

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. Berge 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 (5) program, with two (2) years preliminary UT Southwestern Pediatrics residency, followed by three (3) years of Child Neurology residency
- “Categorical” Child Neurology Basic Neuroscience Research pathway – five-year (5) program, with one (1) year preliminary UT Southwestern Pediatrics residency, one (1) year of ABPN-approved Research, followed by three (3) years of Child Neurology residency
- “Reserved” Child Neurology pathway – three (3) years of Child Neurology residency after completion of two – three (2-3) years of ACGME-accredited Pediatrics residency training
- “Categorical” Neurodevelopmental Disabilities Residency Program – six-year (6) program, with two (2) years preliminary UT Southwestern Pediatrics residency, followed by four (4) years of Neurodevelopmental Disabilities residency
- Fellowship training in many pediatric neurology subspecialties, including Advanced Fellowship in Electroencephalography & 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 CME 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. Tonia Sabo, 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 (mTBI) 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 under the leadership of Dr. Tonia Sabo. Dr. Mathew Stokes, our third Pediatric Headache Medicine fellow, recently joined the Pediatric Headache faculty 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 30 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. Four faculty joined the Division in 2020.

Kaitlin Batley, M.D.

Assistant Professor, Pediatrics and Neurology



B.S., Interdisciplinary Studies, magna cum laude, 2011

M.D.

University of Miami – Miller School of Medical, Miami, FL, 2015

Postdoctoral Training

Residency, Pediatrics

UT Southwestern, 2015 – 2017

Fellowship, Child Neurology

UT Southwestern, 2017 – 2020

Fellowship, Pediatric Neuromuscular Medicine

UT Southwestern, 2020 – 2021

Interests:

Wilmot Bonnet MD

Assistant Professor, Pediatrics and Neurology



B.A., Biology University of Texas, Phi Beta Kappa, 2010

M.D.

University of Texas at Houston Medical School, 2014

Postdoctoral Training

Residency, Pediatrics

UT Southwestern, 2014 – 2016

Fellowship, Child Neurology

UT Southwestern, 2016 – 2019

Fellowship, Pediatric Vascular Neurology

UT Southwestern, 2019 – 2020

Interests: ischemic and hemorrhagic stroke in children, acute interventions for ischemic stroke and recovery in children with acquired brain injury

Honors / Awards

Best Pediatric Specialists, *D Magazine*

- Diana Castro
- Lauren Dengle Sanchez
- Alison Dolce
- Michael Dowling
- Kimberly Goodspeed
- Berge Minassian
- Dallas Mourot Armstrong
- Rana Said
- Mathew Stokes
- Jennifer Thomas
- Jeff Waugh

Texas Super Doctor, *Texas Monthly*

- Susan Iannaccone
- Jennifer Thomas (Rising Star)

Deepa Sirsi

- The Golden Boot Best Educator Award, Pediatric Neurology Division - UT Southwestern Medical Center

Invited Lectures

Diana Castro

- Sociedad Argentina de Neurología Infantil (SANI), Virtual, March 2021
 - “Atrofia Muscular Espinal”
- Muscular Dystrophy Association Annual Conference, Virtual, March 2021
 - “Navigating SMA Treatment Decisions”
- Medscape CME Live Activity, Virtual, March 2021
 - “Understanding Care Across the Age Spectrum in the Era of Effective Therapies”

Xin Chen

- CRSM, Virtual, February 2021
 - “Preclinical results in rodents strongly support clinical evaluation of AAV9 gene therapy for AGU and CLN7 patients”

- 7th DDX3X Foundation 2021 Scientific Conference, Clearwater, FL, 2021
 - Oral Presentation, *“Gene replacement therapy for DDX3X syndrome”*
- 5th Annual AP4-HSP Research Conference, Virtual, Boston, MA 2021
 - Oral Presentation, *“Preclinical Gene Therapy with AAV9/AP4M1 for SPG50 Disease”*
- 17th International Congress on NCL 2021. St. Louis, MO, 2021
 - Oral Presentation, *“Preclinical results in rodents strongly support clinical evaluation of scAAV9/MFSD8 as a potential gene therapy for CLN7 patients”*
- 16th International Symposium on MPS & Related Diseases, Virtual, Barcelona, Spain 2021
 - Oral Presentation, *“Preclinical gene therapy with scAAV9/AGA in aspartylglucosaminuria mice provides evidence for clinical translation”*

Alison Dolce

- UT Southwestern Medical Center Newborn News Educational Podcast, Dallas, TX, May 2021
 - *“Neonatal Seizures”*

Michael Dowling

- Lecture Series for Primary Care Physicians, Dallas, TX, January 2021
 - *“Neurologic Complications of Sickle Cell Disease”*
- Haltalin Conference, UT Southwestern, Dallas, TX, April 2021
 - *“Ethical Issues in the Care of Neurologically Devastated Children”*
- Grand Rounds, University of Oklahoma School of Medicine, Oklahoma City, OK, May 2021
 - *“Stroke in Children”*
- Fourth Annual Aspen Conference on Pediatric Stroke and Cerebrovascular Disease, University of Cincinnati, Cincinnati, OH, June 2021
 - *“Vertebral Artery Dissection: The Toddler with Tantrums, not Trauma”*
- International Pediatric Stroke Organization (IPSO) Conference, Virtual, June 2021
 - Symposium Panelist, *“Current Controversies in Stroke Prevention in Sickle Cell Disease”*

Patricia Evans

- U. S. Frontiers of Child Neurology, UT Southwestern School of Medicine, Dallas, TX, March 2021
 - *“My Child Matters Too. Addressing Inequities of Health Care Delivery to Children in the 21st Century”*

Kimberly Goodspeed

- Grand Rounds, UT Southwestern Medical Center, Dallas, TX, January 2021
 - *“Preparing for an Era of Precision Therapies in Neurodevelopmental Disorders”*
- UTSW O’Donnell Brain Institute Autism Symposium, Dallas, TX, March 2021
 - *“Clinical Trial Readiness in Neurodevelopmental Disabilities”*
- New Frontiers In Pediatric Neurology, UT Southwestern School of Medicine, Dallas, TX, April 2021
 - *“Understanding Autism Spectrum Disorders Through a Genetic Lens”*
- T32 Sleep and Circadian Rhythms Training Program Translational Seminary Series, UT Southwestern, Dallas, TX, January 2021
 - *“A cerebellar contribution to Fragile X Syndrome and autism-related social and sleep behaviors”*
- PlatformQ Health Education, Postgraduate Institute for Medicine, and National Organization for Rare Disorders, April 2021
 - *“Post-gene Therapy: Implications for Patients’ Management”*
- Simons Searchlight Family & Research Virtual Conference, Simons Foundation, July 2021
 - *“Preparing for Clinical Trials in Neurodevelopmental Disabilities”*
- STXBP1 Summit + Virtual Family Conference, STXBP1 Foundation, September 2021
 - *“Preparing for Clinical Trials in STXBP1 Encephalopathy”*

Steven Gray

- Lurie Children’s Hospital Neuroscience of Disease Series, Chicago, Virtual, September 2021
 - *“AAV9-mediated gene transfer as a platform approach for treating neurological disorders”*
- NIH/NINDS Clinical Neuroscience Grand Rounds, Bethesda, MD, Virtual, May 2021
 - *“AAV9-mediated gene transfer as a platform approach for treating neurological disorders”*
- International Child Neurology Teaching Network – All India Institute of Medical Science Neurogenomics Webinar Series, Virtual, April 2021
 - *“AAV-mediated gene therapy for neurological disorders”*
- Seaver Seminar Series – Icahn School of Medicine at Mt. Sinai, Mt. Sinai, NY, Virtual, March 2021
 - *“AAV-mediated gene therapy for neurological disorders”*

Michelle Machie

- Texas Health Dallas NICU Grand Rounds, Texas Health Presbyterian Hospital, Dallas, TX, April 2021
 - *“Crash Course in Neonatal Neuroimaging”*

Berge Minassian

- Grand Rounds, UT Southwestern Medical Center, Dallas, TX, February 2021
 - *“Lafora Disease – From Residency to Therapy”*

Eric Remster

- New Frontiers in Pediatric Neurology, Dallas, TX April 2021
 - *“The First Targeted Migraine Medications: CGRP Monoclonal Antibodies and Molecules”*

Rana Said

- Schwartz Rounds, Children’s Health, Dallas, TX February 2021
 - Invited Panelist, *“Overcoming fears, building hope: Reflections on the COVID-19 Vaccine”*
- Multidisciplinary Case Conference, Department of Pediatrics, UT Southwestern Medical Center, Dallas, TX September 2021
 - Invited Speaker, *“Rasmussen’s Rashes and Ropa – A Complex Case Intertwining Neurologic, Allergic and Hematologic Disease”*

