

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 children at Children’s Health, from newborn to late adolescence, who have disorders of the brain, spinal cord, nerve, or muscle. Faculty members specialize in providing neurological care, consultations, and second opinions for children afflicted by virtually any neurological disorder.

Division faculty conduct a variety of neurologic bench/basic research and clinical studies, including clinical trials. Current studies are focused in the following main 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 of the autism spectrum
- Neuroimaging studies in pediatric movement disorders
- Functional neurological disorders
- Clinical trials in pediatric migraine
- Concussion recovery utilizing cerebrovascular functioning



Berge Minassian, M.D.
Professor, Division Chief

It is a priority in the Division to maintain a competitive Child Neurology (CN) Training Program and Neurodevelopmental Disabilities (NDD) Training Program that will graduate Child Neurologists who have outstanding clinical training and are competitive clinicians, researchers, and educators. We offer several training pathways:

- “Categorical” Child Neurology Residency pathway: five-year program, with two years of preliminary UT Southwestern Pediatrics residency, followed by three years of Child Neurology residency.
- “Categorical” Child Neurology Basic Neuroscience Research pathway: five-year program, with one year preliminary UT Southwestern Pediatrics residency, one year of American Board of Psychiatry and Neurology (ABPN)-approved research, followed by three years of Child Neurology residency.
- “Reserved” Child Neurology pathway: three years of Child Neurology residency after the completion of two to three years of Accreditation Council for Graduate Medical Education (ACGME)-accredited Pediatrics residency training.
- “Categorical” Neurodevelopmental Disabilities Residency Program: six-year program, with two years preliminary UT Southwestern Pediatrics residency, followed by four years of Neurodevelopmental Disabilities residency.
- Fellowship training in many pediatric neurology subspecialties, including Advanced Fellowship in Electroencephalography and Epilepsy, Clinical Neurophysiology, Epilepsy, Fetal and Neonatal Neurology, Headache Medicine (with optional Sports Neurology track), Neuromuscular Medicine, and Vascular Neurology.
- For appropriate residency candidates, the Physician Scientist Training Program in Pediatrics (PSTP2) or the Neurology department’s R25 Research Program are available.

Each year, in collaboration with the Departments of Pediatrics and Neurology at UT Southwestern, Children's Health™, Texas Scottish Rite Hospital for Children, and the Muscular Dystrophy Association, the Division presents the Carrell-Krusen Neuromuscular Symposium for muscular dystrophy clinic directors, case managers, nurses, and members of interdisciplinary care teams. Dr. Susan Iannaccone directs the symposium, and most residents and fellows from the Division of Pediatric Neurology and from the Department of Neurology and Neurotherapeutics present at the symposium.

The Division of Pediatric Neurology hosts the New Frontiers in Pediatric Neurology Symposium in collaboration with UT Southwestern and Children's Health™, covering updates in the field of Pediatric Neurology. This is a one-day continuing medical education event for Pediatric Neurology physicians, advanced practice providers, residents, and fellows. Drs. Diana Castro and Deepa Sirsi direct the symposium.

The Pediatric NeuroConcussion Clinic, led by Medical Director Dr. Mathew Stokes, is the leading clinical enrolling site for the UTSW North Texas Concussion Registry (ConTex), which is a multicenter, prospective longitudinal registry for concussion, established in 2015.

The Clinical Pediatric NeuroConcussion Program has expanded greatly in capacity to diagnose, treat, and help youth with mild traumatic brain injury from sports and non-sports causes. The program recently moved operations to the newly opened Children's Health Pediatric Multispecialty Center at Cityville, which offers the latest in diagnostic and treatment capabilities for clinical care and includes specialized testing equipment, nursing, social work, school services, and rehabilitative services. Medical student and resident education in these facilities allows proper training in safe "Return to Play" and current Neurological published standards for concussion management.

The Pediatric Headache Program offers an unparalleled clinical and academic program in the Southwest. Dr. Mathew Stokes, our third Pediatric Headache Medicine fellow, recently joined the Pediatric Headache faculty and has been appointed as the new Program Director for this fellowship program. Dr. Eric Remster, the first graduate of the Pediatric Headache Fellowship, is involved with the ongoing clinical research studies in the Division that includes a Quality Initiative Care Pathway for Headache and a Pseudotumor database initiative. The clinical Pediatric Headache Program, which is also located in the Cityville Center, has capabilities of urgent headache care to help keep patients out of the Emergency Department and for procedures such as nerve blocks and botulinum therapy. Currently, the Headache Group is enrolling patients in a clinical trial to evaluate migraine therapy in children ages 5-11.

Faculty

The Division has 33 faculty members, five with a major commitment to research and all focused on clinical and teaching responsibilities. They represent a mix of young and mature individuals, each of whom brings special talents and experience that promise to contribute to further growth and development of the Division. Five faculty joined the Division in 2022.

Roha Khalid, M.D.

Clinical Associate Professor, Pediatrics and Neurology



M.D.

Aga Khan University Medical College, Karachi, Pakistan, 2008

Postdoctoral Training

Residency, Pediatrics

University of Texas Medical Branch, Galveston, TX, 2010 – 2012

Residency, Child Neurology

Duke University Hospital/Duke University Medical Center, Durham, NC, 2012 – 2015

Fellowship, Epilepsy

Children's Mercy Hospital, Kansas City, MO, 2019 – 2020

Interests: epilepsy and EEG, stroke, general neurology

Kazi Majeed, M.D.

Clinical Associate Professor, Pediatrics and Neurology



M.D.

Kind Edward Medical University, Lahore, Pakistan, 1990

Postdoctoral Training

Residency, Pediatrics

University of Florida/Health Science Center Jacksonville, Jacksonville, FL, 1992 – 1994

Residency, Child Neurology

Medical College of Georgia, Augusta, GA, 1994 – 1997

Fellowship, Clinical Neurophysiology & Cerebrovascular Disease

Southern Illinois University School of Medicine, Springfield, IL, 1997 – 1999

Interests: general neurology, epilepsy and EEG, headache

Sharmistha Mitra, Ph.D.

Instructor, Pediatrics and Neurology



B.S., Zoology, University of Calcutta, Kolkata, India, 2006

B.S., Biological Sciences, Molecular, Cellular and Developmental, University of New Hampshire, Durham, NH, 2008

Ph.D.

Virginia Tech, Blacksburg, VA, 2013

Postdoctoral Training

Postdoctoral Researcher, Pediatric Neurology

UT Southwestern, 2017 – 2020

Interests: Pediatric neuromuscular diseases involving protein ubiquitination system and identifying role of E3 ubiquitin ligases in neuromuscular disease pathogenesis

Nancy Philip, M.D.

Clinical Associate Professor, Pediatrics and Neurology



M.D.

University of Texas Health Science Center at San Antonio, San Antonio, TX, 2012

Postdoctoral Training

Residency, Pediatrics

University of Florida College of Medicine, Gainesville, FL, 2012 – 2014

Residency, Child Neurology

UT Southwestern, 2014 – 2017

Fellowship, Clinical Neurophysiology

SUNY Downstate Medical Center, Brooklyn, NY, 2021 – 2022

Interests: epilepsy, general neurology, EEG

Kendra Beneski, M.D.

Assistant Instructor, Pediatrics and Neurology



M.D.

University of Texas Health Science Center at Houston (McGovern Medical School), 2015

Postdoctoral Training

Combined Residency in Pediatrics & Pediatric Neurology

UT Southwestern / Children's Health - 2015-2020

Interests: epilepsy, genetics, neonatal neurology; personal interests include yoga, running, reading, spending time with her husband & dog

Honors / Awards

Best Pediatric Specialists, *D Magazine*

- Alison Dolce
- Michael Dowling
- Kimberly Goodspeed
- Andrea Lowden
- Rana Said
- Mathew Stokes
- Afsaneh Talai
- Jennifer Thomas

Texas Super Doctor, *Texas Monthly*

- Susan Iannaccone
- Jennifer Thomas (Rising Star)

Allison Dolce

- Promoted to Associate Professor

Michael Dowling

- Promoted to Professor

Karen Goodspeed

- Gold Humanism Society – UT Southwestern Medical Center

Deepa Sirsi

- Outstanding Physician-PA Team of the Year Nomination – Texas Academy of Physician Assistants

Sharmistha Mitra

- Advanced Imaging Research Center (AIRC) Scan Time Award - UT Southwestern Medical Center
- The Paul D. Wellstone Muscular Dystrophy Award – UT Southwestern Medical Center
- ASIP Summer Research Opportunity Program in Pathology – American Society for Investigation Pathology

Jeff Waugh

- Best Educator of the Year Golden Boot Award – Child Neurology Residents, UT Southwestern Medical Center

Invited Lectures

Rachel Bailey

- International MSD Scientific and Family Conference, Virtual, April 2022
 - *"Intrathecal delivery of scAAV9 gene replacement therapy for Multiple Sulfatase Deficiency"*
- SLC13A5 International Research Roundtable, Austria, September 2022
 - *"SLC13A5: Disease Model to Treatment"*
- American Society of Gene + Cell Therapy (ASGCT) Lunch & Learn, Virtual, February 2022
 - *"Gene Therapy 101: Landscape of Gene Therapy"*
- American Society of Gene + Cell Therapy (ASGCT) Lunch & Learn, Virtual, October 2022
 - *"Meet the Researchers"*
- TESS Research Foundation Clinical Research Conference, Dallas, TX, June 2022
 - *"Gene Therapy for SLC13A5 Deficiency"*
- Global CMT Research Convention, Cambridge, MA, September 2022
 - *"Gene Therapy for GAN and CMT4J"*
- Neuro2022: The CurePSP International Research Symposium, New York, NY, October 2022
 - *"Gene Therapy"*
- American Epilepsy Society Annual Meeting, Nashville, TN, December 2022
 - *"Investigators workshop: Developing Gene Therapies for Neurological Disorders"*

Xin Chen

- TWS (Tom Wahlig Stiftung) Symposium treat HSP meeting 2022. Tübingen, Germany, December 2022
 - *"Development of an Intrathecal AAV9/AP4M1 Gene Replacement Therapy for Hereditary Spastic Paraplegia 50"*
- 7th Meeting Translational Research Conference 2022 for the Management of NCLs. Lombard, IL, November 2022
 - *"Preclinical results in rodents strongly support clinical evaluation of scAAV9/MFSD8 as a potential gene therapy for CLN7 patients"*
- 6th Annual AP4-HSP Research Conference (2022). Boston, MA, October 2022
 - *"Development of an Intrathecal AAV9/AP4M1 Gene Therapy for Hereditary Spastic Paraplegia 50"*
- 2022 International conference and expo on Genetic Disorders and gene therapy: Gene therapy innovations for a better future. Virtual Hilaris Conferences, September 2022
 - *"Development of an Intrathecal AAV9/AP4M1 Gene Therapy for Hereditary Spastic Paraplegia 50"*

Alison Dolce

- EFTX Epilepsy Foundation Summer School Webinar, Virtual, July 2022
 - *"When Epilepsy Grows Up: A journey from the pediatric to the adult world"*
- New Frontiers in Pediatric Neurology Symposium, Dallas, TX, April 2022
 - *"Devices and New Modalities in Diagnosis and Treatment of Pediatric Epilepsy"*
- Lafora Disease Science Symposium, Virtual, September 2022
 - *"Antiseizure Medications in Lafora Disease"*
- Neonatal Brain Matters, Virtual, May 2022
 - *"Zebras in the NICU: Beyond the Acute Symptomatic Seizure"*
- NEO: The Conference for Neonatology, Las Vegas, NV, February 2023
 - *"The Development and Benefits of the Neuro-NICU"*
- Texas Pediatric Neuroscience Network Conference, San Antonio, TX, March 2023
 - *"Treatment Options for Acute Neonatal Seizures"*

Patricia Evans

- Annual Kenneth C. Haltalin Pediatrics for the Practitioner Lecture, Dallas, TX, May 2022
 - *“Neurological Evaluation and Management of Autism”*

Kimberly Goodspeed

- NDD Essentials, UTSW, September 2022
 - *“Epidemiology and Neurological Assessment of NDD Patients”*
- Medical Student Neurology Procedures Workshop –, UTSW, November 2022
 - *“Pediatric Neurological Exam”*
- Leading the Conversations on Health, UTSW – Old Parkland, October 2022
 - *Southwestern Medical Foundation Sprague Award, Panelist discussing innovations in gene therapy for neurological disease*
- UTSW Center for Translational Medicine, Translational Science Forum, October 2022
 - *Clinical Trial Readiness for SLC6A1-related Neurodevelopmental Disorder*
- SLC6A1 Connect Annual Symposium 2022, Nashville, TN, December 2022
 - *Clinical Trial Readiness for SLC6A1-Related Neurodevelopmental Disorder*
- Facilitate Advanced Therapies Week 2023, Miami, FL, January 2023
 - *Begin with the End in Mind: Clinical Trial Readiness in Rare Disease*
- American Society of Gene and Cell Therapy Lunch and Learn, virtual webinar, March 2023
 - *Roadblocks to Developing a Treatment – Panelist*
- Pediatric Emergency Medicine Fellowship Didactics, UTSW, April 2023
 - *“Autism Spectrum Disorder in the Emergency Department”*
- Child Neurology Society Webinar, April 2023 (virtual)
 - *Carving the Path Towards Clinical Trials in Rare Disease*
- UTSW Neuropsychology Didactics, UTSW, April 2023
 - *Poetry in Code – Genetic through analogy and metaphor*

Steven Gray

- American Society of Cell and Gene Therapy Insight Series - Immunological Responses to AAV Gene Therapy in Neurological Compartments: Mechanisms and Management. Virtual, November 2022.
 - *“Possible mechanisms (and mitigation strategies) for AAV-mediated DRG toxicity”*
- Federation of European Biochemical Societies 360 Lysosome Course. Izmir, Turkey. October 2022.
 - *“AAV-mediated gene therapy for lysosome disorders”*
- Neurology Grand Rounds. UT Southwestern Medical Center, Dallas, TX. September 2022.
 - *“AAV9/AP4M1 gene therapy for spastic paraplegia type 50” (Steven Gray) and “Journey to cure Michael” (Terry Pirovolakis)”*
- American College of Medical Genetics and Genomics Summer Gene Therapy Education Series: Advancement in gene therapy options for rare diseases. Virtual, July 2022.
 - *“Gene therapy targets: broadening the scope of therapy”*
- Garrod Symposium. Calgary, Canada. May 2022.
 - *“Approaches to gene therapy for inborn errors of metabolism” [Keynote Address]*
- Neurology Grand Rounds. UT Southwestern Medical Center, Dallas, TX. March 2022.
 - *“AAV9-mediated gene transfer as a platform approach for treating neurological disorders”*

