholarship.
- UTSW pediatric epilepsy members became an associate site of The Epilepsy Study Consortium (TESC), and key faculty attended the TESC introductory meeting at the 2022 American Epilepsy Society (AES) Annual Meeting. A roundtable discussion about rare epilepsies was held the first week of February 2023.
- UTSW Pediatrics was also represented at the Pediatric Epilepsy Research Consortium, with faculty members and leaders of special interest groups studying epilepsy surgery, developmental epileptic encephalopathies, sunflower syndrome, epilepsy with myoclonic atonic seizures, neuromodulation, and psychogenic nonepileptic seizures.
- Faculty members were invited to speak at meetings and regional symposiums, and to give Grand Rounds at other institutions.
- Epilepsy faculty have published 15 manuscripts, including editorials, in peer-reviewed journals.
- One faculty member served as a contributing editor to *Epilepsy Currents*, and other faculty served as ad hoc reviewers for child neurology and epilepsy-focused journals.

Education

- The ACGME Pediatric Epilepsy and Neurophysiology Fellowship Programs continue to attract talented applicants. A third track for fellowship training was added in 2021: the Advanced EEG and Epilepsy Research Fellowship.
- These competitive fellowships have been filling all positions and training highly competent pediatric epilepsy physicians, many of whom go on to university faculty positions at reputable institutions, including some fellows

who continue their careers at UTSW and increase the reach of this team to provide high-quality care to children with severe epilepsy.

- Pediatric epilepsy faculty are leaders and members of multiple national neurology and epilepsy education committees, including the American Board of Psychiatry and Neurology Epilepsy Exam Writing and Recertification Committee, the American Academy of Neurology (AAN) Pipeline Subcommittee, the AES Diversity Equity and Inclusion Committee, the AES Pediatric Content Committee, and the American Clinical Neurophysiology Society Maintenance of Certification Committee.