Sarah Sinnett

- Taysha Gene Therapies, Dallas, Virtual, December 2021
 - *“Unveiling the full potential of RTT therapies”*
- Regis University, Denver, Virtual, November 2021
 - *“Feedback-enabled gene therapy for Rett syndrome”*

Jennifer Thomas

- New Frontiers in Pediatric Neurology, UT Southwestern Medical Center, Dallas, TX, April 2021
 - *“Neonatal Encephalopathy: Diagnosis, Management and Outcome”*

Conference Presentations

American Academy of Neurology Conference, Virtual, April 2021

Khan T, Dolce A, Goodspeed K

Poster Presentation, *“A case off Bainbridge-Ropers syndrome with breath holding spells: challenges in diagnosis and management”*

Said R

Panelist, *"How to Address Patient Bias Against Neurologists of Diverse Backgrounds"*

Moderator, *"Clinical Educator Career Path Panel Discussion"*

Invited Speaker, Medical Student Symposium - *"Innovations in Neurology"*

Course Director and Invited Speaker, *"The (Neuro) Anatomy of an Impactful and effective Program Director"*

Course Director and Moderator, Medical Student Symposium - *"NeuroEquity"*

Invited Course Director and Speaker, *"Child Neurology: A Case-based Approach"*

Lorenzo J, Dolce A, Lowden A

Poster Presentation, *"Electroclinical Features in MECP2 Duplication Syndrome: Pediatric Case Series"*

Gray SJ

Gene Therapy for Neurological Diseases, Boston, MA, December 2021

Invited speaker, *"Intra-CSF administration of AAV9. Promise, caveats, and unanswered questions"*

Neurodevelopmental Drug Development Summit, Virtual, December 2021

Invited speaker and panelist, *"Limitations and future prospects for AAV-mediated MeCP2 gene transfer for the treatment of Rett syndrome"*

International Congress of Inborn Errors of Metabolism 2021, Sydney, Australia, November 2021

KEYNOTE: *"CNS Targeted Gene Therapy: Update on Gene Therapy for Lysosomal Diseases"*

Société française pour l'étude des erreurs innées du métabolisme (CETL & SFEIM), France (Virtual), June 2021

KEYNOTE: *"Update on gene therapy for lysosomal diseases"*

United Mitochondrial Disease Foundation Annual meeting, (Virtual), June 2021

Invited speaker: *"Gene therapy for SURF1-related Leigh syndrome"*

American Academy of Pediatrics National Conference, Virtual, October 2021

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

Poster Presentation, *"Are There Differences in Concussion-Related Characteristics and Return-to-Play in Soccer Positions?"*

Althoff C, Worrall H, Miller S, Chung J, **Stokes M**, Cullum CM, Jones J

Poster Presentation, *"Are There Differences in Reported Symptoms and Outcomes Between Pediatric Patients With and Without Obsessive Compulsive Disorder After a Concussion?"*

American Epilepsy Society Conference, Chicago, IL, December 2021

Choudhari, P, **Lowden A, Dolce A**

"Electrographic Features and Predictive Value of EEG in Children with Complex Febrile Seizures"

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

"Efficacy and Safety of Lacosamide in Infants"

Sirsi D, Said R

Abstract Presentation, *"A Novel EEG Curriculum for Child Neurology Residents Using a Flipped Learning Method."*

Child Neurology Society Annual Meeting, Boston, MA, September 2021

Evans P

Poster Presentation, *“Autism Communication Enhancement (AE): Skill Training for Pediatric Neurologist during Telehealth Visits with Communication Impaired Children during the COVID Pandemic.”*

Cooper D, Dooley K, **Goodspeed K**

Poster Presentation, *“A comparison of the adaptive and autistic behavior in three developmental epileptic encephalopathies – SLC6A1, SCN2A, STXBP1”*

O’Donnell Brain Institute Autism Symposium, UT Southwestern, Dallas, TX, March 2021

Bone ME, **Goodspeed KD**

Poster Presentation, *“A zoo based social skills intervention for preschoolers with autism”*

Bone ME, **Goodspeed KD**

Poster Presentation, *“A case report of topiramate for severe breath-holding spells in a patient with Pitt-Hopkins Syndrome”*

Neonatal Brain Matters Conference

Chalak L, **Thomas J**

Conference Host

Texas Academy of Physician Assistants (TAPA) Annual Meeting, Virtual, February 2021

Dolce A

“Evaluation & Treatment of Seizure in Children”

Said R

“Use of Cannabidiol for Treatment of Refractory Epilepsy in Children”

Sirsi D

“Practitioners guide to First and second seizure in otherwise healthy children”

WORLDSymposium, Virtual, February 2021

Chen X

Oral Presentation *“Preclinical results in rodents strongly support clinical evaluation of scAAV9/MFSD8 as a potential gene therapy for CLN7 patients”*

LaBounty Phillips K, McGinnis E, Seda J, **Goodspeed K**, Gerhart M, Prasad S

Poster Presentation, *“Co-creating a gene therapy clinical trial with GM2 gangliosidosis caregivers: a virtual approach to patient engagement”*

Other Conferences

Balagura G, Kaufman M, 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, Coppola A, Dilena R, Faggioli R, Ferrari A, Fiorini E, Madia F, Gennaro E, Giordano L, Iacomino M, Marini C, Mancardi MM, Mastrangelo M, Messina T, Minetti C, Nobili L, Pisano T, Russo A, Salpietro V, Savasta S, Scala M, Spalice R, Trivisano M, Tzadok M, Valeriani M, Vari MS, Vigeveno F, Vignoli A, Viri M, Zara F, Helbig I, Striano P

International Epilepsy Congress, International League Against Epilepsy (ILAE), Virtual, September 2021
Abstract Presentation, *"Developmental trajectories in STXBP1-DEE"*

Batley K, Castro D

Carrell Krusen Neuromuscular Symposium, Dallas, TX, February 2021
Oral Presentation, *"Four sisters with progressive weakness"*

Bonnet W

International Stroke Conference 2021, Virtual, March 2021
Poster Presentation, *"Incidence and Clinical Characteristics of Large Vessel Occlusion in Pediatric Stroke"*

Bunt SC, Presley C, Tarkenton T, Wilmoth K, Didehbani N, **Stokes M**, Miller SM, Bell K, Batjer H, Cullum CM
Sports Neuropsychology Symposium, Virtual, 2021

Poster Presentation, *"Concussion Symptom Reporting In Sport-Related Concussions During The COVID-19 Pandemic"*

Chen X

American Society of Gene and Cell Therapy, Virtual, May 2021
Poster Presentation, *"Novel AAV Capsids for Enhanced Gene Transfer to the Cerebellum, Spinal Cord, and Schwann Cells"*

Daesco V, Horton D, Goodspeed K

American Neurological Association Annual Meeting, Virtual, October 2021
Presentation, *"Characterizing the CDG-SRD5A3 Clinical Spectrum"*

Gray SJ

NIH Workgroup on gene-targeted therapies (early diagnosis and delivery), Virtual, June 2021
Panelist and speaker, *"Workgroup 1, the who, what, and when"*

NIH/NIMH Virtual gene therapy workshop, Virtual, January 2021

Panelist and speaker, *"Limitations and future prospects for AAV-mediated MeCP2 gene transfer for the treatment of Rett syndrome"*

Heinzelmann M, **Stokes M**, Bunt S, Didehbani N, Miller S, Hynan L, Cullum CM

American Academy of Neurology Sports Concussion Conference, Virtual, July 2021

Poster Presentation, *"Impact of Playing Surface on Concussion Symptoms in Young American Football Players"*

Lowden A

Child Neurology Society and International Child Neurology Association Annual Meeting, Virtual, October 2021
Poster Presentation, *"Autism Communication Enhancement (AE): Skill Training for Pediatric Neurologist during Telehealth Visits with Communication Impaired Children during the COVID Pandemic"*

Machie M

Hershey Conference on Developmental Brain Injury, June 2021

Poster Presentation, *"Reliability of the Kidokoro MRI Score in an Ethnically and Socioeconomically Diverse Very Preterm Infant Population"*

Mehta A, **Sirsi D**, Veltkamp D, Pfeifer C

121st ARRS Virtual Annual Meeting, April 2021

Poster Presentation, *“Stereoencephalography as an Emerging Adjunct to Seizure Localization in Pediatric Epilepsy”*

Muquith M, Pham T, Patel A, **Goodspeed K**

American Medical Association Annual Meeting (AMA Research Challenge 2021), October 2021, Poster Presentation

Poster Presentation, *“Prevalence of Diagnostic Genetic Testing Among Male Patients with Concomitant Neurodevelopmental Disabilities and Genital Abnormalities”*

Nitschke F

Biomarker Symposium of the Adult Polyglucosan Body Disease Research Foundation, Virtual, November 2021

Invited Speaker, *“CSF α -Glucans and Glycogen Synthase - Potential for biomarker and target engagement in APBD”*