Saima Kayani

- American Academy of Neurology Annual Meeting (Medical Student Seminar), Seattle, WA, April 2022
 - *“Gene Therapy in Neurologic Diseases”*
- Annual WORLD Symposium, San Diego, CA, February 2022
 - *“Preliminary safety data of a phase 1 first in human clinical trial support the use of high dose intrathecal AAV9/CLN7 for the treatment of patients with CLN7 disease”*
- Neurology Grand Rounds UT Southwestern, Dallas, TX, July 2022
 - *“Phase 1 first in human clinical trial support the use of high dose intrathecal AAV9/CLN7 for the treatment of patients with CLN7 disease”*

Andrea Lowden

- Federación Latinoamericana de psiquiatría de la Infancia, Adolescencia, La Romana, Dominican Republic, May 2022
 - *“Tics, new presentations and Covid 19”*
 - *“Advances in Epilepsy”*
 - *“Epilepsy and Psychiatric Disorders”*

Felix Nitschke

- Adult Polyglucosan Body Disease Scientific & Community Conference, Virtual, May 2022
 - *“Conquer from Within – Treating APBD by viral delivery of cross-correction-enabled amylase”*

Rana Said

- American Epilepsy Society – Summer Internship Webinars, Virtual, June 2022
 - *“Epilepsy: Treatment including Surgery”*
- Epilepsy Foundation of Texas, Summer Webinar, Virtual, June 2022
 - *“Vacationing with Epilepsy”*
- American Academy of Neurology Annual Meeting, Seattle, WA – April 2022
 - *The (Neuro)-Anatomy of an Effective and Impactful Program Director*
 - *Child and Adolescent Neurology for the Adult Neurologist*
 - *Course Director, Medical Student Symposium*

Sarah Sinnett

- Associazione Italian Rett Peschiera del Garda, Italy, Virtual, June 2022
 - *“Feedback-enabled gene therapy for Rett syndrome”*
- PURA Syndrome Virtual Conference, June 2022
 - *“Making an impossible gene therapy possible: Our scientific journey for Rett Syndrome (RTT)”*

Cynthia Wang

- Siegel Rare Neuroimmune Association, Rare Neuroimmune Disorders Symposium, Los Angeles, CA, October 2022
 - *“APERTURE: New Insights on ADEM”*

Jeff Waugh

- Neuroscience/Premedical Students, University of Dallas, Richardson, TX, April 2022
 - *“The Neuroanatomy of Compulsion”*

Conference Presentations

American Academy of Neurology Conference, Seattle, WA, April 2022

Shoaib A, **Machie M, Thomas J, Dolce A**

Poster Presentation, *“Use of Lacosamide in Neonatal Seizures “*

Choudhari P, Lowden A, Dolce A

Poster Presentation, *“Presence and Predictive Value of Midline Vertex Discharges on EEG in Children with Complex Febrile Seizures “*

American Medical Society for Sports Medicine’s (AMSSM) Annual Meeting, Austin, TX, April 2022

Worrall HM, Althoff CE, Miller SM, Jones JC., **Stokes MA**, Tow S, Cullum CM, Chung JS

Poster Presentation, *“Concussion trends in pediatric athletes presenting to outpatient clinics pre- and post-COVID-19 pandemic shutdown.”*

Poster Presentation, *“Anxiety history and injury characteristics among young athletes who did and did not receive a CT scan after a concussion.”*

Althoff CE, Worrall HM, Chung JS, **Stokes MA**, Tow S, Miller SM, Cullum CM, Jones JC

Poster Presentation, *“Does time of season impact concussion presentation and outcomes in youth football athletes?”*

Pediatric Academic Societies Meeting, Denver, CO, April 2022

Sirsi D

Abstract Presentation, *“Early Electroencephalography in predicting neurocognitive and imaging outcomes in pediatric patients with non-accidental traumatic brain injury.”*

Raman L, **Sirsi D, Said R**, Miles D

Oral Presentation, *“Early EEG as a Predictor of Neurologic and Imaging Outcomes in Young Children with Non-Accidental Traumatic Brain Injury”*

American Society of Gene + Cell Therapy (ASGCT) Annual Meeting, Washington, DC, May 2022

Bailey L, Schackmuth M, Garza I, Knight K, Holmes S, **Bailey RM**

Poster Presentation, *“Gene therapy treatment in young SLC13A5 deficient mice”*

Bailey RM, Armao D, Garza I, Yuhui H, Holmes S, Rozenberg A, Price A, Gray SJ

Oral Presentation, *“Vagus nerve delivery of AAV9 to treat autonomic nervous system dysfunction in Giant Axonal Neuropathy”*

Bailey RM, Garza I, Pearce K, Holmes S, Knight K

Oral Presentation, *“Vectorized delivery of tau reduction therapy as a treatment approach for tauopathies”*

Chen X

Oral Presentation, *“Development of an Intrathecal AAV9/AP4M1 Gene Therapy for Hereditary Spastic Paraplegia 50 (SPG50)”*

Eller MM, Garza I, Knight K, **Bailey RM**

Poster Presentation, *“Development of AAV gene therapy for ECHS1 Deficiency”*

Garza IT, Holmes S, Knight K, **Bailey RM**

Poster Presentation, *“Effect of age on intrathecal AAV9 transduction in mice.”*

Lyons C, Gray SJ, **Sinnott SE**

Poster Presentation, *“Non-invasive interventions may synergistically enhance MECP2 gene therapy”*

Prasad S, Flora G, Ramesh N, Tavakkoli F, Newman M, Gray SJ, **Sinnott S**, Sadhu C

Poster Presentation, *“Safety Assessment of High-Dose miniMECP2 AAV9 gene-replacement therapy (TSHA-102) for Rett Syndrome in Rats”*

Prasad S, Flora G, Tavakkoli F, Newman M, Gray SJ, **Sinnott S**, Sadhu C.

Poster Presentation, *“Safety and Biodistribution Assessment in Non-human Primates (NHPs) of a miniMECP2 AAV9 Vector for Gene-replacement Therapy of Rett Syndrome”*

Presa M, **Bailey RM**, Ray S, Bailey L, Tata S, Murphy T, Combs H, Gray SJ, Lutz C

Poster Presentation, *“Preclinical use of a new scAAV9/SUMF1 vector for the treatment of multiple sulfatase deficiency”*

Sadhu C, Tetens S, Flora G, Ramesh N, Tavakkoli M, Prasad S, Gray SJ, **Sinnott SE**

Published abstract, *“A human-ready regulated AAV9/miniMECP2 gene therapy improves survival and respiratory health after translationally relevant treatment ages in mice modeling Rett syndrome (RTT)”*

ASECND 2022 Rett Syndrome National Summit, Nashville, TN, April 2022

Armstrong D

Oral Presentation, *“What is that? – Seizures, Rett Episodes and Movement Disorders”*

Sinnott S

Oral Presentation and Poster Presentation, *“Non-invasive interventions may synergistically enhance MECP2 gene therapy”*

Other Conferences

Evans P

Essentials of Neurodevelopmental Disabilities (NDD) Virtual Meeting, September 2022, Dallas, TX
Creator and director, National Daylong Meeting Targeting Child Neurologists and Pediatricians

Presa M, **Bailey RM**, Ray S, Bailey L, Tata S, Murphy T, Combs H, Gray SJ, Lutz C

Annual WORLDSymposium, San Diego, CA, February 2022

Oral Presentation, *“Efficacy of a scAAV9/SUMF1 viral vector for the treatment of multiple sulfatase deficiency”*

Presley C, Goette W, Wilmoth K, Caze T, Miller S, Bell K, **Stokes M**, Didehbani N, Cullum CM

International Neuropsychological Society (INS) Annual Meeting, New Orleans, LA, February 2022

Paper Presentation, *“Predictors of recovery in adolescents with preexisting mental health problems following sports-related concussion.”*

Waugh J, Herbert L, Hassan A, Kim R, Wilkinson-Smith A

International Conference on Functional Neurological Disorders, Boston, MA, June 2022

Abstract Presentation, *“When neurologists diagnose functional neurological disorder, why don't they code for it?”*

Worrall HM, Miller SM, Chung JS, **Stokes MA**, Tow S, Cullum CM, Jones JC
Annual Pediatrics Research in Sports Medicine Society (PRISM) Meeting, Houston, TX, January 2022
Podium Presentation, *“Injury characteristics between pediatric athletes who did and did not receive a CT scan following concussion.”*

Education and Training

Dr. Rana Said is the Director of Education for the Division of Pediatric Neurology and the Program Director for the Child Neurology residency. She oversees all the training programs in the Division, including undergraduate medical education, the Child Neurology residency, the Neurodevelopmental Disabilities residency, and all fellowship programs. She also provides direct mentorship and support to each of 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, the program has grown considerably. Along with the growth of the program, Dr. Lauren Sanchez has joined as the Associate Program Director of the Child Neurology Residency Program. The program offers an 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 HealthSM Dallas/UT Southwestern Medical Center. We also offer a “Categorical” Child Neurology Basic Neuroscience Research pathway (five-year program, with one year preliminary UT Southwestern Pediatrics residency, one year of ABPN-approved research, followed by three years of Child Neurology residency), and for appropriate residency candidates, PSTP2 or the Neurology department’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 continues to receive “Commended” status from the ACGME for the past review cycles. Graduates are eligible for the American Board of Pediatrics and the American Board of Psychiatry and Neurology with special qualifications in Child Neurology.

Dr. Patricia Evans 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 HealthSM Dallas/UT Southwestern Medical Center. Graduates are eligible for three boards, specifically the American Board of Pediatrics, the American Board of Psychiatry and Neurology with special qualifications in Child Neurology, and the American Board of Psychiatry and Neurology 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.

Dr. Alison Dolce is the Director of the Pediatric Clinical Neurophysiology fellowship program. Graduates are eligible for the American Board of Psychiatry and Neurology in Clinical Neurophysiology.

Dr. Deepa Sirsi is the Director of the Pediatric Epilepsy fellowship program. This ACGME-accredited program accepts two fellows per year. Graduates are board-eligible in Epilepsy.

Dr. Jennifer Thomas 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.

Dr. Mathew Stokes 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 the United Council for Neurology Subspecialties Board in Headache Medicine. There is a Sports Neurology track for interested candidates.

Dr. Kimberly Goodspeed is the Associate Program Director of the Neurodevelopmental Disabilities residency program and serves on the Resident Recruitment Committee, contributing to curriculum development, mentoring, and selection of the

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 mentoring trainee-initiated scholarly projects. She also manages a shared educational resource of clinical research methodologies for faculty and trainees to foster collaborations and training on protocol development, clinical research design, as well as scientific and grant writing resources.

Dr. Michael Dowling is the Associate Program Director for the Vascular Neurology program. This is an ACGME-accredited program that accepts one Pediatric track fellow per year. Graduates are eligible for the American Board of Psychiatry and Neurology in Vascular Neurology.

The Neuromuscular Medicine program is an ACGME-accredited program that accepts one Pediatric track fellow per year. The Pediatric Neuromuscular Medicine faculty serve in a Site Director capacity for the Pediatric subspecialty fellow. Graduates are eligible for the American Board of Psychiatry and Neurology in Neuromuscular Medicine.

Fellowship Program in Development:

- Pediatric Movement Disorders: This is a TMB-approved program which will accept one Pediatric subspecialty fellow per year. Dr. Waugh will be the Site Director for the Pediatric subspecialty fellow.

Research Activities

Division faculty conduct a variety of neurologic bench/basic research and clinical studies, including clinical trials. Current studies are focused in 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 include:

- Berge 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, with his colleague Dr. Gray and others on the faculty, building the premiere national Gene Therapy Center. 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 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 on-going patient registry study, for which the clinic received a grant in 2021. She works with Dr. Hoang Nguyen in cardiology as well as neurology trainees on projects related to cardiac electrophysiology in girls and women with Rett syndrome and with medical students on caregiver burden in boys with severe neonatal encephalopathy related to MECP2. She works 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 dataset as well as on a revision of the clinical practice guidelines in Rett syndrome. She 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, she is working with a neurology trainee as well as colleagues across institutions on a seizure semiology smart phone application.



- Xin Chen is the Primary Investigator (PI) of three research projects (DDX3X, GNAO1, and CMT4A) initially funded by Taysha Gene Therapy and two new research projects (PDHA1 and LNPk) funded by private 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. Steven 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 Health in Dallas, TX, in collaboration with UTSW (clinicaltrials.gov NCT04737460). In 2022, IND application to initiate a Phase I intrathecal gene transfer trial for AAV9/AP4M1 was approved by the U.S. FDA and the trial is enrolling SPG50 patients at Children's Health in Dallas, TX, 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 in developing novel adeno-associated virus (AAV) capsids with superior efficacy, higher specificity, and less toxicity, supporting several projects in other laboratories and other institutions, and mentoring laboratory personnel.
- Lauren Sanchez is involved in clinical research in the areas of rare neurogenetic disorders, neurometabolic disorders, and Neuro-Oncology.
- Michael 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 is involved in clinical research in areas of ischemic and hemorrhagic stroke in children, acute interventions for ischemic stroke, and recovery in children with acquired brain injury.
- Patricia 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.
- Kimberly Goodspeed is involved in 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 several specific genetic conditions including Pitt-Hopkins Syndrome, SLC6A1-Related Neurodevelopmental Disorder, SLC13A5 Deficiency Disorder, AGU, and Phelan McDermid Syndrome. She 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 aspartylglucosaminuria, 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 also consults for several gene therapy companies, including Astellas Gene Therapy, Jaguar Gene Therapy, and Taysha Gene Therapy. 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 non-invasive heart rate monitoring to predict challenging behaviors in the classroom among children with autism spectrum disorder. Dr. Goodspeed was also awarded the Dedman Family Scholar in Clinical Care Award 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 program in Clinical Sciences through the UTSW Center for Translation Medicine and completed her Master of Science in Clinical Sciences in the spring of 2022. Dr. Goodspeed 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.
- Steven 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, 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, 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 noteworthy publications in the Journal of Clinical Investigation (Chen X et al, 2022; Chen X et al, 2023).