Sanchez L

Children’s Tumor Foundation NF Virtual Case Conference, October 2021

Oral Presentation, *“Pediatric Vasculopathy in NF1”*

Waugh J

Annual Pediatric Neurology Symposium, May 2021, Memphis, TN

Invited Lecture, *“How to Recognize Abnormal Movements in Children”*

Invited Lecture, *“The Brain Circuits Behind Dystonia”*

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

Society of Clinical Research Associates (SOCRA) Annual Conference, Virtual, September 2021

Poster Presentation, *“Differences in Concussion-Related Characteristics and Return-to-Play in Soccer Positions”*

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 of 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/Site Directors for each of these programs. In her capacity as Program Director of the Child Neurology Residency Program (core program) for the past 17.5 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 (3) training program, including a combined five-year (5) “Categorical” program with two (2) years of ACGME-accredited Pediatrics training and three (3) 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 (5) program, with one (1) year preliminary UT Southwestern Pediatrics residency, one (1) year of ABPN-approved Research, followed by three (3) years of Child Neurology residency), and for appropriate residency candidates, the Physician Scientist Training Program in Pediatrics (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 (6) “Categorical” program with two (2) years of ACGME-accredited Pediatrics training and four (4) 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. Susan Arnold was the Director of the Pediatric Clinical Neurophysiology fellowship program until June 2021. Dr. Alison Dolce became the Director of the Pediatric Clinical Neurophysiology fellowship program beginning in June 2021. Graduates are eligible for the American Board of Psychiatry and Neurology in Clinical Neurophysiology. This ACGME-accredited program accepts one (1) fellow per year.

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 (2) fellows per year. Graduates are board-eligible in Epilepsy. Dr. Deepa Sirsi is also the Director of TMB accredited Advanced EEG and Epilepsy Fellowship program.

Dr. Jennifer Thomas is the Director of the Fetal and Neonatal Neurology fellowship program. This is a Texas Medical Board (TMB)-approved program, which accepts one (1) fellow per year, who has completed prior training in either Child Neurology or Neonatal-Perinatal Medicine.

Dr. Tonia Sabo is the Director of the Pediatric Headache fellowship program. This program is TMB-approved and a United Council for Neurologic Subspecialties (UCNS)-accredited program. This program accepts one (1) 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.

The Neuromuscular Medicine program is an ACGME-accredited program that accepts one (1) 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.

Dr. Mathew Stokes is the Director of the Pediatric Headache fellowship program. This program is TMB-approved and a United Council for Neurologic Subspecialties (UCNS)-accredited program. This program accepts one (1) 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.

The Neuromuscular Medicine program is an ACGME-accredited program that accepts one (1) 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.

Dr. Michael Dowling is the Associate Program Director for the Vascular Neurology program. This is an ACGME-accredited program that accepts one (1) Pediatric track fellow per year. Graduates are eligible for the American Board of Psychiatry and Neurology in Vascular Neurology.

Fellowship Programs in Development:

- Pediatric Movement Disorders. This is a TMB-approved program, which will accept one (1) Pediatric subspecialty fellow per year. Dr. Waugh will be the Site Director for the Pediatric subspecialty fellow.
- Neurodevelopmental Disabilities. This will be a one-year (1) ACGME-accredited fellowship program for fellows who completed prior training in Child Neurology. Graduates will be eligible for the American Board of Psychiatry and Neurology in Neurodevelopmental Disabilities, upon passing of the American Board of Pediatrics and American Board of Psychiatry and Neurology with Specialty Qualifications in Child Neurology.

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.
- Susan Arnold is involved in multiple industry-sponsored clinical research trials. In 2017, she was the site-principal investigator for three epilepsy clinical trials sponsored by UCB Pharma. She was also site principal investigator for a multi-center study of the pharmacokinetics of antiepileptic drugs in obese children funded by the National Institute of Child Health and Human Development. She has worked with Drew Thodeson on an evaluation of the utility of next generation gene sequencing in the evaluation and management of childhood epilepsy.
- 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 designation in 2021 from the International Rett Syndrome Foundation (IRSF) and will begin an on-going patient registry study in 2022, for which the clinic received a grant in 2021. She works with Dr. Hoang Nguyen in cardiology on projects related to cardiac electrophysiology in girls and women with classic Rett syndrome.
- Diana Castro is the co-director of the Pediatric Muscular Dystrophy Association (MDA) Clinic, co-director of the Parent Project Muscular Dystrophy Clinic, director of the Cure SMA care center network as well as the site co-director for the neuromuscular fellowship at Children's Health. Since completing training in pediatric neuromuscular medicine, Dr. Castro has conducted multiple clinical research trials, as PI or Co-PI in Spinal Muscular Atrophy (SMA), Duchenne Muscular Dystrophy (DMD) and Charcot-Marie-Tooth (CMT). The pediatric neuromuscular research group is very active; they currently run over 14 clinical trials, including natural history and treatment-related studies. Other research interest includes myasthenia gravis and acquired neuropathies. She serves as a medical advisor for the Myasthenia Gravis Foundation and the GBS/CIDP Foundation.
- Xin Chen is the investigator of three new research projects (DDX3X, GNAO1, and CMT4A) funded by Taysha Gene Therapy. He is also the co-investigator of multiple research projects funded by private foundations, industry, and NIH to Dr. Steven Gray. Dr. Chen's main research areas are Aspartylglucosaminuria (AGU), CLN7 Batten disease, SPG50 disease, SLC52A2 deficiency disease, and SRD5A3 deficiency disease. In 2021, Investigational New Drug application #19766 to initiate a Phase I intrathecal gene transfer trial for AAV9/*MFSD8* was approved by the US FDA and the trial is enrolling CLN7 patients at Children's Health in Dallas, TX in collaboration with UTSW Medical Center (clinicaltrials.gov NCT04737460). Dr. Chen is currently working to facilitate the initiation of gene therapy clinical trials for AGU and SPG50 gene therapies. He is also helping in developing novel 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 disorders of Autism Spectrum Disorders as well as Fragile X Syndrome. She participates in a range of studies, including the genetics of Autism Spectrum Disorders, and mechanisms of Fragile X Syndrome cognitive deficits.
- Kimberly Goodspeed is involved in clinical research of rare neurogenetic and neurodevelopmental disorders and directs specialty clinics for Pitt-Hopkins Syndrome and SLC6A1-Related Disorder. She is also heavily involved in the development of the UTSW Gene Therapy program, alongside Dr. Minassian and other faculty colleagues. She is the principal investigator for several gene therapy disease programs including clinical trial readiness studies for aspartylglucosaminuria, SLC6A1 Related Disorder, SLC13A5 Deficiency Disorder, and SRD5A3-CDG. Additionally, she is working with Dr. Sanchez on a retrospective natural history study for mucopolysaccharidosis type 3C and 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. 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. She was also awarded the Dedman Family Scholar in Clinical Care to establish a deep phenotyping registry for children with neurodevelopmental disorders. She earned her graduate certificate program in Clinical Sciences through the UTSW Center for Translation Medicine and is completing her Master of Science in Clinical Sciences in the spring of 2022.
- 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 CNS gene transfer, NIH funding to test a gene therapy treatment for Niemann-Pick Type C disease, an NIH R01 subcontract to develop a gene therapy approach for Krabbe disease, grants from the Cure SPG50 foundation, the Sappani Foundation, the Drake Rayden Foundation, and Hannah's Hope Fund, a sponsored research agreement from Roche, and multiple sponsored research agreements from Taysha Gene Therapies. His funded research projects include the development of gene therapy treatments for CLN7 Batten disease, Multiple Sulfatase Deficiency, Rett syndrome, Angelman Syndrome, SLC6A1 disease, SPG50 disease, SLC52A2 disease, GNAO1, DDX3X, Charcot-Marie Tooth disease type 4A, Fragile X, SRD5A3, Non-Ketonic Hyperglycinemia, and Leigh Syndrome. Dr. Gray is created a core facility for research and clinical AAV manufacture, and also facilitate the initiation of gene therapy clinical trials for multiple rare neurological diseases including CLN7 Batten disease at UTSW and the following disorders elsewhere: Giant Axonal Neuropathy, CLN1 Batten disease, CLN5 Batten disease, Spastic Paraplegia type 50, and GM2 gangliosidosis (Tay-Sachs and Sandhoff diseases).
- Sharmistha Mitra is the co-investigator of multiple basic science research projects funded by private foundations, industry, and NIH to Dr. Berge Minassian and to Dr. Mitra. Dr. Mitra focuses much of her research in 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, role of E3-ubiquitin ligases in Polyglucosan body diseases and targeted molecular therapy for certain pediatric neuromuscular diseases. She supervises and mentor senior research scientists and multiple technicians in Dr. Minassian's laboratory.
- Susan Iannaccone has expanded the Pediatric Neuromuscular Clinical Trials Program (Pediatric NMCTP) that she started in 2005. She and the NM team are running 12 clinical trials for our patients with rare neuromuscular diseases including gene replacement therapy for Spinal Muscular Atrophy. Funding for studies in spinal muscular atrophy and Duchenne muscular dystrophy is largely through industry contracts, making it possible for us to offer our patients access to state of the art and cutting-edge research. She is Associate Director of the Wellstone Muscular Dystrophy Center at UTSW for which the focus is gene editing for Duchenne muscular dystrophy.
- Saima Kayani has been actively involved in development of clinical aspects of gene therapy trials at UT Southwestern under my supervision and serves as PI and Co-I for NIH and industry sponsored trials. These trials are ongoing at UT Southwestern/Children's Medical Center (Dallas) and others are upcoming sponsored programs. Saima Kayani has carried out various research responsibilities including handling and writing informed consent documents, writing clinical protocols for institutional review board, 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. Saima Kayani has also been actively involved in development of clinical section of IND for first in human Phase I intra thecal 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 intra thecal gene therapy in CLN7.