- Sharmistha Mitra is the Co-Investigator of multiple basic science research projects funded by private foundations, industry, and the NIH under Dr. Berge Minassian and Dr. Mitra. This includes Wellstone Foundation Award and multi-PI, Chan Zuckerberg Initiative--funded grant to investigate molecular mechanism of Lafora Disease where she is one of the PIs. She is also PI of UTSW's internally (Small Animal Center Research award) and externally foundation supported (American Society of Investigative Pathology) projects to decipher 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 is Director of Clinical Research for the Division of Pediatric Neurology. Since 2015, she has been Associate Director of the UTSW Wellstone Muscular Dystrophy Center, funded by the NIH and led by Dr. Eric Olson, 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 NeuroNEXT grant from NINDS (2018-2023) and has trained/mentored more than 20 pediatric neuromuscular fellows and medical students. Dr. Iannaccone is site PI or site Co-Principal Investigator (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, and is currently working with the UTSW Gene Therapy Program as mentor for Early-Stage Investigators, as part of the team performing investigational new drug (IND), applications and as PI or co-PI for human trials such as:
 - 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)
 - Phase 1 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 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 and others are upcoming sponsored programs. 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 Advisor 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 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 clinical trial and overseeing all regulatory compliance. As a key opinion leader, Dr. Kayani has provided extensive clinical insight and strategic planning, along with proficient clinical operations, to Biotech companies such as Taysha Gene Therapies in partnership with the gene therapy program. This involves regular standing weekly/monthly meetings discussing the 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).

- Mathew Stokes is involved in clinical research studies focused on biomarkers and phenotypes of concussion. He is the Children's Medical Center 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.
- Rana 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 Lauren Sanchez, Daniel Bowers (Neuro-oncology), Elizabeth Maher (Internal Medicine), Rebekah Clarke (Neuroradiology), and Robert Bachoo (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 to 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. MR 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 status epilepticus, refractory status epilepticus, new onset refractory status epilepticus, and infantile spasms.
- Sarah Sinnett is the PI of several projects funded by industry. 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. She is co-inventor on several provisional patent applications; sole inventor on a fourth provisional patent application; and two of these inventions have been licensed to industry. Dr. Sinnett's 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 industry. The ultimate goal of this intellectual property is to streamline and accelerate gene research and development for multiple disease applications. She has been PI of the Rett syndrome gene therapy project at UTSW since 2020.
- Deepa 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 non-accidental 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 genetic causes of epilepsy and neurodevelopmental disorder. She participates in multicenter research studies on infantile spasms conducted by the Pediatric Epilepsy Research Consortium.
- Alison 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-Investigator for the Lafora Natural History Study.
- Peter Tsai 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. Dr. Tsai 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 R01 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 non-invasive 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 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 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 University of Lübeck 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 National Institute for Neurological Diseases and Stroke, the Children's Clinical Research Advisory Committee, and the Brain and Behavior Research Foundation.

- Felix Nitschke is a PI investigating 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 NIH (National Institute of Neurological Disorders and Stroke) and as Co-PI on a collaborative grant from the Chan-Zuckerberg Initiative.

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.

Pediatric Neurology 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, tremor, 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

Center of Autism and Developmental Disabilities (CADD)

The Center of Autism and Developmental Disabilities (CADD) is a multi-disciplinary center that provides assessment and management for children, adolescents, and young adults with cognitive and/or developmental disabilities, with or without autism. CADD integrates neurology, psychology, both diagnostic and therapeutic, and psychiatry. Dr. Patricia Evans serves as one of three clinical co-directors for the center and directs one of only seven ACGME-accredited NDD training programs in the nation. Dr. Evans is head of the fragile X syndrome clinic at UTSW, which is based at CADD. Dr. Kimberly Goodspeed directs three rare neurodevelopmental disorders clinics, including those for Phelan McDermid Syndrome, Pitt Hopkins Syndrome, and SLC6A1 Related Disorders. Both Drs. Evans and Goodspeed participate in clinical research as well as clinical trials for these rare syndromes.

Comprehensive Epilepsy Center

The Comprehensive Epilepsy Center at Children's Medical Center Dallas strives to provide excellence in all areas of epilepsy care. Priorities are patient access, excellence in management of both straightforward and complex conditions, promotion of research and education, and community outreach to families and health care providers.

Access

The Epilepsy Center currently sees outpatients at three locations. The majority of patients are seen at Children’s Medical Center Dallas, with a smaller group seen at Texas Health Presbyterian Hospital and UTSW Frisco.

- Patient demand remained high in 2022 with 1,550 new patient referrals to the epilepsy center.
- With the expansion of services to Children’s Health Plano for neurology and epilepsy, three physicians were hired and started in fall 2022 to support the expanding need for neurology, epilepsy, EEG, and Epilepsy Monitoring Unit (EMU) services.
 - COVID, flu, and respiratory syncytial virus infections increased demands on acute care services and hospital beds, which limited the availability of these beds for scheduled admissions to the EMU in Plano in fall and winter 2022.
 - Epilepsy physicians who were hired to staff the EMU in Plano started in fall 2022, and onboarding and credentialing processes also affected scheduling EMU admissions.

Epilepsy Surgery Program

The Comprehensive Epilepsy Center is certified as a level 4 epilepsy center by the National Association of Epilepsy Centers, indicating provision of the highest level of complexity of epilepsy surgery services. Favorable epilepsy outcomes with surgery at Children’s Medical Center Dallas are comparable to other big centers and pediatric series published in peer-reviewed literature.

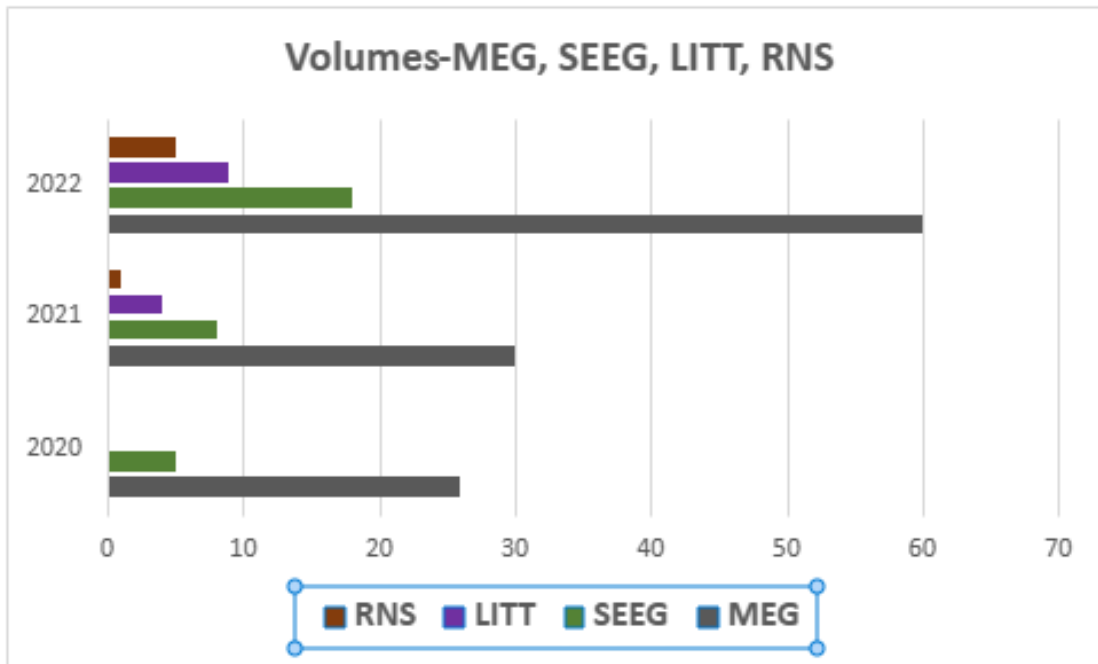
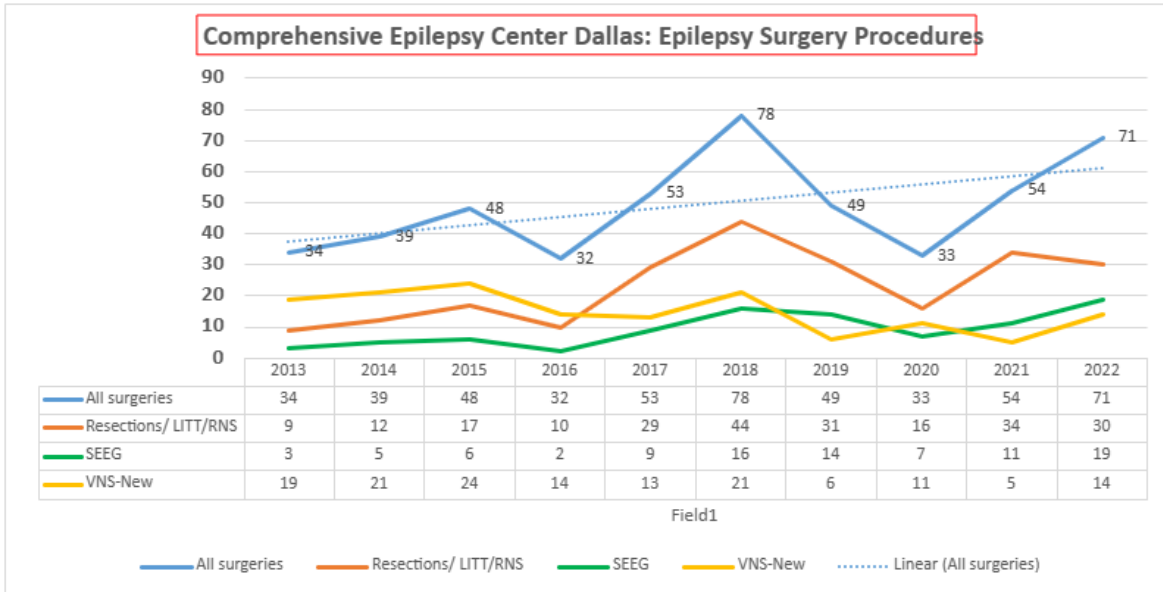
Procedures done in 2022 (reference numbers for 2021):

- Sixty magnetoencephalography procedures in children – MEG (30 in 2021)
- Nineteen stereo EEG procedures – SEEG(eight in 2021)
- Five laser interstitial thermoablation- LITT/Visualase (none in 2021)
- Five responsive neurostimulation –RNS/Neuropace (one in 2021)

The surgery program has shown steady growth over time and went through a dramatic expansion in late 2018 with the addition of robot-assisted stereotactic surgical procedures.

Threats

- There has been a significant increase in demand for physician time devoted to surgical procedures, and this continues to be an area of challenge for the program since the time needed for these surgical evaluations is not proportional to the reimbursements.
- Patients are individually discussed at multidisciplinary epilepsy surgery conferences, where the committee discusses recommendations. Continued support for epileptologists to participate in these conferences is essential and necessary.



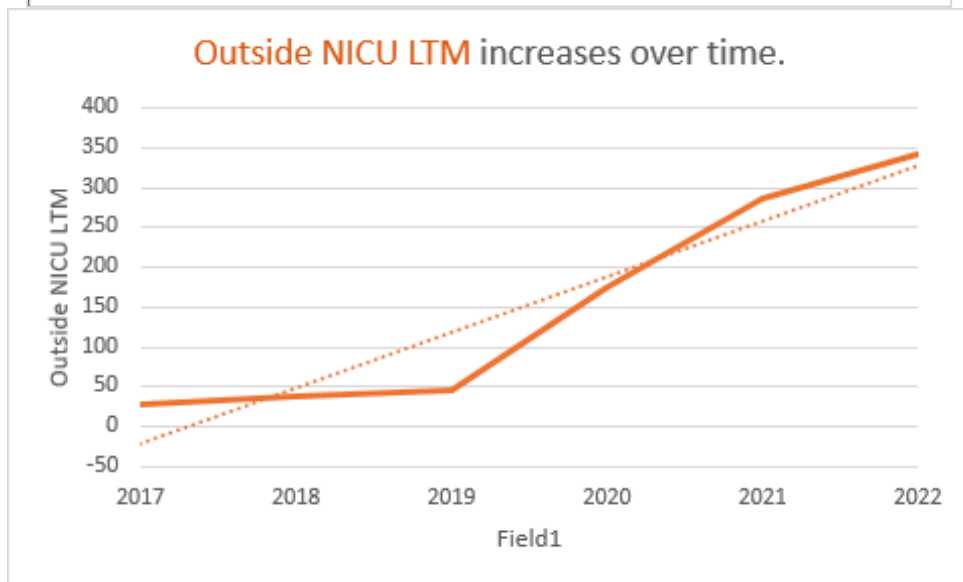
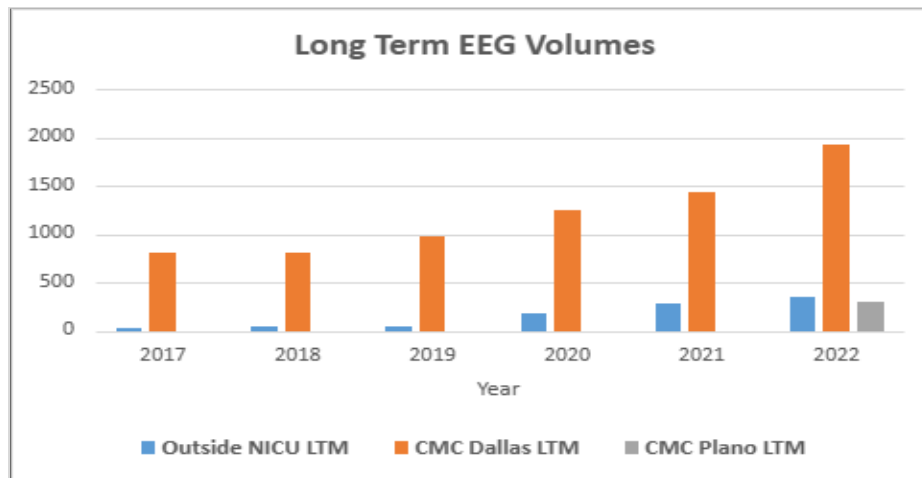
Neurophysiology Services