As the co-investigator for CLN7 Phase 1 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, 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 start of a prospective natural history trial for rare brain disease SURF-1 related Leigh Syndrome (Starting early 2022).

- Tonia Sabo is involved in clinical industry related studies in Pediatric Headache. She is involved in faculty led research in Intracranial Hypertension. She is involved in Quality Initiative Research on the treatment of headache, and she is the Children's Medical Center Site Principal Investigator for the UTSW North Texas Concussion Registry (ConTex), which is a multicenter, prospective longitudinal registry for concussion established in 2015 that has enrolled over 900 patients to date. Dr. Sabo has been studying cerebrovascular recovery in concussed collegiate athletes. Currently, a new clinical trial focusing on treatment of pediatric migraine with triptan therapy started enrollment.
- 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 the UTSW North Texas Concussion Registry (ConTex), which is a multicenter, prospective longitudinal registry for concussion established in 2015. Additionally, he is a co-investigator on a multicenter study, CARE4KIDS, looking at objective biomarkers in concussive head injury.
- Rana Said is involved in multiple industry-sponsored clinical research trials. In addition, she is currently the Co-Principal Investigator 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 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. We will also be assessing biomarkers of efficacy by screening for 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 principal investigator in a novel treatment for refractory status epilepticus, Dr. Said is also 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 (NORSE), and infantile spasms.
- Sarah Sinnett is the Principal Investigator of several projects funded by industry. She has also been funded by 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 and two of these inventions have been licensed to industry. Her recent first-author publication in *Brain* has an attention score ranking it within the top 5% of nearly 20 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.
- Deepa Sirsi is involved in a range of clinical research studies concerning EEG: 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, SLC6A1 syndrome. She is the site PI for industry funded anti-seizure medication trials. She collaborates with other clinicians and basic scientists in research involving treatment of genetic causes of epilepsy and neurodevelopmental impairments such as GLUT1 disorder.
- Jennifer Thomas is involved in clinical research in the area of neonatal neurology. She is currently site co-principal investigator in a multi-center trial designed to study the efficacy of high dose erythropoietin for neuroprotection in term infants with hypoxic-ischemic encephalopathy, known as the HEAL trial.
- Alison Dolce is involved in a range of clinical research studies including those in the area 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 and Tuberous Sclerosis. Peter Tsai is the PI on numerous federally funded pre-clinical and translational research projects. These include NIH RO1 and DOD grants to delineate circuit mechanisms underlying autism-relevant behaviors in Tuberous Sclerosis, an NIH RO1 grant to explore the circuits contributing to abnormal behaviors in Fragile X Syndrome, and a DOD funded

grant examining brain mechanism and the potential benefits of non-invasive brain stimulation for children with autism. Dr. Tsai 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 studying biomarkers in demyelinating and other immune-mediated brain conditions such as multiple sclerosis, neuromyelitis optica spectrum disorder, and autoimmune encephalitis. She is the PI on a study of longitudinal and neuropsychological outcomes of patients with acute disseminated encephalomyelitis (APERTURE). She has published case reports, case series, and original research in the fields 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, PH. He has current grant funding from the American Academy of Neurology and the Collaborative Center for X-linked Dystonia Parkinsonism.
- Felix Nitschke is a Principal Investigator investigating metabolic pathways involved in several neurodegenerative diseases, such as Lafora disease and Adult polyglucosan body disease. His work currently focusses on glycogen metabolism and the treatment of associated neurological diseases with novel gene therapy approaches. He recently received funding from the Orphan Disease Center at University of Pennsylvania.

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 and adolescents who have autistic or severe developmental disabilities. CADD provides the integration of psychiatry, psychology, and neurodevelopmental disabilities specialists, as well as behavioral therapies. Dr.

Patricia Evans serves as one of three clinical co-directors for the center. Dr. Kimberly Goodspeed directs three rare neurodevelopmental disorders clinics, including that for Phelan McDermid Syndrome, Pitt Hopkins Syndrome, and SLC6A1 Related Disorders, and participates in clinical research for these rare syndromes, and participates in clinical trials for these rare syndromes. The center also houses a clinic for rare genetic variant Fragile X Syndrome.

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

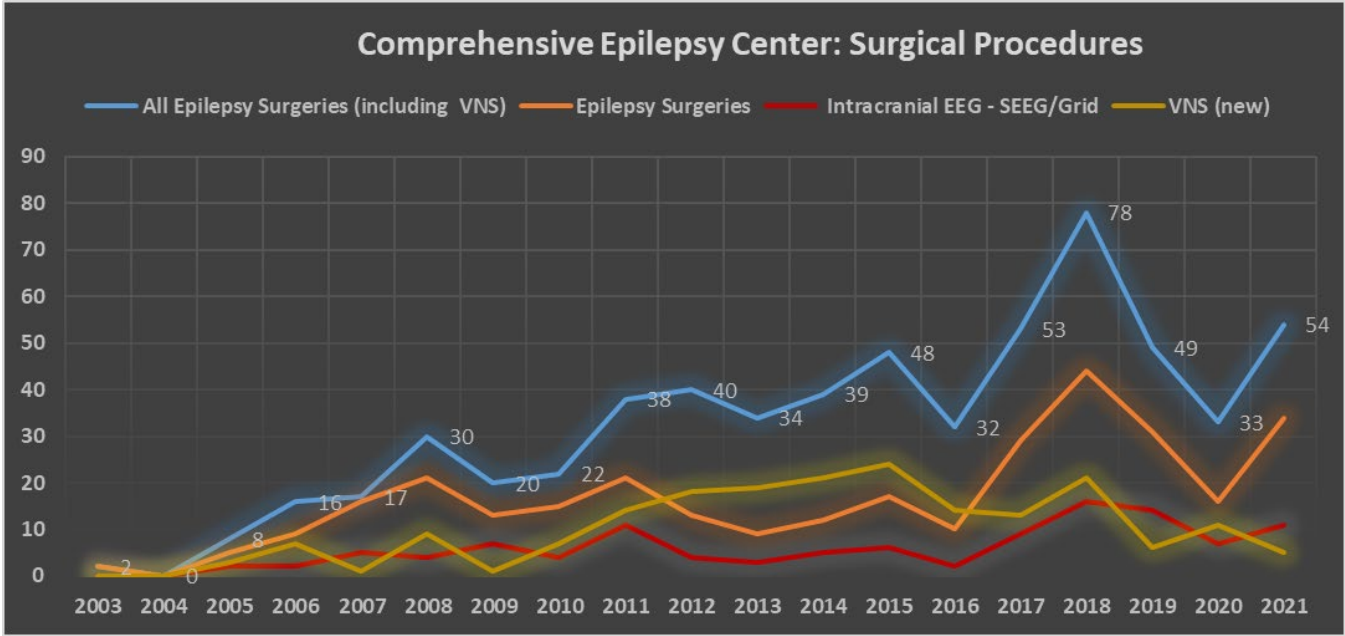
The Epilepsy Center currently sees outpatients at three locations. The majority of patients are seen in the CHST clinic in downtown Dallas, with a smaller group seen at the CHST clinic at Texas Heath Presbyterian Hospital. The UTSW Pediatric Neurology and Epilepsy Clinic opened in Frisco, and began seeing patients in 2020, with increasing patient volume in 2021 as it continues to work on expanding its referral base to increase new patient referrals. Patient demand remained high in 2021 with 1320 new patient referrals to the CHST Epilepsy Center. Epilepsy Center physicians and advanced practice providers completed 7320 outpatient visits in 2021 (22% increase from 2020). In addition, we continued to respond to the fluctuating needs and restrictions related to COVID pandemic by utilizing telemedicine for our patients. In 2021, Dallas Epilepsy clinic completed 2,125 telemedicine visits. This was an unprecedented surge going from no telemedicine in 2019 to a well-established, quality measured system for our patients to access care safely.

The previous epilepsy center director Dr. Susan Arnold retired in June 2021 and Dr. Deepa Sirsi is the current medical director of the epilepsy center since July 2021. A senior epilepsy physician from Denver Children’s Hospital was hired to join the group and will start in March 2022 and this will ensure continued access for new patient appointments, along with maintaining and supporting the expanding need for EEG services and will also focus on expanding epilepsy research and clinical trials.

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. In 2020 the program began performing pediatric MEG studies on the UTSW North Campus, which has enhanced our ability to assess patients and offer epilepsy surgery to the most complex epilepsy patients, in line with other large pediatric epilepsy centers. There were 26 pediatric MEG studies performed and interpreted by pediatric epileptologists in 2020 and this increased to 30 in 2021. It is anticipated that this volume will continue to increase in 2022 as MEG is increasingly used as an additional and valuable tool in epilepsy surgery. On the Children’s Medical Center Dallas campus, the center performed the first responsive neurostimulation device placement in August 2021 and this volume is anticipated to increase in 2022. It is also anticipated that deep brain stimulation for epilepsy will be employed as a treatment option for children with intractable epilepsy in 2022. The epilepsy surgery program is also a pioneer in the use of laser interstitial thermoablation therapy, a minimally invasive surgical approach which reduces surgical complications and allows for much faster recovery time. Seizure-free outcomes for surgery at CMC Dallas equal or exceed those in pediatric series published in peer-reviewed literature.

The surgery program has shown steady growth over time and went through a dramatic expansion in late 2017 with the addition of robot assisted stereotactic surgical procedures. With this increase, there has been a significant increased demand for physician time devoted to surgical procedures, and this continues to be an area of challenge for the program. The surgical numbers were lower in 2020 compared to prior years due to the COVID pandemic and limitation from this for elective procedures. However, in 2021 the surgical numbers more than doubled from 2020 and reached and surpassed 2019 numbers. Approximately 45% of the Center’s surgical patients originate as consultations from the Children’s Health inpatient or ER services, 32% come from referrals for second opinions, and the remainder from our new-onset seizure and epilepsy clinic.

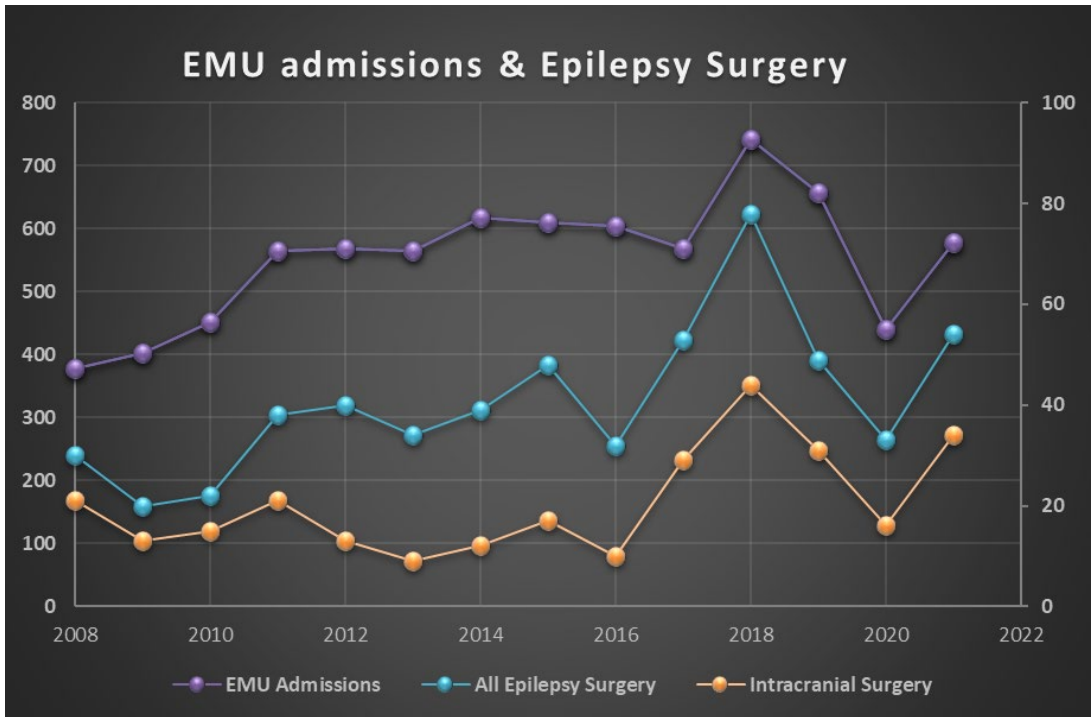
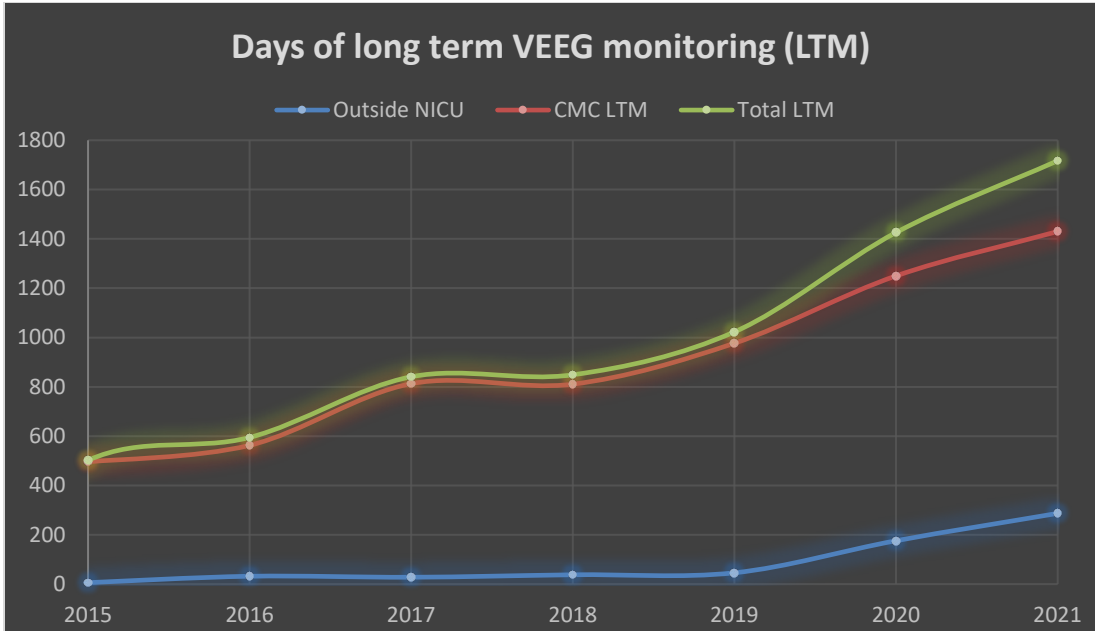