There has been a significant increase in inpatient long-term video EEG monitoring, especially in ICUs, as video EEG monitoring is increasingly utilized for monitoring children who are at high risk for seizures, which could further affect their outcome. The continued growth in ICU monitoring follows a national trend. The center also provides neonatal intensive care unit (NICU) EEG interpretation for

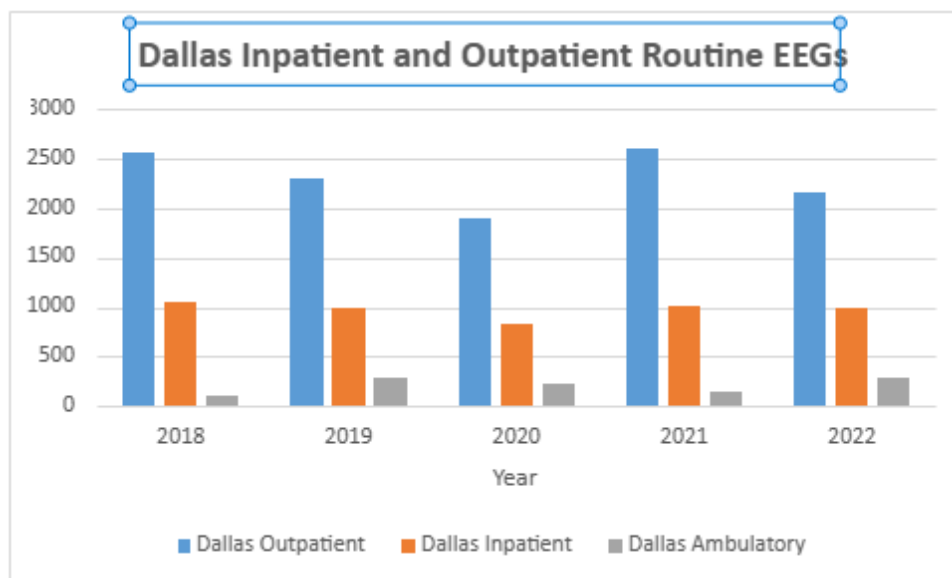
- Parkland Memorial Hospital
- Texas Health Presbyterian Dallas
- Clements University Hospital
- Texas Health Presbyterian Frisco

Threats

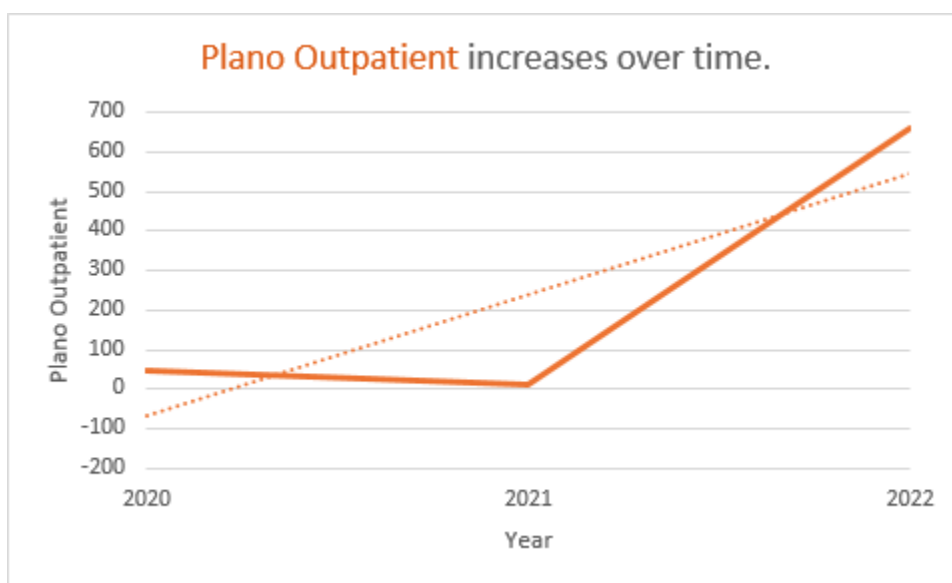
- Increasing volumes of ICU monitoring and increasing NICUs that epilepsy physicians are providing EEG interpretation services to on a daily basis are a challenge to the Epilepsy Center physicians due to their high acuity and since these studies are initiated at any time of day or night.



- EEGs performed by the Neurophysiology lab remains steady in the inpatient and outpatient settings.
- The graph below demonstrates volumes of routine inpatient and outpatient EEGs performed in Dallas.



- Plano Neurophysiology lab has seen an increase in volumes in 2022 since UTSW Neurology started service.
 - Three physicians were hired in 2022 and were onboarded between September and November 2022. There has been a significant increase in long-term EEGs, routine inpatient EEGs, and outpatient EEGs in 2022. Outpatient EEG volumes are below.



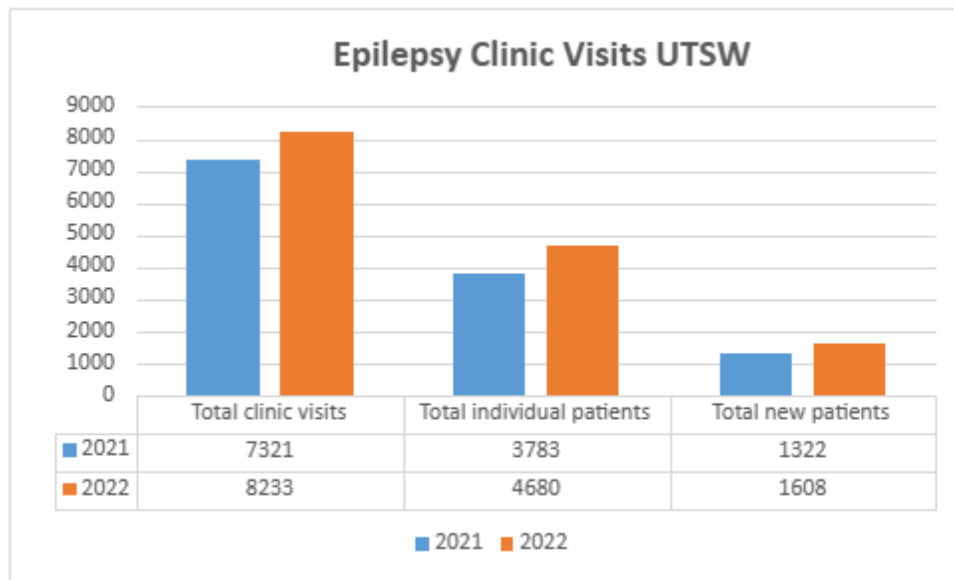
- (EMU) admissions for diagnosis, guidance with management and presurgical assessments. EMU volumes in Dallas have recovered after a decline in 2020 due to COVID.
- EMU volumes in Plano decreased in 2022 due to multiple reasons. Children’s Medical Center Plano experienced unprecedented inpatient acute care medical volumes during 2022. The facility’s obligation to provide care to patients entering from our Emergency Department limited elective admissions, including EMU admissions.
- On Aug. 1, 2022, the Plano EMU transitioned to the UTSW Neurology Group. Three new physicians were hired for Plano Neurology and Epilepsy services and onboarded between September to November 2022.

Opportunities for Improvement

Delays in credentialing and the need for completion have led to delays in physicians start dates and their ability to start providing clinical care. An increase in credentialing resources is needed along with an effort to improve the credentialing process.

Epilepsy Clinic

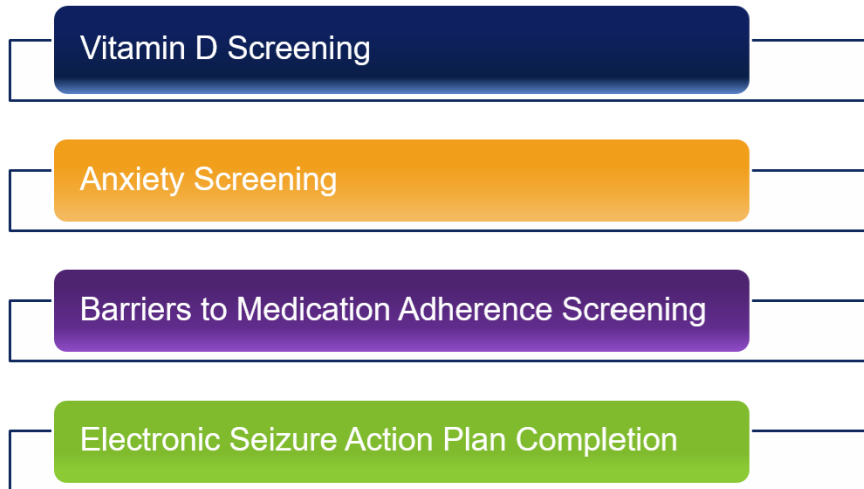
- UTSW Epilepsy physicians and advanced practice providers provide outpatient epilepsy care to over 4,500 patients through clinics located at Children’s Medical Center Dallas, UTSW Frisco, and Texas Health Presbyterian Dallas. The volumes of patients seen in 2022 were increased from 2021, as seen below.



National Recognition for Quality Care

- The Comprehensive Epilepsy Center was first certified in 2011 by the Joint Commission Disease Specific program for Distinction in the management of pediatric epilepsy and continues to be the only pediatric epilepsy center in the country with this certification. In 2022, this program received recertification for this distinction. The Joint Commission discontinued disease-specific certification in 2022. However, our epilepsy program continues to actively pursue quality improvement processes in the epilepsy clinic. Below are the QI measures undertaken in 2022. We track our performance on these measures every month and discuss this at our monthly epilepsy team meetings and look for barriers and areas for improvement on these measures.

Program Quality Improvement Initiatives 2022



children'shealth?

Proprietary and Confidential

1

- In 2020, the program joined a multicenter collaborative initiative, the Epilepsy Health Learning System (ELHS), and our center is one of the national leaders for quality improvement in outpatient pediatric epilepsy care. The quality measures supporting this distinguished status are:
 - Documenting and discussing seizure types, seizure frequency during each visit.
 - Assessing medication adherence at each patient visit.
 - Screening for depression and anxiety using standardized scales.
- ELHS registry: information specific to seizure frequency, medication adherence, and quality of life is collected from members of the ELHS on a monthly basis, and multicenter data is analyzed by the ELHS data collection center.
 - Data reports for both team-specific and network aggregate outcomes are reported back to the center.
 - Children's Health is working on completing the legal paperwork in early 2023 and will then start sharing deidentified data with ELHS and participate in this.
- In 2021 The Rett Syndrome Clinic was established and was granted recognition as a center of excellence and is drawing patients with this diagnosis from within and outside Texas to receive specialized care and to avail opportunities for participation in research.

Psychology Services

Psychological services in the Pediatric Epilepsy Center include behavioral health screening, clinical assessment, evidence-based intervention, and program development.

In 2017, the Pediatric Epilepsy Center began screening all teens for depressive symptoms. In 2022, screening expanded to include anxiety symptoms. Identification of depressive and anxiety symptoms provides quality, comprehensive care.

The Pediatric Epilepsy Center psychologist is actively involved in national workgroups and leadership roles.

Plans for 2023 include continued program development and expanding the Pediatric Epilepsy Center's new Psychogenic Nonepileptic Seizure Clinic that was started in 2022.

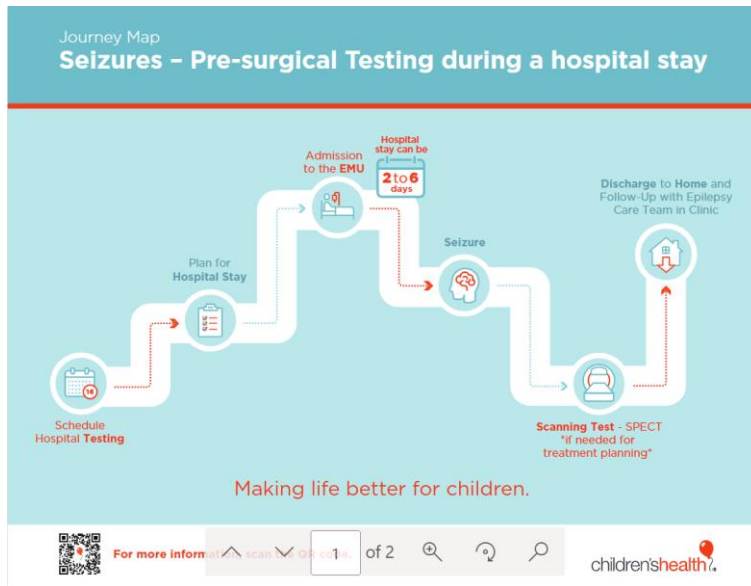
Outreach:

- **Epilepsy Camps:**
 - The faculty and staff of the Epilepsy Center continue to partner with the Epilepsy Foundation of Texas and provide medical staff to three epilepsy camps each year. The largest and most complex of these camps is the summer teen camp, Camp Kaleidoscope, which provides services to 100 teens with epilepsy and related neurological disabilities and is fully staffed by UTSW and Children's Medical Center staff for this weeklong camp.

- **Support Groups and Transition Education:**
 - Teen and Spanish support groups continued to meet in 2022.
 - Spanish support group meetings returned to in-person meetings with excellent patient and family participation.
 - The teen group participation was dwindling, likely due to virtual format of meetings, and we hope to rejuvenate the teen group meetings in 2023.

- **Family Conferences and Community Outreach:**
 - The Epilepsy Center newsletter is an important tool for epilepsy education in the outpatient clinic. This quarterly newsletter includes a patient feature, educational topic, clinic updates and introduction of new team members to our patients.
 - Epilepsy Center physicians also continue to serve on the professional advisory board of the Epilepsy Foundation of Texas and are regular speakers at their community events.
 - Epilepsy faculty were speakers at the epilepsy summer school webinar series conducted by epilepsy foundation of North Texas. Five of the nine speakers for the webinar series were UTSW pediatric epilepsy faculty.

- **Patient Resources:**
 - **EMU presurgical admission journey map:** collaboration with Child Life, Epilepsy team



- **Resources for barriers to medication adherence** made available in clinic rooms.

Patients who identify missed medication doses on the screening questionnaire are then asked about the types of barriers and specific tools that are being provided to address the barrier. An example is below:

Making Anti-Seizure Medicine Easier to Take



Sometimes taking anti-seizure medicine can be hard. Cutting pills in half, carrying liquid medicine, and keeping medicine cold can be a lot of work. Here are tips to make taking medicine easier.

Cutting Pills

- Ask your pharmacist about using a pill cutter. They are an easy tool to use if you have to cut your pills.
- Cut all the pills you will need for the week at the same time. This will make taking them fast and easy all week long!



Carrying Liquids

If it's hard to keep a full size bottle of liquid medicine with you, think about putting the anti-seizure medicine you need in a syringe. Place the syringe in a Ziploc® baggie or plastic container and keep it in a safe place.

- Local drug stores have syringes you can use for free, so be sure to ask. Try to get ones with a cap if you can.
- Ask your doctor/nurse for other tips on how to carry liquid medicine.



Keeping Anti-Seizure Medicines Cool

Some anti-seizure medicines need to stay cold. If you are on the go, this is hard!

- Packing medicine in a closed bag of ice or carrying medicine in a small cooler may help
- Be sure to ask your doctor/nurse if doing this would keep your medicine at the right temperature.
- If not, ask if your doctor/nurse can think of any ways to keep your medicine at the right temperature while you are on the go.



Taking Many Doses of Anti-Seizure Medicines

If taking more than one dose of anti-seizure medicine a day is hard, talk with your doctor/nurse about ways to change when you take your medicine.

- If you have trouble remembering to take your medicine consider setting up a reminder. You can use a pillbox, an alarm on your phone or a seizure diary to help you. Ask your doctor or nurse about what option might be right for you.



Find your reminder message



THE EPILEPSY FOUNDATION IS A SUSTAINING PARTNER OF THE EPILEPSY LEARNING HEALTHCARE SYSTEM (ELHS)

© 2017 adapted for epilepsy © 2020 Epilepsy Foundation.

- **Standardized epilepsy after visit summary** was developed by the epilepsy team, which includes clinic information, when to expect test results and educational information such as seizure safety precautions, the importance of vitamin D, local resources for support, and information about sudden unexpected death in epilepsy patients.
- **Transition work group:** the epilepsy team has created a transition work group and is collaborating with the transition team at Children's Health, care coordination, adult epilepsy team, and our patient family partners to create a systematic gradual approach for the transition of patients with epilepsy to adulthood. This work group was formed in 2022 and plans to continue to work on this in 2023 with iterative measures until the goal is achieved.
- **Epilepsy Awareness Month 2022 —Communications Key Takeaways**
 - During November, Epilepsy Awareness Month was featured in 23 social media posts, which is an increase of four posts from 2021.
 - Posts about Epilepsy Awareness Month on the Children's Health social media channels led to recognition for the team and encouragement for our patients.

Facebook Posts



Hayden Patient Story

Imp.	Eng.	Eng. Rate	Link Clicks
19,492	2,362	12.12%	N/A



Jack and Luke Patient Story

Imp.	Eng.	Eng. Rate	Link Clicks
28,860	1,940	6.72%	520



Reena Patient Story

Imp.	Eng.	Eng. Rate	Link Clicks
5,192	233	4.49%	N/A

- Dr. Price was interviewed live by WFAA; her interview was also posted online.
- Dr. Said was honored with the Person of the Year award by the Epilepsy Foundation of Texas for her outstanding contributions to the community.

LinkedIn Posts



Epilepsy Team

Imp.	Eng.	Eng. Rate	Clicks
6,191	2,343	37.85%	2,177



Dr. Rana Said

Imp.	Eng.	Eng. Rate	Clicks
19,122	1,981	10.36%	1,446



Hayden Patient Story

Imp.	Eng.	Eng. Rate	Clicks
3,342	185	5.54%	125



Nolan Patient Story

Imp.	Eng.	Eng. Rate	Clicks
3,382	180	5.32%	113

LinkedIn does not separate clicks, so this number reflects link, picture, hashtag, profile visits and tagged page clicks.

Research

- The Comprehensive Epilepsy Center continues to participate in clinical research with the faculty involved in IRB-approved clinical research studies as primary investigators (industry sponsored, non-funded) and co-investigators in NIH-funded trials.
- There were two industry clinical trials that started recruitment in 2022 and one that closed out after successful enrollment.
- There are multiple industry sponsored trials that were in the startup stage in 2022 with anticipated initiation in 2023.
- There are 10 non-funded research projects that were initiated in 2022 and are in various stages at UTSW amongst 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. There will be a roundtable discussion about rare epilepsies in the first week of February 2023.

- UTSW Pediatrics is 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 non-epileptic seizures.
- Many of our UTSW epilepsy physicians attended the AES Annual Meeting in Nashville, Tennessee, in December 2022. They had a strong presence and presented 10 posters at the conference. One faculty member also chaired the special interest group session on autoimmune epilepsy during the meeting, increasing the visibility of the organization at this premier national conference.
- Faculty members were invited to speak at meetings, regional symposiums, and to give grand rounds at other institutions.
 - In 2022, epilepsy physicians were invited to speak at the Latin American Congress of Childhood and Adolescent Psychiatry and associated subspecialties, the American Epilepsy Society meeting, the International Rett Syndrome Foundation national meeting, the International Child Neurology Association national meeting, and Epilepsy Awareness Day at Disneyland.
- Epilepsy faculty have published 15 manuscripts, including editorials, in peer-reviewed journals.
- One faculty member serves as a contributing editor to *Epilepsy Currents*, and other faculty serve 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, among others.

Priorities for 2023

The ultimate priority for the Comprehensive Epilepsy Center is to provide the highest quality medical care to children and adolescents with seizures and epilepsy. As part of this priority:

- Integrating EEG reports into the Children’s Health EPIC medical record system. The kick-off for this process started in August 2022 and is currently in the implementation stages. We are working closely with EPIC and information technology specialists to establish and implement this direct EPIC EEG reporting for Children’s Health patients.
- With the expanding surgical treatments for intractable epilepsy and an increase in referrals for surgical evaluations, we anticipate an increase in referrals for epilepsy surgery evaluations, magnetoencephalography, and intracranial EEG, which will lead to an increase in epilepsy surgery procedures, including laser thermoablation, RNS, and other devices for intractable epilepsy. The ability to perform presurgical evaluations at the Dallas and Plano EMUs will be essential. Dedicated EMU beds in Plano are needed to meet these needs.
- The EMU aims to meet the national standards for EEG technicians to monitor patients undergoing videoEEG studies at a 4:1 ratio of studies to technicians. We have faced the challenges of recruiting and retaining well-trained EEG technicians in 2022 for the Dallas and Plano EEG labs. Considerations are being given to looking for gap coverage for continuous EEG monitoring by technicians from external companies to fill this need. There is a need to train EEG technicians to monitor the ongoing EEGs, detect seizures when they occur, and report this to the physicians. In 2023, the EEG lab and epilepsy teams plan to work on developing a structured EEG technician training and competency evaluation program.
- The UTSW Division of Pediatric Neurology has provided leadership for the Plano campus level 3 epilepsy center since August 2022 and anticipates an increase in volumes for EEGs and EMU admissions in 2023. We hope to receive continued support to build the EMU and epilepsy center program.

- Expanding outreach services for teens with epilepsy is another important priority. Center physicians will continue to provide medical staff to the Epilepsy Foundation of Texas' epilepsy camp for more than 110 teens living with epilepsy each year.
- The center has started discussions and plans to establish epilepsy subspecialty clinics for Angelman syndrome, tuberous sclerosis, and infantile spasms in 2023-24. These dedicated clinics would provide collaborative, multidisciplinary care that incorporates national standards and guidelines, improve access for patients to clinical care, track outcomes, and allow for novel research advances. The center hopes to gain recognition as Angelman and Tuberous Sclerosis Complex Clinic/Centers of Excellence in 2023-24. As we build specialty clinics that focus on genetic epilepsy syndromes, there is a need for a dedicated genetic counselor for epilepsy, and we want to focus on adding that in 2023.
- Our Epilepsy Center's participation in the ELHS will continue to further promote local and national initiatives in quality care. We plan to complete the legal process for participation in the ELHS registry.
- Increasing involvement in research is an important priority for 2023 and the future. The time taken to get clinical trials initiated due to regulatory and contract agreements and other research requirements has been over 9 –12 months, and this is a major deterrent to attracting organizations to UTSW and Children's Health for significant epilepsy trials. Participation in multicenter studies through the Pediatric Epilepsy Research Consortium and participation in natural history studies, anti-seizure device and medication trials, epilepsy surgery, and neuromodulation trials are key priorities.
- Increasing regional, national, and international visibility through lectures, publications, leadership, and advertisements at conferences is a goal for 2023.

Current Grant/Contract Support

Dallas Armstrong

Contractor: International Rett Syndrome Foundation
Title of Project: IRSF Center of Excellence Clinical Support Agreement
Role: Principal Investigator
Dates: 07/2021 – 05/2023

Rachel Bailey

Grantor: Hannah's Hope Fund
Title of Project: GAN Vagus Nerve Injection Studies
Role: Co-Principal Investigator
Dates: 11/2019 – 01/2022

Kaitlin Batley

Grantor: CURE SMA
Title of Project: Cure SMA Clinical Data Registry
Role: Principal Investigator
Dates: 06/2019 – 12/2023

Grantor: CURESPG50
Title of Project: CureSPG50
Role: Co-Principal Investigator
Dates: 01/2023 – 07/2028

Contractor: Fibrogen Inc

Title of Project: Trial of Pamrevlumab (FG-3019), a Monoclonal Antibody to Connective Tissue Growth Factor, in Non-Ambulatory Subjects with Duchenne Muscular Dystrophy

Role: Principal Investigator

Dates:

Contractor: Medpace Inc

Title of Project: Phase 2 Active Treatment Study to Evaluate the Efficacy and Safety of SRK-015 in Patients with Later-Onset Spinal Muscular Atrophy

Role: Principal Investigator

Dates:

Contractor: Fibrogen Inc

Title of Project: Phase 3, Randomized, Double-Blind, Trial of Pamrevlumab (FG-3019) or Placebo in Combination with Systemic Corticosteroids in Subjects with Non-ambulatory Duchenne Muscular Dystrophy (DMD)

Role: Principal Investigator

Dates:

Wilmot Bonnet

Grantor: Hospital for Sick Children

Title of Project: Canadian Pediatric Stroke Imaging Research Platform: Harnessing An International Focus

Role: Principal Investigator

Dates: 08/2020 – 03/2022

Xin Chen

Grantor: CURESPG50

Title of Project: SPG50 Gene Therapy

Role: Co-Principal Investigator

Dates: 02/2020 – 07/2023

Contractor: The Orphan Disease Center/University Of Pennsylvania

Title of Project: Conquer from Within – Treating APBD by viral delivery of cross-correction-enabled amylase

Role: Co-Investigator

Dates: 02/2021 – 01/2022

Contractor: Sappani Family Foundation

Title of Project: SRD5A3 Gene Therapy

Role: Co-Investigator

Dates: 02/2020 – 07/2022

Contractor: Taysha Gene Therapies

Title of Project: CLN7 Gene replacement therapy for Batten disease

Role: Co-Investigator

Dates: 10/2021 – 06/2022

Contractor: Taysha Gene Therapies

Title of Project: DDX3X Gene Therapy

Role: Investigator

Dates: 12/2020 – 11/2022

Contractor: Taysha Gene Therapies
Title of Project: CMT4A Gene Therapy
Role: Investigator
Dates: 03/2021 – 02/2024

Contractor: Taysha Gene Therapies
Title of Project: GNAO1 Gene Replacement Therapy
Role: Investigator
Dates: 04/2021 – 03/2023

Contractor: DDX3X Foundation
Title of Project: DDX3X Gene Therapy
Role: Investigator
Dates: 12/2022 – 11/2023

Michael Dowling

Grantor: Nih-National Inst of Neuro Disord & Strk/University of California, San Francisco
Title of Project: The Vascular effects of Infection in Pediatric Stroke (VIPS II)
Role: Principal Investigator
Dates: 07/2021 – 06/2023

Grantor: NIH-National Inst of Neuro Disord & Strk/University of California, San Francisco
Title of Project: Seizures and Children's Outcomes after Stroke (SCOUTS)
Role: Principal Investigator
Dates: 01/2022 – 12/2026

Grantor: Nih-National Inst of Neuro Disord & Strk/Washington University in St. Louis
Title of Project: The role of metabolic and hemodynamic reserve in age-related brain vulnerability in pediatric sickle cell anemia
Role: Principal Investigator
Dates: 06/2021 – 03/2026

Patricia Evans

Grantor: National Fragile X Foundation
Title of Project: National Fragile X Clinical & Research Consortium (FXCRX)
Role: Principal Investigator
Dates: 10/2011 – 08/2025

Grantor: National Fragile X Foundation
Title of Project: National Fragile X Clinical & Research Consortium (FXCRX)
Role: Principal Investigator
Dates: 09/2020 – 08/2025

Grantor: Zynherba Pharmaceuticals Inc.
Title of Project: A Randomized, Double-Blind, Placebo-Controlled Multiple-Center, Efficacy and Safety Study of ZYN002 Administered as a Transdermal Gel to Children and Adolescents with Fragile X Syndrome - RECONNECT
Role: Principal Investigator
Dates:

Kimberly Goodspeed

Grantor: SLC6A1 CONNECT

Title of Project: A Natural History and Outcome Measure Discovery Study of Developmental and Epileptic Encephalopathies

Role: Principal Investigator

Dates: 07/2020 – 06/2025

Grantor: Board of Trustees of the Leland Stanford/Tess Research Foundation

Title of Project: International SLC13A5 Natural History Study

Role: Principal Investigator

Dates: 11/2020 – 11/2023

Contractor: Neuren Pharmaceuticals Ltd

Title of Project: An Open-Label Study of the Safety, Tolerability, and Pharmacokinetics of Oral NNZ-2591 in Pitt-Hopkins Syndrome

Role: Principal Investigator

Dates: 03/2023 – 03/2024

Contractor: UTSW KL2 Scholar

Title of Project: NIH-funded career development award supporting transition to independence and development of the SLC6A1-NDD clinical research program

Role: Co-Investigator

Dates: 2022 – 2024

Contractor: Dedman Scholar In Clinical Care Award

Title of Project: Endowed scholar award for excellence in clinical care and clinical research. Funds support many projects in genetic neurodevelopmental clinical characterization and a patient registry for genetic NDDs

Role: Principal Investigator

Dates: 2020 – 2024

Contractor: SLC6A1 Connect

Title of Project: Patient foundation research award to support the pilot clinical trial readiness study of SLC6A1-NDD

Role: Principal Investigator

Dates: 2020 – 2025

Contractor: Stanford University/TESS Research Foundation

Title of Project: Natural history study of SLC13A5 deficiency disorder that includes both remote and in-person participation

Role: Principal Investigator

Dates: 2020 – 2024

Contractor: Community Based Research Award/UTSW Endowment

Title of Project: Pilot award to measure efficacy of a social-emotional learning program for pre-school-aged children with autism spectrum disorder in partnership with the Dallas Zoo

Role: Principal Investigator

Dates: 2020 – 2022

Contractor: Neuren Pharmaceuticals Ltd

Title of Project: An Open-Label Study of the Safety, Tolerability, and Pharmacokinetics of Oral NNZ-2591 in Pitt-Hopkins Syndrome

Role: Principal Investigator

Dates: 2022

Contractor: Bespoke Gene Therapy Consortium

Title of Project: NIH Consortium award to support a gene transfer therapy clinical trial for Multiple Sulfatase Deficiency

Role: Co-Investigator

Dates: 2023

Contractor: NIH Ultra Rare Gene Therapy (URGenT, PAR-22-030)

Title of Project: NIH U01 to support a gene transfer therapy clinical trial for aspartylglucosaminuria.