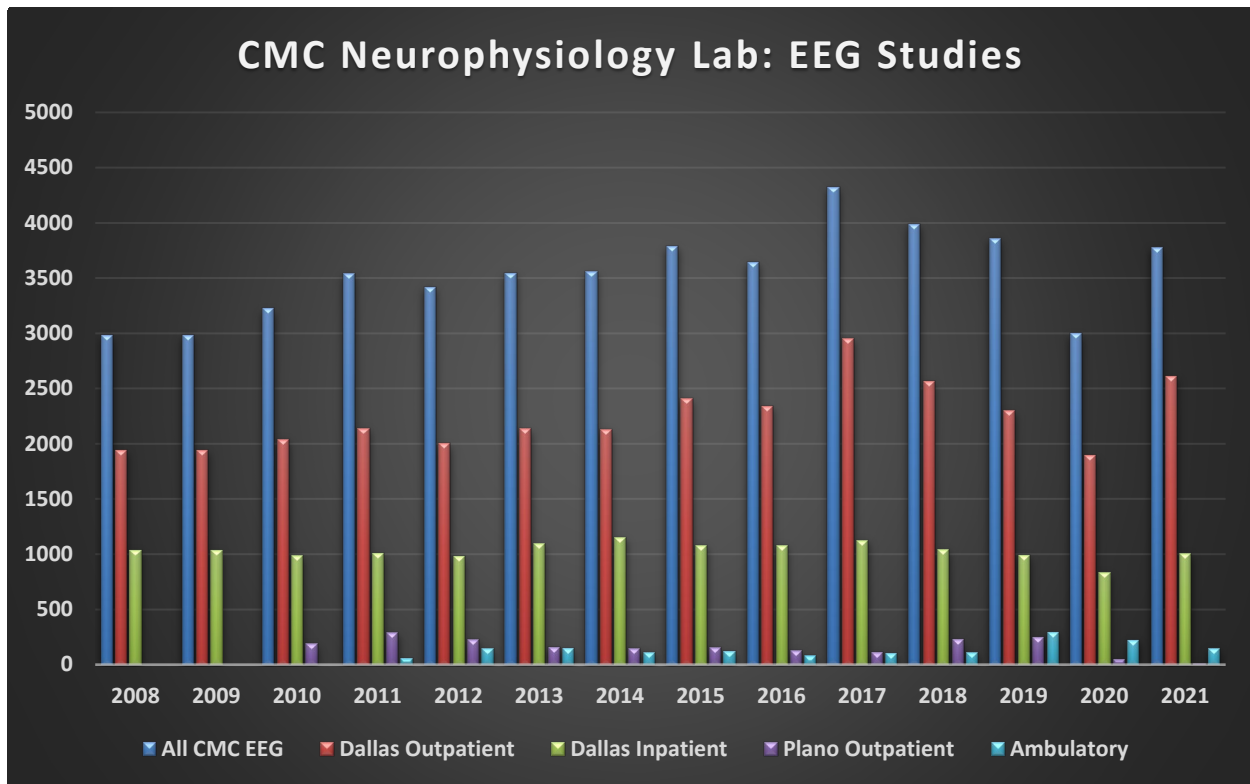
Neurophysiology services

Neurophysiology services have remained stable, with a rebounding of routine EEGs and EMU studies compared to 2020 when numbers decreased due to COVID. 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, and represents a challenge to the Epilepsy Center physicians due to the time, labour intensive nature of interpreting these studies and high acuity of these patients. Active recruitment efforts are ongoing to hire additional faculty members in 2022 to help meet this need.

Outpatient ambulatory studies remained steady. The center also provides EEG interpretation for Parkland NICU since 2020 and continues to provide NICU EEG interpretation services at Texas Health Presbyterian Hospital Dallas and Clements University Hospital. In 2022 epilepsy physicians will start providing NICU EEG interpretation services for Texas Health Presbyterian Hospital Frisco.

The CMC Dallas Neurophysiology Lab maintained accreditation by the American Board of Registration of Electroencephalographic and Evoked Potential Technologists (ABRET), and is one of the few accredited pediatric neurophysiology laboratories in the region.





Dallas Inpatient includes routine 1–2-hour EEGs, EMU & Prolonged LTM studies are included in All CMC EEG

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 2020, the program joined a multicenter collaborative initiative, the Epilepsy Health Learning System (ELHS) and is taking its place as a national leader for quality improvement in outpatient pediatric epilepsy care. The quality measures supporting this distinguished status are:

- Appropriate diagnostic evaluation with EEG
- Epilepsy safety counseling (seizure precautions and first aid)
- Ease of access (speed of scheduling ER referrals)
- Patient and Family Understanding of the Diagnosis
- Screening of teenage patients for depression
- Documenting and discussing seizure types, seizure frequency during each visit
- Measuring quality of life at each patient visit
- Assessing medication adherence at each patient visit
- Adoption of telemedicine and use of MyChart to communicate effectively with patients and families.

Center faculty maintain active involvement in national and international Epilepsy initiatives. Dr Arnold, the prior medical director until June 2021, is a past board member of the National Association of Epilepsy Centers and chaired their accreditation committee.

In June 2021 Children’s Health and UTSW Medical Center received the designation of Rett Center of Excellence with Dr. Dallas Armstrong as the Medical Director.

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 staff also provides educational programming for both children and parents through the camp programs. 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.

Support groups and Transition education:

Teen and Spanish Support groups continued to meet in 2021, predominantly with virtual support groups zoom in 2021. Our bi-monthly transition group for teens and parents began in 2017 in collaboration with the Epilepsy Foundation of Texas and focusses on preparing families for the transition to adulthood.

Family Conferences and Community outreach:

The Epilepsy Center newsletter is an important tool for epilepsy education in the outpatient clinic. Several 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.



Dr. Lauren Sanchez

Research and Education

The Comprehensive Epilepsy Center continues to participate in clinical research with the faculty and fellows all involved in IRB approved clinical research studies. In 2021 there were two active clinical trials for new anti-seizure medications, and a study of triheptanoin oil for Glut-1 deficiency. The center is also a participating site for several natural history studies for genetic epilepsies. Five faculty members presented research at national meetings in 2021, and many epilepsy faculty spoke at regional symposiums. Six epilepsy faculty authored or co-authored a total of 10 papers in peer reviewed journals in 2021.

The Epilepsy Center physicians continues to have an international presence with Dr. Andrea Lowden participating in Global Health initiatives and giving lectures about pediatric epilepsy and EEG in Cuba and the Dominican Republic in 2019. In 2020 - 2021 these activities were on hiatus due to COVID pandemic and travel restrictions.

The ACGME Pediatric Epilepsy and Neurophysiology Fellowship programs continue to attract talented applicants and the majority of fellows go on to university faculty positions. In addition, a third track for fellowship training was added in 2021, the Advanced EEG and Epilepsy research fellowship.

Current Grant/Contract Support

Dallas Armstrong

Grantor: International Rett Syndrome Foundation

Title of Project: Rett Center of Excellence Patient Registry

Role: Principal Investigator

Dates: 07/2021 – 06/2023

Diana Castro

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: Biogen

Title of Project: Study of Nusinersen (BIIB058) in Participants With Spinal Muscular Atrophy (DEVOTE)

Role: Site Principal Investigator

Dates: 2020 – Present

Contractor: Neurogene

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

Role: Site Principal Investigator

Dates: 2019 – Present

Contractor: Cure SMA

Title of Project: Cure SMA Clinical Data Registry

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: Fibrogen

Title of Project: A phase 3 Trial Pamrevlumab or Placebo in Combination with Systemin Corticosteroids, in Subject with Non-Ambulatory Duchenne Muscular Dystrophy (DMD)

Role: Site Principal Investigator

Dates: 2021 - Present

Contractor: Fibrogen

Title of Project: Trial of Pamrevlumab (FG-3019), in Non-Ambulatory Subjects with Duchenne Muscular Dystrophy (DMD)

Role: Site Principal Investigator

Dates: 2016 - 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

Contractor: Biogen/ IONIS Pharmaceutical

Title of Project: A Phase 3 Study to Assess the Efficacy and Safety of ISIS-SMN Rx (CS11) in Infants and Patients with Later-onset Spinal Muscular Atrophy

Role: Site Principal Investigator

Dates: 2015 – Present

Contractor: PTC Pharmaceutical

Title of Project: An open label study for previously treated Ataluren (PTC124) patients with nonsense mutation dystrophinopathy

Role: Site Co-Investigator

Dates: 2016 – Present

Xin Chen

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: CURESPG50

Title of Project: SPG50 Gene Therapy

Role: Co-Investigator

Dates: 02/2020 – 07/30/2022

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

Lauren Dengle Sanchez

Contractor: Phoenix Nest

Title of Project: A Retrospective Natural History study of Mucopolysaccharidosis type IIIC

Role: Principal Investigator

Dates: 11/2020 – Present

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: Site Principal Investigator
Dates: 09/2017 – 06/2022

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: Site 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: Site Principal Investigator
Dates: 09/2020 – 12/2021

Grantor: National Fragile X Foundation
Title of Project: Deciphering mechanisms, marker
Role: Site Principal Investigator
Dates: 09/2010 - 12/2021

Kimberly Goodspeed

Grantor: SLC6A1 CONNECT
Title of Project: A Natural History and Outcome Measure Discovery Study of Developmental and Epileptic Encephalopathies
Role: Site Principal Investigator
Dates: 07/2020 – 06/2025

Grantor: Dedman Family Scholar in Clinical Care
Title of Project: A Registry and Clinical Biorepository for Genetic Neurodevelopmental Disorders
Role: Site Principal Investigator
Dates: 09/2020 – 08/2024

Grantor: UTSW Community Based Research Award
Title of Project: Social Skills Building through Wildlife Heroes Club at the Dallas Zoo
Role: Site Principal Investigator
Dates: 07/2020 – 08/2022

Steven Gray

Contractor: Nih-National Inst Of Neuro Disord & Strk/University Of California
Title of Project: Gray_Autoimmune Mechanisms in Peripheral Neuropathy
Role: Principal Investigator
Dates: 09/2019 – 08/2021

Contractor: CURESPG50
Title of Project: SPG50 Gene Therapy
Role: Co-Investigator
Dates: 02/2020 – 07/2022

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 – 11/2021

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 – 03/2023

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-Principal Investigator
Dates: 12/2020 – 11/2023

Contractor: Taysha Gene Therapies
Title of Project: DDX3X Gene Therapy
Role: co-Principal 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 – 07/2022

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

Contractor: Nih-National Inst Of Neuro Disord & Strk/University of Pennsylvania
Title of Project: Combination Therapy, Biomarkers and Imaging in Canine Krabbe Disease - YR3
Role: Site Principal Investigator
Dates: 06/2018 – 05/2021