Role: Co-Investigator

Dates: 2023

Contractor: UTSW OCHRE Community Health Research Program

Title of Project: Phase 1: Planning grant to support partnership with the Vanguard School, a private school for individuals with autism spectrum disorder. Study will develop a protocol to assess efficacy of non-invasive heart rate monitoring as a biomarker of psychological distress in the classroom.

Role: Principal Investigator

Dates: 2023

Contractor: NIH-NINDS K23 Career Development Award

Title of Project: Initial submission of a K23 application to the NIH-NINDS career development award program. The project focuses on clinical trial readiness, biomarker development, and disease concept model development in SLC6A1-NDD

Role: Principal Investigator

Dates: 2023

Steven Gray

Contractor: DDX3X Foundation

Title of Project: Timing of therapeutic intervention for DDX3X syndrome (Predoctoral fellowship)

Role: Mentor

Dates: 12/2022 – 11/2023

Contractor: DDX3X Foundation

Title of Project: DDX3X Gene Therapy

Role: Principal Investigator

Dates: 12/2022 – 11/2023

Contractor: Phoenix Nest

Title of Project: MPS IIIC gene therapy: preclinical in vivo assessment of treatment efficacy

Role: Principal Investigator

Dates: 12/2022 – 11/2024

Contractor: MOAD Foundation

Title of Project: MOA-A/B Gene Therapy

Role: Principal Investigator

Dates: 04/2022 – 03/2024

Contractor: Form Bio

Title of Project: Genome Stability and Optimization of AAV Vector Genomes

Role: Principal Investigator

Dates: 12/2022 – 11/2023

Contractor: NIH-National Inst of Neuro Disord & Strk/University of Pennsylvania

Title of Project: AAV-mediated gene therapy for CNS disease correction in feline NPC1 disease

Role: Principal Investigator

Dates: 12/2020 – 11/2025

Contractor: NIH-National Inst of Neuro Disord & Strk

Title of Project: Directed Evolution of Novel AAV Capsids for Global CNS Gene Delivery in Rodents and Primates

Role: Principal Investigator

Dates: 08/2018 – 11/2023

Contractor: Foundation for Prader-Willi Research

Title of Project: shRNA/AAV9 Gene Therapy for the Treatment of Prader-Willi Syndrome

Role: Co-Investigator

Dates: 08/2022 – 01/2024

Contractor: CURESPG50

Title of Project: SPG50 Gene Therapy

Role: Principal Investigator

Dates: 02/2020 – 07/2023

Contractor: Hannah's Hope Fund

Title of Project: GAN Vagus Nerve Injection Studies

Role: Principal Investigator

Dates: 11/2019 – 01/2022

Contractor: Hannah's Hope Fund

Title of Project: GAN Vagus Nerve Translational Studies

Role: Principal Investigator

Dates: 12/2022 – 11/2024

Contractor: NIH-National Inst of Chld Hlth & Hmn Dev

Title of Project: First murine animal model and adeno-associated virus (AAV)-based gene therapy for MTATP6 mitochondrial diseases

Role: Mentor

Dates: 08/2022 – 07/2023

Contractor: Taysha Gene Therapies

Title of Project: FMR1 Gene replacement therapy for Fragile X syndrome

Role: Principal Investigator

Dates: 09/2020 – 08/2022

Contractor: Drake Rayden Foundation

Title of Project: NKH (GLDC) Gene Therapy: First Steps

Role: Principal Investigator

Dates: 08/2020 – 01/2022

Contractor: Taysha Gene Therapies

Title of Project: Vagus Nerve non-human primate proof of concept

Role: Principal Investigator

Dates: 07/2020 – 06/2022

Contractor: Taysha Gene Therapies

Title of Project: Preclinical assessment of a regulated miniMeCP2 vector

Role: Co-Investigator

Dates: 05/2020 – 04/2022

Contractor: Taysha Gene Therapies
Title of Project: SURF1 Gene Therapy
Role: Principal Investigator
Dates: 04/2020 – 09/2023

Contractor: Taysha Gene Therapies
Title of Project: SLC6A1 Gene Therapy
Role: Principal Investigator
Dates: 04/2020 – 04/2024

Contractor: Taysha Gene Therapies
Title of Project: Sponsored Research Agreement C
Role: Principal Investigator
Dates: 01/2022 – 09/2022

Contractor: Taysha Gene Therapies
Title of Project: CLN7 gene replacement for Batten disease
Role: Principal Investigator
Dates: 10/2021 – 09/2022

Contractor: Taysha Gene Therapies
Title of Project: CMT4A Gene Therapy
Role: Co-Investigator
Dates: 12/2020 – 11/2023

Contractor: Taysha Gene Therapies
Title of Project: DDX3X Gene Therapy
Role: Co-Investigator
Dates: 12/2020 – 11/2022

Contractor: Taysha Gene Therapies
Title of Project: Gene therapy for autism and tumor phenotypes related to PTEN Hamartoma Tumor Syndrome
Role: Co-Investigator
Dates: 06/2020 – 05/2022

Contractor: Cure SPG50 Foundation
Title of Project: AP4M1 gene therapy
Role: Principal Investigator
Dates: 02/2020 – 07/2023

Contractor: Sappani Foundation
Title of Project: SRD5A3 gene therapy
Role: Principal Investigator
Dates: 02/2020 – 07/2022

Susan Iannaccone

Contractor: Muscular Dystrophy Association
Title of Project: MDA care center
Role: Principal Investigator

Dates: 07/2021 – 06/2024

Contractor: NIH-National Inst of Neuro Disord & Strk

Title of Project: UTSW Clinical Research Site for Network of Excellence in Neuroscience Clinical Trials (NeuroNEXT)

Role: Co- Investigator

Dates: 07/2018 – 06/2023

Contractor: NIH-National Inst of Neuro Disord & Strk

Title of Project: Myoediting of Duchenne muscular dystrophy

Role: Co-Principal Investigator

Dates: 09/2020 – 08/2025

Contractor: Parent Project Muscular Dystrophy (PPMD)

Title of Project: Parent Project Muscular Dystrophy Certification

Role: Principal Investigator

Dates: 01/2022 – 12/2022

Contractor: US Department of Defense/Massachusetts General Hospital

Title of Project: W81XWH-20-1-0293, “Extracellular RNA Biomarkers of Myotonic Dystrophy”

Role: Principal Investigator

Dates: 06/2020 – 05/2023

Contractor: SAREPTA

Title of Project: Long-term, Open-label Extension Study for Patients with Duchenne Muscular Dystrophy Enrolled in Clinical Trials Evaluating Casimersen or Golodirsen

Role: Principal Investigator

Dates:

Contractor: SAREPTA

Title of Project: A Double-Blind, Placebo-Controlled, Multi-Center Study with an Open-Label Extension to Evaluate the Efficacy and Safety of SRP-4045

Role: Principal Investigator

Dates:

Contractor: Ionis Pharmaceuticals Inc

Title of Project: An Open label Extension Study for Patients with Spinal Muscular Atrophy who Previously Participated in Investigational Studies of ISIS 396443

Role: Principal Investigator

Dates:

Contractor: SAREPTA

Title of Project: A Phase 3 Multinational, Randomized, Double-Blind, Placebo-Controlled Systemic Gene Delivery Study to Evaluate the Safety and Efficacy of SRP-9001 in Subject With Duchenne Muscular Dystrophy (EMBARK)

Role: Principal Investigator

Dates:

Contractor: BIOGEN MA INC

Title of Project: A Long-Term Extension Study of Nusinersen (BIIB058) Administered at Higher Doses in Participants With Spinal Muscular Atrophy Who Previously Participated in an Investigational Study With Nusinersen

Role: Principal Investigator

Dates:

Contractor: SAREPTA

Title of Project: A Phase 2, Two-Part, Multiple-Ascending-Dose Study of SRP-5051 for Dose Determination, then Dose Expansion, in Patients with Duchenne Muscular Dystrophy Amenable to Exon 51-Skipping Treatment

Role: Principal Investigator

Dates:

Contractor: US Department of Defense/Massachusetts General Hospital

Title of Project: Extracellular RNA Biomarkers of Myotonic Dystrophy

Role: Principal Investigator

Dates: 06/2020 – 05/2023

Contractor: PTC Therapeutics

Title of Project: An Open-Label, Safety Study for ATALUREN (PTC124) Patients with Nonsense Mutation Dystrophinopathy

Role: Site Principal Investigator

Dates: Present

Contractor: AveXis/Novartis

Title of Project: Phase 1 Open-Label, Dose Comparison Study of AVXS-101 for Sitting but Non-Ambulatory Patients with Spinal Muscular Atrophy

Role: Site Principal Investigator

Dates: Present

Contractor: AveXis/Novartis

Title of Project: A Long-term Follow-up Study of Patients in the Clinical Trials for Spinal Muscular Atrophy Receiving AVXS-101

Role: Site Principal Investigator

Dates: Present

Contractor: Capricor

Title of Project: DMD cell based therapy

Role: Site Principal Investigator

Dates: Present

Contractor: RegenexBIO

Title of Project: DMD gene therapy

Role: Site Principal Investigator

Dates: Present

Contractor: NIH

Title of Project: Wellstone Muscular Dystrophy Center

Role: Associate Director and co-chair of Administrative Core, PI Eric Olson, PhD

Dates: 2020 – 2025

Contractor: MDA Pediatric Care Center

Title of Project: Support for PI and clinic nurse time

Role: PI (Co-Director)

Dates: 2018 – 2023

Contractor: Parent Project for Muscular Dystrophy (PPMD) Certified Clinic

Title of Project: Support for clinic activities in support of patient special needs

Role: PI (Co-Director)

Dates: 2019 – 2023

Contractor: Sarepta

Title of Project: A Study of the Natural History of Patients with LGMD2E/R4, LGMD2D/R3 and LGMD2C/R5;>4 Years of Age, Who are Managed in Routine Clinical Practice

Role: Site Principal Investigator

Dates: 2021 – Present

Contractor: Neurogene

Title of Project: A Natural History Study of Charcot-Marie-Tooth 4J (CMT4J)

Role: Site Principal Investigator

Dates: 2019 – Present

Contractor: Scholar Rock, Inc.

Title of Project: An Active Treatment Study of SRK-015 in Patients With Type 2 or Type 3 Spinal Muscular Atrophy (TOPAZ)

Role: Site Principal Investigator

Dates: 2019 – Present

Contractor: ReveraGen BioPharma, Inc.

Title of Project: A phase 2a open label, multiple ascending dose study to assess the safety, tolerability, pharmacodynamics and exploratory efficacy of Vamorolone in Boys with Duchenne Muscular Dystrophy

Role: Site Principal Investigator

Dates: 2016 – Present

Saima Kayani

Contractor: NIH-National Inst of Neuro Disord & Strk

Title of Project: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D)

Role: Co-Principal Investigator

Dates: 09/2016 – 01/2023

Contractor: NIH-National Inst of Neuro Disord & Strk

Title of Project: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D)

Role: Co-Principal Investigator

Dates: 08/2021 – 07/2022

Contractor: Investigator Initiated

Title of Project: Phase 1 intrathecal lumbar administration of AAV/CLN7 for treatment of CLN7 disease

Role: Co-Investigator

Dates: 2020 – 2024

Berge Minassian

Contractor: NIH-National Inst Of Neuro Disord & Strk/University of Kentucky Research Fndn

Title of Project: Lafora Epilepsy - Basic Mechanisms to Therapy (Admin Core)

Role: Principal Investigator

Dates: 12/2022 – 11/2024

Contractor: The Orphan Disease Center/University of Pennsylvania

Title of Project: Conquer from Within – Treating APBD by viral delivery of cross-correction-enabled amylase

Role: Principal Investigator

Dates: 02/2021 – 07/2023

Contractor: Chan Zuckerberg Initiative

Title of Project: Closing the knowledge gaps in the fatal Lafora neurodegenerative disease
Role: Principal Investigator
Dates: 07/2021 – 06/2023

Contractor: UTSW Internal
Title of Project: Quantification of Glucose-6-Phosphate (G6P) and Gamma-Aminobutyric Acid (GABA) in the brain using 7T MRI or 3T Spectroscopy in Lafora Disease and Neurogenetic Diseases.
Role: Principal Investigator
Dates: 06/2023 – 05/2024

Contractor: Taysha Gene Therapies
Title of Project: SRA202002-0010
Role: Principal Investigator
Dates: 06/2020 – 05/2023

Contractor: Taysha Gene Therapies
Title of Project: Sponsored Research Agreement S
Role: Principal Investigator
Dates: 2020 – 2023

Contractor: Ionis Pharmaceuticals Inc.
Title of Project: Prospective, Longitudinal, Obs
Role: Principal Investigator
Dates: 06/2023 – 05/2024

Sharmistha Mitra

Contractor: Chan Zuckerberg Initiative
Title of Project: Closing the knowledge gaps in the fatal Lafora neurodegenerative disease
Role: Principal Investigator
Dates: 12/2022 – 11/2024