Contractor: Hannah's Hope Fund
Title of Project: GAN vagus nerve injection studies
Role: Principal Investigator
Dates: 11/2019 – 01/2022

Contractor: Cure AHC Inc.
Title of Project: ATP1A3 Gene Therapy
Role: Principal Investigator
Dates: 12/2018 – 06/2021

Contractor: NIH/NINDS
Title of Project: Giant Axonal Neuropathy Gene Therapy
Role: Principal Investigator
Dates: Present

Contractor: Nih-Office Of The Director/Jackson Laboratory
Title of Project: The Jackson Laboratory Center for Precision Genetics: From New Models to Novel Therapeutics
Role: Site Principal Investigator
Dates: 12/2018 – 06/2021

Contractor: International Rett Syndrome Foundation
Title of Project: Can non-invasive interventions synergistically enhance the efficacy of MECP2 gene therapy?
Role: Mentor (Principal Investigator Sarah Sinnett)
Dates: 01/2019 – 10/2021

Contractor: NIH/NINDS

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

Role: Principal Investigator

Dates: 08/2018 – 11/2022

Contractor: Global Foundation For Peroxisomal Disord

Title of Project: Gene Therapy for Zellweger Spectrum Disorders

Role: Co-Investigator

Dates: 12/2018 – 07/2021

Susan Iannaccone

Contractor: SAREPTA

Title of Project: Long-term, Open-label Extension Study for Patients with DMD

Role: Site Co-Investigator (with Diana Castro, MD)

Dates: 06/2019 - 12/2040

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: Site Principal Investigator

Dates: Present

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: Site Principal Investigator

Dates: Present

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: DOD
Title of Project: Extracellular RNA markers of myotonic dystrophy
Role: SitePI, sub contract with Massachusetts General Hospital, PI Thurman Wheeler, MD
Dates: 2019 – 2023

Contractor: NIH
Title of Project: Network of Excellence in Neuroscience Clinical Trial (NeuroNEXT)
Role: Co-investigator (Richard Dewey, PI)
Dates: 2018 - 2023

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

Saima Kayani

Contractor: NIH
Title of Project: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D)
PI: Juan Pascual, MD
Role: MD, Co-Investigator
Dates: 2017 - 2021

Contractor: NIH
Title of Project: Compatibility of C7 with Ketogenic Diet in Patients Diagnosed with G1D
Role: Co-Investigator
Dates: 2017 - 2022

Contractor: NIH
Title of Project: Treatment Development of Triheptanoin (G1D)
PI: Juan Pascual, MD
Role: Co-Investigator
Dates: 2017 - 2022

Contractor: Investigator Initiated

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

PI: Ben Greenberg, MD

Role: Co-Investigator

Dates: 2020 - 2024

Berge Minassian

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

Title of Project: Genome Editing, mRNA Suppression, and Glycogen Chain Termination as Therapy for Lafora Disease YR5

Role: Principal Investigator

Dates: 07/2020 – 06/2021

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

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

Role: Principal Investigator

Dates: 07/2021 – 06/2022

Contractor: Ultragenyx Pharmaceutical Inc

Title of Project: Viral Gene Replacement Therapy for Unverricht-Lundborg Disease

Role: Principal Investigator

Dates: 12/2018 – 01/2021

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: Present

Sharmistha Mitra

Contractor: Dr. Roy Elterman Pediatric Neurology Research Fund

Title of Project: Investigating role of malin E3 ubiquitin ligase in Lafora Disease

Role: Principal Investigator

Dates: 09/2020 - 08/2021

Contractor: The Paul D. Wellstone Muscular Dystrophy Award

Title of Project: Investigating Ubiquitination of Bach1 in a Polyglucosan Body Myopathy-1 disease mouse model

Role: Principal Investigator

Dates: 01/2022 - 09/2022

Felix Nitschke

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 – 07/2022

Eric Remster

Contractor: Lilly USA, LLC
Title of Project: LY2951742 Protocol Addendum I5Q-MC-CGAS(1.1)
Role: Site Principal Investigator
Dates: 01/2019 – 12/2024

Tonia Sabo

Grantor: Impax Laboratories, LLC
Title of Project: A Clinical Study to Evaluate the Efficacy and Safety of Zolmitriptan Nasal Spray
Role: Site Principal Investigator
Dates: 01/2018 – 11/2023

Grantor: Impax Laboratories, LLC
Title of Project: A Multicenter, randomized,
Role: Site Principal Investigator
Dates: 2017 - 2021

Rana Said

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: 07/2020 – Present

Contractor: Zogenix
Title of Project: An Open-Label Extension Trial to Assess the Long-Term Safety of XZ008 (Fenfluramine hydrochloride) Oral Solution as an Adjunctive Therapy for Seizures in Patients with Rare Seizure Disorders such as Epileptic Encephalopathies including Davet Syndrome and Lennox-Gastaut Syndrome (STU 2019-0939)
Role: Co-Principal Investigator
Dates: 09/2019 – Present

Contractor: UCB Pharma
Title of Project: An open-label study to determine safety, tolerability and efficacy of long-term oral Lacosamide (LCM) as adjunctive therapy in children with epilepsy (STU 032017-109)
Role: Co-Principal Investigator
Dates: 03/2017 – Present

Contractor: UCB Pharma
Title of Project: A multicenter, open-label, long-term study to investigate the efficacy and safety of Lacosamide as adjunctive therapy in pediatric subjects with epilepsy with partial-onset (STU 112015-045)
Role: Co-Principal Investigator
Dates: 11/2017 – Present

Contractor: UCB Pharma

Title of Project: A multicenter, double-blind, randomized, placebo-controlled, parallelgroup study to investigate the efficacy and safety of Lacosamide as adjunctive therapy in subjects with epilepsy greater or equal to 1 month to less than 4 years of age with partial-onset seizures (STU 122013-035)

Role: Co-Principal Investigator

Dates: 12/2017 – Present

Sarah Sinnett

Grantor: Taysha Gene Therapies

Title of Project: PTEN Hamartoma Tumor Syndrome

Role: Principal Investigator

Dates: 6/2020 – 11/2022

Grantor: Taysha Gene Therapies

Title of Project: FOXP1 Gene Therapy Feasibility and Risk Assessment

Role: Principal Investigator

Dates: 07/2020 – 6/2022

Grantor: Taysha Gene Therapies

Title of Project: Pre-clinical assessment of a regulated miniMECP2 vector

Role: Principal Investigator

Dates: 05/2022 – 04/2023

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 – 11/2022

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

Deepa Sirsi

Grantor: GW Pharmaceuticals Inc

Title of Project: An Open-Label Exploratory Inve

Role: Principal Investigator

Dates: Present

Grantor: NIH/NINDS

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

Role: Co-Investigator

Dates: 07/2016 – Present

Grantor: NIH / NINDS

Title of Project: Treatment development of triheptanoin (C7) for Glucose transporter type I deficiency (G1D): A Phase I Maximum Tolerable Dose Trial

Role: Co-Investigator

Dates: 07/2016 - 05/2022

Grantor: NIH/NINDS

Title of Project: Compatibility of C7 with the Ketogenic Diet in Patients Diagnosed with Glucose Transporter Type 1 Deficiency G1D

Role: Co-Investigator

Dates: 01/2019 – 03/2022

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: Co-Investigator

Dates: 05/2020 - Present

Matthew Stokes

Grantor: Texas Institute of Brain Injury and Repair

Title of Project: North Texas Sports Concussion Network (CON-TEX)

Role: Prospective Registry

Dates: 2019 – Present

Grantor: National Institute of Neurological Disorders and Stroke

Title of Project: Endophenotypes of Persistent Post-Concussive Symptoms in Adolexcents (CARE4KIDS)

Role: Co-Investigator

Dates: 2019 – Present

Jeff Waugh

Grantor: American Academy of Neurology, Career Development Award

Title of Project: Evaluating a potential mechanism for dystonia: the role of impaired inhibition

Role: Principal Investigator

Dates: 02/2018 – 01/2022

Grantor: Elterman Family Foundation

Title of Project: Identifying a final common pathway in autism spectrum disorders: a striatal compartment analysis

Role: Principal Investigator

Dates: 10/2021 – Present

Grantor: Clinical and Translational Science Awards

Title of Project: Improving the efficacy of deep brain stimulation by adjusting surgical targets for individual anatomic variation

Role: Principal Investigator

Dates: 02/2022 – Present

Peer-Reviewed Publications

1. Aboujaoude A, **Minassian B**, **Mitra S**. [LUBAC: a new player in polyglucosan body disease](#). *Biochem Soc Trans*. 2021 Nov 1;49(5):2443-2454. PMID:34709403
2. Acsadi G, Crawford TO, Müller-Felber W, Shieh PB, Richardson R, Natarajan N, **Castro D**, Ramirez-Schrempp D, Gambino G, Sun P, Farwell W. [Safety and efficacy of nusinersen in spinal muscular atrophy: The EMBRACE study](#). *Muscle Nerve*. 2021 May;63(5):668-677. PMID:33501671