Felix Nitschke

Contractor: NIH-National Inst of Neuro Disord & Strk
Title of Project: Uncovering cellular mechanisms to keep glycogen water-soluble
Role: Principal Investigator
Dates: 05/2022 – 04/2027

Contractor: The Orphan Disease Center/University of Pennsylvania
Title of Project: Conquer from Within – Treating APBD by viral delivery of cross-correction-enabled amylase
Role: Principal Investigator
Dates: 02/2021 – 07/2023

Contractor: Chan Zuckerberg Initiative
Title of Project: Closing the knowledge gaps in the fatal Lafora neurodegenerative disease
Role: Co-Principal Investigator
Dates: 12/2022 – 11/2024

Eric Remster

Contractor: Eli Lilly & Company

Title of Project: Protocol I5Q-MC-CGAS(f)A Randomized, Double-Blind, Placebo-Controlled Study of Galcanezumab in Patients 6 to 17 Years of Age with Episodic Migraine – the REBUILD-1 Study

Role: Site Principal Investigator

Dates:

Contractor: Lilly USA, LLC

Title of Project: LY2951742 Protocol Addendum I5Q-MC-CGAS(1.1)

Role: Site Principal Investigator

Dates: 01/2019 – 12/2024

Rana Said

Contractor: NIH-National Inst of Neuro Disord & Strk/Baylor College of Medicine

Title of Project: Pediatric Dose Optimization for Seizures in EMS (PediDOSE)

Role: Co-Principal Investigator

Dates: 08/2021 – 11/2022

Contractor: Marinus

Title of Project: A Double-Blind, Randomized, Placebo-Controlled Study to Evaluate the Efficacy and Safety of Intravenous Ganaxolone in Status Epilepticus (STU 2020-0740)

Role: Principal Investigator

Dates: 06/2020 – 11/2024

Sarah Sinnett

Grantor: UTSW

Title of Project: Inventor funds (non-grant funding)

Role: Principle Investigator

Dates: 09/2022 – no expiration

Grantor: The Orphan Disease Center/University of Pennsylvania

Title of Project: Conquer from Within – Treating APBD by viral delivery of cross-correction-enabled amylase

Role: Co-Investigator

Dates: 02/2021 – 07/2023

Grantor: Taysha Gene Therapies

Title of Project: FOYG1 Gene Therapy Feasibility and Risk Assessment

Role: Principal Investigator

Dates: 07/2020 – 07/2023

Grantor: Taysha Gene Therapies

Title of Project: Efficacy of FOYG1 Gene Therapies

Role: Principal Investigator

Dates: 01/2022 – 12/2023

Grantor: Taysha Gene Therapies
Title of Project: Pre-clinical assessment of a regulated miniMECP2 vector
Role: Principal Investigator
Dates: 05/2020 – 11/2023

Grantor: Taysha Gene Therapies
Title of Project: Gene therapy for autism and tumor phenotypes related to PTEN Hamartoma Tumor Syndrome
Role: Principal Investigator
Dates: 06/2020 – 05/2023

Deepa Sirsi

Grantor: NIH-National Inst of Neuro Disord & Strk
Title of Project: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D)
Role: Co-Principal Investigator
Dates: 09/2016 – 01/2023

Grantor: NIH-National Inst of Neuro Disord & Strk
Title of Project: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D)
Role: Co-Principal Investigator
Dates: 08/2021 – 07/2022

Grantor: Tess Research Foundation/Board of Trustees of The Leland Stanford
Title of Project: International SLC13A5 Natural History Study
Role: Co-Principal Investigator
Dates: 12/2021 – 11/2024

Grantor: Zogenix, Inc.
Title of Project: An Open-Label Extension Trial to Assess the Long-Term Safety of ZX008 (Fenfluramine Hydrochloride) Oral Solution as an Adjunctive Therapy for Seizures in Patients with Rare Seizure Disorders Such as Epileptic Encephalopathies including Dravet Syndrome
Role: Principal Investigator
Dates: 05/2020 – 08/2022

Contractor: Neurelis, Inc
Title of Project: An Open-Label, Single-Dose, Pharmacokinetics Study of Valtoco® With Open-Label Safety Period in Pediatric Subjects With Epilepsy
Role: Principal Investigator
Dates: 12/2022 – Present

Mathew Stokes

Grantor: Texas Institute of Brain Injury and Repair
Title of Project: Endophenotypes of Persistent Post-Concussive Symptoms in Adolescents: CARE4KIDS
Role: Co-Investigator
Dates: 09/2021 – 08/2022

Grantor: Texas Institute of Brain Injury and Repair
Title of Project: Endophenotypes of Persistent Post-Concussive Symptoms in Adolescents: CARE4KIDS
Role: Co-Investigator
Dates: 09/2022 – 08/2023

Grantor: Texas Institute of Brain Injury and Repair
Title of Project: North Texas Sports Concussion Network (CON-TEX)
Role: Principal Investigator
Dates: 07/2019 – 08/2023

Jeff Waugh

Grantor: NIH-National Inst of Neuro Disord & Strk
Title of Project: The structural underpinnings of disinhibition in dystonia
Role: Principal Investigator
Dates: 08/2022 – 07/2027

Grantor: American Academy of Neurology,
Title of Project: 2017 Career Development Award: Evaluating a potential mechanism for dystonia: the role of impaired inhibition
Role: Principal Investigator
Dates: 02/2018 – 01/2023

Peer-Reviewed Publications

1. Abend N, Adams E, Al Balushi A, Alburaki W, Appendino J, Barbosa VS, Birca A, Bonifacio S, Branagan A, Chang T, Chowdhury R, Christou H, Chu C, Cilio MR, Comani S, Corsi-Cabrera M, Croce P, Cubero-Rego L, Dawoud F, de Vries L, Dehaes M, Devane D, Duncan A, El Ters N, El-Dib M, Elshibiny H, Esser M, Fairchild K, Finucane E, Franceschini MA, Gallagher A, Ghosh A, Glass H, Venkata SKRG, Baillet TH, Herzberg E, Hildrey E, Hurley T, Inder T, Jacobs E, Jefferies K, Jermendy A, Khazaei M, Kilmartin K, King G, Lauronen L, Lee S, Leijser L, Lind J, Llaguno NS, **Machie M**, Magalhães M, Mahdi Z, Maluomi J, Marandyuk B, Massey S, McCulloch C, Metsäranta M, Mikkonen K, Mohammad K, Molloy E, Momin S, Munster C, Murthy P, Netto A, Nevalainen P, Nguyen J, Nieves M, Nyman J, Oliver N, Peeters C, Pietrobom RFR, Pijpers J, Pincheffsky E, Ping YB, Quirke F, Raeisi K, Ricardo-Garcell J, Robinson J, Rodrigues DP, Rosati J, Scott J, Scringer-Wilkes M, Shellhaas R, Smit L, Soul J, Srivastava A, Steggerda S, Sunwoo J, Szakmar E, Tamburro G, Thomas S, Toiviainen-Salo S, Toma AI, Vanhatalo S, Variane GFT, Vein A, Vesoulis Z, Vilan A, Volpe J, Weeke L, Wintermark P, Wusthoff C, Zappasodi F, Zein H, Zempel J. [Proceedings of the 13th International Newborn Brain Conference: Neonatal Neurocritical Care, Seizures, and Continuous EEG monitoring.](#) *J Neonatal Perinatal Med.* 2022;15(2):467-485. PMID:35431189
2. Bain JM, Snyder LG, Helbig KL, Cooper DD, Chung WK, **Goodspeed K.** [Consistency of parent-report SLC6A1 data in Simons Searchlight with Provider-Based Publications.](#) *J Neurodev Disord.* 2022 Jun 28;14(1):40. PMID:35761184
3. Balagura G, Xian J, Riva A, Marchese F, Ben Zeev B, Rios L, **Sirsi D**, Accorsi P, Amadori E, Astrea G, Baldassari S, Beccaria F, Boni A, Budetta M, Cantalupo G, Capovilla G, Cesaroni E, Chiesa V, Coppola A, Dilena R, Faggioli R, Ferrari A, Fiorini E, Madia F, Gennaro E, Giacomini T, Giordano L, Iacomino M, Lattanzi S, Marini C, Mancardi MM, Mastrangelo M, Messina T, Minetti C, Nobili L, Papa A, Parmeggiani A, Pisano T, Russo A, Salpietro V, Savasta S, Scala M, Accogli A, Scelsa B, Scudieri P, Spalice A, Specchio N, Trivisano M, Tzadok M, Valeriani M, Vari MS, Verrotti A, Vigeveno F, Vignoli A, Toonen R, Zara F, Helbig I, Striano P. [Epilepsy Course and Developmental Trajectories in STXBP1-DEE.](#) *Neurol Genet.* 2022 Jun;8(3):e676. PMID:35655584
4. Beslow LA, Agner SC, Santoro JD, Ram D, Wilson JL, Harrar D, Appavu B, Fraser SM, Rossor T, Torres MD, Kossorotoff M, Zuñiga Zambrano YC, Hernández-Chávez M, Hassanein SMA, Zafeiriou D, **Dowling MM**, Kopyta I, Stence NV, Bernard TJ, Dlamini N, International Pediatric Stroke Study Group.

[International Prevalence and Mechanisms of SARS-CoV-2 in Childhood Arterial Ischemic Stroke During the COVID-19 Pandemic.](#) *Stroke*. 2022 Aug;53(8):2497-2503. PMID:35380052

5. Bone M, **Goodspeed K**. [A Case Report of Topiramate for Severe Breath Holding Spells in a Teenage Boy with Pitt-Hopkins Syndrome.](#) *Child Neurol Open*. 2022 Jan-Dec;9():2329048X221093172. PMID:35449882
6. Bone M, **Goodspeed K**, **Sirsi D**. [Epilepsy and electroencephalography in Pitt-Hopkins syndrome.](#) *J Transl Genet Genom*. 2022;6:168-178. <https://doi.org/10.20517/jtgg.2021.56>
7. Bunt SC, Didehbani N, LoBue C, **Stokes M**, Heinzelmann M, Rossetti H, Miller SM, Nakonezny PA, Bell K, Batjer H, Cullum CM. [Sex differences in reporting of concussion symptoms in adults.](#) *Clin Neuropsychol*. 2022 Aug;36(6):1290-1303. PMID:33258703
8. Campbell C, Leu C, Feng YA, Wolking S, Moreau C, Ellis C, Ganesan S, Martins H, Oliver K, Boothman I, Benson K, Molloy A, Brody L, Epi4K Collaborative, Genomics England Research Consortium, Michaud JL, Hamdan FF, **Minassian BA**, Lerche H, Scheffer IE, Sisodiya S, Girard S, Cosette P, Delanty N, Lal D, Cavalleri GL, Epi25 Collaborative. [The role of common genetic variation in presumed monogenic epilepsies.](#) *EBioMedicine*. 2022 Jul;81():104098. PMID:35679801
9. **Chen X**, Dong T, Hu Y, Shaffo FC, Belur NR, Mazzulli JR, **Gray SJ**. [AAV9/MFSD8 gene therapy is effective in preclinical models of neuronal ceroid lipofuscinosis type 7 disease.](#) *J Clin Invest*. 2022 Mar 1;132(5):. PMID:35025759
10. Cousin MA, Veale EL, Dsouza NR, Tripathi S, Holden RG, Arelin M, Beek G, Bekheirnia MR, Beygo J, Bhambhani V, Bialer M, Bigoni S, Boelman C, Carmichael J, Courtin T, Cogne B, Dabaj I, Doummar D, Fazilleau L, Ferlini A, Gavrilova RH, Graham JM Jr, Haack TB, Juusola J, Kant SG, **Kayani S**, Keren B, Ketteler P, Klöckner C, Koopmann TT, Kruisselbrink TM, Kuechler A, Lambert L, Latypova X, Lebel RR, Leduc MS, Leonardi E, Lewis AM, Liew W, Machol K, Mardini S, McWalter K, Mignot C, McLaughlin J, Murgia A, Narayanan V, Nava C, Neuser S, Nizon M, Ognibene D, Park J, Platzer K, Poirsier C, Radtke M, Ramsey K, Runke CK, Guillen Sacoto MJ, Scaglia F, Shinawi M, Spranger S, Tan ES, Taylor J, Trentesaux AS, Vairo F, Willaert R, Zadeh N, Urrutia R, Babovic-Vuksanovic D, Zimmermann MT, Mathie A, Klee EW. [Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome.](#) *Genome Med*. 2022 Jun 13;14(1):62. PMID:35698242
11. Davidson BL, Gao G, Berry-Kravis E, Bradbury AM, Bönnemann C, Buxbaum JD, Corcoran GR, **Gray SJ**, Gray-Edwards H, Kleiman RJ, Shaywitz AJ, Wang D, Zoghbi HY, Flotte TR, Tauscher-Wisniewski S, Tiffet CJ, Sahin M, Gene Therapy Workshop Faculty. [Gene-based therapeutics for rare genetic neurodevelopmental psychiatric disorders.](#) *Mol Ther*. 2022 Jul 6;30(7):2416-2428. PMID:35585789
12. Fredrich S, **Wang C**, Narayan R, Tardo L, Blackburn KM, Vernino S. [Refractory Anti-NMDA Receptor Encephalitis in Early Pregnancy: A Case Report of Treatment Course and Pregnancy Outcomes.](#) *Neurol Neuroimmunol Neuroinflamm*. 2022 Sep;9(5):. PMID:35728968
13. **Goodspeed K**, Horton D, **Lowden A**, Sguigna PV, Booth T, Wang ZJ, Edgar VB. [A cross-sectional natural history study of aspartylglucosaminuria.](#) *JIMD Rep*. 2022 Sep;63(5):425-433. PMID:36101820
14. **Goodspeed K**, Liu JS, Nye KL, Prasad S, Sadhu C, Tavakkoli F, Bilder DA, **Minassian BA**, **Bailey RM**. [SLC13A5 Deficiency Disorder: From Genetics to Gene Therapy.](#) *Genes (Basel)*. 2022 Sep

15;13(9):. PMID:36140822

15. **Goodspeed K, Bailey RM**, Prasad S, Sadhu C, Cardenas JA, Holmay M, Bilder DA, **Minassian BA**. [Gene Therapy: Novel Approaches to Targeting Monogenic Epilepsies](#). *Front Neurol*. 2022;13():805007. PMID:35847198
16. Govindarajan R, Vu AN, Salas RME, Miller AM, Sandness DJ, **Said RR**, Southerland AM, Fernandez A, Romano S, Sennott BJ, Patino-Murillas J, Soni M, AAN Undergraduate Education Subcommittee and Education Committee. [Accelerated Implementation of a Virtual Neurology Clerkship Amid a Global Crisis](#). *Neurology*. 2022 Feb 15;98(7):279-286. PMID:34921103
17. Gumusgoz E, Kasiri S, Guisso DR, Wu J, Dear M, Verhalen B, **Minassian BA**. [AAV-Mediated Artificial miRNA Reduces Pathogenic Polyglucosan Bodies and Neuroinflammation in Adult Polyglucosan Body and Lafora Disease Mouse Models](#). *Neurotherapeutics*. 2022 Apr;19(3):982-993. PMID:35347645
18. Heinzelmann M, Woods C, Vargas BB, **Stokes M**. [Posttraumatic Headache](#). *Semin Neurol*. 2022 Aug;42(4):428-440. PMID:36041477
19. Herzberg EM, **Machie M**, Glass HC, Shellhaas RA, Wusthoff CJ, Chang T, Abend NS, Chu CJ, Cilio MR, Bonifacio SL, Massey SL, McCulloch CE, Soul JS, Neonatal Seizure Registry study group. [Seizure Severity and Treatment Response in Newborn Infants with Seizures Attributed to Intracranial Hemorrhage](#). *J Pediatr*. 2022 Mar;242():121-128.e1. PMID:34780777
20. **Iannaccone ST**. [New results for risdiplam in spinal muscular atrophy](#). *Lancet Neurol*. 2022 Dec;21(12):1065-1066. PMID:36244363
21. Khan TR, **Dolce A, Goodspeed K**. [A case of Bainbridge-Ropers syndrome with breath holding spells and intractable epilepsy: challenges in diagnosis and management](#). *BMC Neurol*. 2022 Feb 16;22(1):60. PMID:35172777
22. Lidstone SC, Costa-Parke M, Robinson EJ, Ercoli T, Stone J, FMD GAP Study Group. [Functional movement disorder gender, age and phenotype study: a systematic review and individual patient meta-analysis of 4905 cases](#). *J Neurol Neurosurg Psychiatry*. 2022 Jun;93(6):609-616. PMID:35217516
23. MacCannell D, Berger Z, Kirschner J, Mercuri E, Farrar MA, **Iannaccone ST**, Kuntz NL, Finkel RS, Valente M, Muntoni F. [Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model](#). *CNS Drugs*. 2022 Feb;36(2):181-190. PMID:35080757

24. McKnight D, Morales A, Hatchell KE, Bristow SL, Bonkowsky JL, Perry MS, Berg AT, Borlot F, Esplin ED, Moretz C, Angione K, Ríos-Pohl L, Nussbaum RL, Aradhya S, ELEVATE Consortium, Haldeman-Englert CR, Levy RJ, Parachuri VG, Lay-Son G, de Montellano DJD, Ramirez-Garcia MA, Benítez Alonso EO, Ziobro J, Chirita-Emandi A, Felix TM, Kulasa-Luke D, Megarbane A, Karkare S, Chagnon SL, Humberson JB, Assaf MJ, Silva S, Zarroli K, Boyarchuk O, Nelson GR, Palmquist R, Hammond KC, Hwang ST, Boutlier SB, Nolan M, **Batley KY**, Chavda D, Reyes-Silva CA, Miroshnikov O, Zuccarelli B, Amlie-Wolf L, Wheless JW, Seinfeld S, Kanhangad M, Freeman JL, Monroy-Santoyo S, Rodriguez-Vazquez N, Ryan MM, **Machie M**, Guerra P, Hassan MJ, Candee MS, Bupp CP, Park KL, Muller E 2nd, Lupo P, Pedersen RC, Arain AM, Murphy A, Schatz K, Mu W, Kalika PM, Plaza L, Kellogg MA, Lora EG, Carson RP, Svystilnyk V, Venegas V, Luke RR, Jiang H, Stetsenko T, Dueñas-Roque MM, Trasmonte J, Burke RJ, Hurst ACE, Smith DM, Massingham LJ, Pisani L, Costin CE, Ostrander B, Filloux FM, Ananth AL, Mohamed IS, Nechai A, Dao JM, Fahey MC, Aliu E, Falchek S, Press CA, Treat L, Eschbach K, Starks A, Kammeyer R, Bear JJ, Jacobson M, Chernuha V, Meibos B, Wong K, Sweney MT, Espinoza AC, Van Orman CB, Weinstock A, Kumar A, Soler-Alfonso C, Nolan DA, Raza M, Rojas Carrion MD, Chari G, Marsh ED, Shiloh-Malawsky Y, Parikh S, Gonzalez-Giraldo E, Fulton S, Sogawa Y, Burns K, Malets M, Montiel Blanco JD, Habela CW, Wilson CA, Guzmán GG, Pavliuk M. [Genetic Testing to Inform Epilepsy Treatment Management From an International Study of Clinical Practice](#). *JAMA Neurol*. 2022 Dec 1;79(12):1267-1276. PMID:36315135
25. **Mitra S**, Gumusgoz E, **Minassian BA**. [Lafora disease: Current biology and therapeutic approaches](#). *Rev Neurol (Paris)*. 2022 Apr;178(4):315-325. PMID:34301405
26. Mo A, Saffari A, Kellner M, Döbler-Neumann M, Jordan C, Srivastava S, Zhang B, Sahin M, Fink JK, Smith L, Posey JE, Alter KE, Toro C, Blackstone C, Soldatos AG, **Christie M**, Schüle R, Ebrahimi-Fakhari D. [Early-Onset and Severe Complex Hereditary Spastic Paraplegia Caused by De Novo Variants in SPAST](#). *Mov Disord*. 2022 Dec;37(12):2440-2446. PMID:36103453
27. Moreau C, Tremblay F, Wolking S, Girard A, Laprise C, Hamdan FF, Michaud JL, **Minassian BA**, Cossette P, Girard SL. [Assessment of burden and segregation profiles of CNVs in patients with epilepsy](#). *Ann Clin Transl Neurol*. 2022 Jul;9(7):1050-1058. PMID:35678011
28. **Nitschke F**, Montalbano AP. [Novel Cross-Correction-Enabled Gene Therapy for CDKL5-Deficiency Disorder](#). *Neurotherapeutics*. 2022 Oct;19(6):1878-1882. PMID:36266502
29. Nitschke S, Sullivan MA, **Mitra S**, Marchioni CR, Lee JPY, Smith BH, Ahonen S, Wu J, Chown EE, Wang P, Petković S, Zhao X, DiGiovanni LF, Perri AM, Israelian L, Grossman TR, Kordasiewicz H, Vilaplana F, Iwai K, **Nitschke F**, **Minassian BA**. [Glycogen synthase downregulation rescues the amylopectinosis of murine RBCK1 deficiency](#). *Brain*. 2022 Jul 29;145(7):2361-2377. PMID:35084461
30. Orsini A, Ferrari D, Riva A, Santangelo A, Macrì A, Freri E, Canafoglia L, D'Aniello A, Di Gennaro G, Massimetti G, Minetti C, Zara F, Michelucci R, Tumber A, Vincent A, **Minassian BA**, Striano P. [Ocular phenotype and electroretinogram abnormalities in Lafora disease and correlation with disease stage](#). *J Neurol*. 2022 Jul;269(7):3597-3604. PMID:35184210

31. Ozlu C, Chelliah P, Dahshi H, Horton D, Edgar VB, Messahel S, **Kayani S**. [ECHS1 deficiency and its biochemical and clinical phenotype](#). *Am J Med Genet A*. 2022 Oct;188(10):2908-2919. PMID:35856138
32. Pham T, Patel A, Muquith M, Zimmern V, **Goodspeed K**. [Abnormal Genetic Testing in Males With Concomitant Neurodevelopmental Disabilities and Genital Malformation](#). *Pediatr Neurol*. 2022 Sep;134():72-77. PMID:35841714
33. Rajasekaran K, Ma Q, Good LB, Kathote G, Jakkamsetti V, Liu P, Avila A, Primeaux S, Enciso Alva J, Markussen KH, Marin-Valencia I, **Sirsi D**, Hacker PMS, Gentry MS, Su J, Lu H, Pascual JM. [Metabolic modulation of synaptic failure and thalamocortical hypersynchronization with preserved consciousness in Glut1 deficiency](#). *Sci Transl Med*. 2022 Oct 5;14(665):eabn2956. PMID:36197967
34. Rowe AA, **Chen X**, Nettesheim ER, Issioui Y, Dong T, Hu Y, Messahel S, **Kayani SN**, **Gray SJ**, Wert KJ. [Long-term progression of retinal degeneration in a preclinical model of CLN7 Batten disease as a baseline for testing clinical therapeutics](#). *EBioMedicine*. 2022 Nov;85():104314. PMID:36374771
35. Strauss KA, Farrar MA, Muntoni F, Saito K, Mendell JR, Servais L, McMillan HJ, Finkel RS, Swoboda KJ, Kwon JM, Zaidman CM, Chiriboga CA, **Iannaccone ST**, Krueger JM, Parsons JA, Shieh PB, Kavanagh S, Wigderson M, Tauscher-Wisniewski S, McGill BE, Macek TA. [Onasemnogene abeparvovec for presymptomatic infants with three copies of SMN2 at risk for spinal muscular atrophy: the Phase III SPR1NT trial](#). *Nat Med*. 2022 Jul;28(7):1390-1397. PMID:35715567
36. Strauss KA, Farrar MA, Muntoni F, Saito K, Mendell JR, Servais L, McMillan HJ, Finkel RS, Swoboda KJ, Kwon JM, Zaidman CM, Chiriboga CA, **Iannaccone ST**, Krueger JM, Parsons JA, Shieh PB, Kavanagh S, Tauscher-Wisniewski S, McGill BE, Macek TA. [Onasemnogene abeparvovec for presymptomatic infants with two copies of SMN2 at risk for spinal muscular atrophy type 1: the Phase III SPR1NT trial](#). *Nat Med*. 2022 Jul;28(7):1381-1389. PMID:35715566
37. Szabo SM, Klimchak AC, Qian C, Iannaccone S, Popoff E, Gooch KL. [Characterizing the Occurrence of Key Clinical Milestones in Duchenne Muscular Dystrophy in the United States Using Real-World Data](#). *J Neuromuscul Dis*. 2022;9(6):689-699. PMID:36245384
38. Tardo L, **Wang C**, Rajaram V, Greenberg BM. [Pediatric paraneoplastic neuromyelitis optica spectrum disorder associated with ovarian teratoma](#). *Mult Scler*. 2022 Jan;28(1):160-163. PMID:34423664
39. Thompson K, Bianchi L, Rastelli F, Piron-Prunier F, Ayciriex S, Besmond C, Hubert L, Barth M, Barbosa IA, Deshpande C, Chitre M, Mehta SG, Wever EJM, Marcotelles P, Donkervoort S, Saade D, Bönnemann CG, Chao KR, Cai C, **Iannaccone ST**, Dean AF, McFarland R, Vaz FM, Delahodde A, Taylor RW, Rötig A. [Biallelic variants in TAMM41 are associated with low muscle cardiolipin levels, leading to neonatal mitochondrial disease](#). *HGG Adv*. 2022 Apr 14;3(2):100097. PMID:35321494
40. Tuncay IO, Parmalee NL, Khalil R, Kaur K, Kumar A, Jimale M, Howe JL, **Goodspeed K**, **Evans P**, Alzghoul L, Xing C, Scherer SW, Chahrour MH. [Analysis of recent shared ancestry in a familial cohort identifies coding and noncoding autism spectrum disorder variants](#). *NPJ Genom Med*. 2022 Feb 21;7(1):13. PMID:35190550

41. Wang L, Wang Y, Makhmoudova A, **Nitschke F**, Tetlow IJ, Emes MJ. [CRISPR-Cas9-mediated editing of starch branching enzymes results in altered starch structure in Brassica napus](#). *Plant Physiol*. 2022 Mar 28;188(4):1866-1886. PMID:34850950
42. **Waugh JL**, Hassan A, Kuster JK, Levenstein JM, Warfield SK, Makris N, Brüggemann N, Sharma N, Breiter HC, Blood AJ. [An MRI method for parcellating the human striatum into matrix and striosome compartments in vivo](#). *Neuroimage*. 2022 Feb 1;246():118714. PMID:34800665
43. Wu YW, Comstock BA, Gonzalez FF, Mayock DE, Goodman AM, Maitre NL, Chang T, Van Meurs KP, Lampland AL, Bendel-Stenzel E, Mathur AM, Wu TW, Riley D, Mietzsch U, Chalak L, Flibotte J, Weitkamp JH, Ahmad KA, Yanowitz TD, Baserga M, Poindexter BB, Rogers EE, Lowe JR, Kuban KCK, O'Shea TM, Wisnowski JL, McKinstry RC, Bluml S, Bonifacio S, Benninger KL, Rao R, Smyser CD, Sokol GM, Merhar S, Schreiber MD, Glass HC, Heagerty PJ, Juul SE, HEAL Consortium. [Trial of Erythropoietin for Hypoxic-Ischemic Encephalopathy in Newborns](#). *N Engl J Med*. 2022 Jul 14;387(2):148-159. PMID:35830641
44. Zimmern V, **Minassian B**, Korff C. [A Review of Targeted Therapies for Monogenic Epilepsy Syndromes](#). *Front Neurol*. 2022;13():829116. PMID:35250833

Book Chapter

1. Kern H, **Waugh J** (2022) [Movement Disorders](#). In: Rinaldi RJ, Srinivasan R (Eds.), *Handbook of Pediatric Rehabilitation Medicine* (pp. 75-84). New York, NY: Springer Publishing.
2. Wilkinson-Smith A, **Waugh JL** (2022) [Functional Movement Disorder in Children](#). In: LaFaver K, Maurer CW, Nicholson TR, Perez DL (Eds.), *Functional Movement Disorder: An Interdisciplinary Case-Based Approach* (pp. 183-196). Cham, Switzerland: Springer Publishing.