3. Ahonen S, Nitschke S, Grossman TR, Kordasiewicz H, Wang P, Zhao X, Guisso DR, Kasiri S, **Nitschke F, Minassian BA.** [Gys1 antisense therapy rescues neuropathological bases of murine Lafora disease.](#) *Brain.* 2021 Nov 29;144(10):2985-2993. PMID:33993268
4. Alkhatir RA, Ahonen S, **Minassian BA.** [SUCLA2 Arg407Trp mutation can cause a nonprogressive movement disorder - deafness syndrome.](#) *Ann Clin Transl Neurol.* 2021 Jan;8(1):252-258. PMID:33231368
5. Appavu B, Deng D, **Dowling MM,** Garg S, Mangum T, Boerwinkle V, Abruzzo T. [Arteritis and Large Vessel Occlusive Strokes in Children After COVID-19 Infection.](#) *Pediatrics.* 2021 Mar;147(3):. PMID:33277353
6. Armao D, Bouldin TW, Bailey RM, **Gray SJ.** [Extensive rod and cone photoreceptor-cell degeneration in rat models of giant axonal neuropathy: implications for gene therapy of human disease.](#) *Ophthalmic Genet.* 2021 Oct;42(5):600-603. PMID:33955818
7. Arraj P, Robbins K, **Dengle Sanchez L,** Veltkamp DL, Pfeifer CM. [MRI findings in juvenile Huntington's disease.](#) *Radiol Case Rep.* 2021 Jan;16(1):113-115. PMID:33204383
8. Banning A, Zakrzewicz A, **Chen X, Gray SJ,** Tikkanen R. [Knockout of the CMP-Sialic Acid Transporter SLC35A1 in Human Cell Lines Increases Transduction Efficiency of Adeno-Associated Virus 9: Implications for Gene Therapy Potency Assays.](#) *Cells.* 2021 May 19;10(5):. PMID:34069698
9. Barry M, Barry D, Kansagra AP, Hallam D, Abraham M, Amlie-Lefond C, Thrombolysis in Pediatric Stroke (TIPSTER) Investigators*.. [Higher-Quality Data Collection Is Critical to Establish the Safety and Efficacy of Pediatric Mechanical Thrombectomy.](#) *Stroke.* 2021 Apr;52(4):1213-1221. PMID:33719517
10. **Batley K, Castro D.** Evaluation of a hypotonic infant *Pediatric Society of Greater Dallas Newsletter.* 2021.
11. Beslow LA, Linds AB, Fox CK, Kossorotoff M, Zuñiga Zambrano YC, Hernández-Chávez M, Hassanein SMA, Byrne S, Lim M, Maduaka N, Zafeiriou D, **Dowling MM,** Felling RJ, Rafay MF, Lehman LL, Noetzel MJ, Bernard TJ, Dlamini N, International Pediatric Stroke Study Group.. [Pediatric Ischemic Stroke: An Infrequent Complication of SARS-CoV-2.](#) *Ann Neurol.* 2021 Apr;89(4):657-665. PMID:33332607
12. Bharucha-Goebel DX, Norato G, Saade D, Paredes E, Biancavilla V, Donkervoort S, Kaur R, Lehky T, Fink M, Armao D, **Gray SJ,** Waite M, Debs S, Averion G, Hu Y, Zein WM, Foley AR, Jain M, Bönnemann CG. [Giant axonal neuropathy: cross-sectional analysis of a large natural history cohort.](#) *Brain.* 2021 Nov 29;144(10):3239-3250. PMID:34114613
13. Braga BP, Sillero R, Pereira RM, Urgan K, Swift DM, Rollins NK, Hogge AJ, **Dowling MM.** [Dynamic compression in vertebral artery dissection in children: apropos of a new protocol.](#) *Childs Nerv Syst.* 2021 Apr;37(4):1285-1293. PMID:33155060
14. Budhu JA, Velazquez AI, **Said RR,** Jordan JT. [Opinion & Special Articles: Maximizing Inclusiveness and Diversity Through Virtual Residency Applications and Interviews.](#) *Neurology.* 2021 Sep 28;97(13):647-650. PMID:34266922

15. Bunt SC, Meredith-Duliba T, Didehkhani N, Hynan LS, LoBue C, **Stokes M**, Miller SM, Bell K, Batjer H, Cullum CM. [Resilience and recovery from sports related concussion in adolescents and young adults.](#) *J Clin Exp Neuropsychol.* 2021 Sep;43(7):677-688. PMID:34720048
16. Calame DG, **Hainlen M**, Takacs D, Ferrante L, Pence K, Emrick LT, Chao HT. [EIF2AK2-related Neurodevelopmental Disorder With Leukoencephalopathy, Developmental Delay, and Episodic Neurologic Regression Mimics Pelizaeus-Merzbacher Disease.](#) *Neurol Genet.* 2021 Feb;7(1):e539. PMID:33553620
17. Caruso JP, Janjua MB, **Dolce A**, Price AV. [Retrospective analysis of open surgical versus laser interstitial thermal therapy callosotomy in pediatric patients with refractory epilepsy.](#) *J Neurosurg Pediatr.* 2021 Jan 22;27(4):420-428. PMID:33482643
18. **Challa L, Bonnet W, Goodspeed K**, Pfeifer C. [Microembolic Thrombi in a patient with Vaping Related Lung Injury.](#) *Pediatric Stroke.* 2021
19. **Chen X**, Kang R, Kroemer G, Tang D. [Targeting ferroptosis in pancreatic cancer: a double-edged sword.](#) *Trends Cancer.* 2021 Oct;7(10):891-901. PMID:34023326
20. **Chen X**, Zeh HJ, Kang R, Kroemer G, Tang D. [Cell death in pancreatic cancer: from pathogenesis to therapy.](#) *Nat Rev Gastroenterol Hepatol.* 2021 Nov;18(11):804-823. PMID:34331036
21. **Chen X**, Snanoudj-Verber S, Pollard L, Hu Y, Cathey SS, Tikkanen R, **Gray SJ**. [Pre-clinical Gene Therapy with AAV9/AGA in Aspartylglucosaminuria Mice Provides Evidence for Clinical Translation.](#) *Mol Ther.* 2021 Mar 3;29(3):989-1000. PMID:33186692
22. Day JW, Finkel RS, Chiriboga CA, Connolly AM, Crawford TO, Darras BT, **Iannaccone ST**, Kuntz NL, Peña LDM, Shieh PB, Smith EC, Kwon JM, Zaidman CM, Schultz M, Feltner DE, Tauscher-Wisniewski S, Ouyang H, Chand DH, Sproule DM, Macek TA, Mendell JR. [Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy in patients with two copies of SMN2 \(STR1VE\): an open-label, single-arm, multicentre, phase 3 trial.](#) *Lancet Neurol.* 2021 Apr;20(4):284-293. PMID:33743238
23. Donkervoort S, Krause N, Dergai M, Yun P, Koliwer J, Gorokhova S, Geist Hauserman J, Cummings BB, Hu Y, Smith R, Uapinyoying P, Ganesh VS, Ghosh PS, Monaghan KG, Edassery SL, Ferle PE, Silverstein S, Chao KR, Snyder M, Ellingwood S, Bharucha-Goebel D, **Iannaccone ST**, Dal Peraro M, Foley AR, Savas JN, Bolduc V, Fasshauer D, Bönnemann CG, Schwake M. [BET1 variants establish impaired vesicular transport as a cause for muscular dystrophy with epilepsy.](#) *EMBO Mol Med.* 2021 Dec 7;13(12):e13787. PMID:34779586
24. Erdei C, Liu CH, **Machie M**, Church PT, Heyne R. [Parent mental health and neurodevelopmental outcomes of children hospitalized in the neonatal intensive care unit.](#) *Early Hum Dev.* 2021 Mar;154():105278. PMID:33221031
25. Fox J, Thodeson DM, **Dolce AM**. [Nicotine: A Targeted Therapy for Epilepsy Due to nAChR Gene Variants.](#) *J Child Neurol.* 2021 Apr;36(5):371-377. PMID:33284031
26. **Goodspeed K**. Autism Spectrum Disorder *Pediatric Society of Greater Dallas Newsletter.* 2021 4th Quarter.

27. **Goodspeed K**, Feng C, Laine M, Lund TC. [Aspartylglucosaminuria: Clinical Presentation and Potential Therapies.](#) *J Child Neurol.* 2021 Apr;36(5):403-414. PMID:33439067
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29. Gumusgoz E, Guisso DR, Kasiri S, Wu J, Dear M, Verhalen B, Nitschke S, **Mitra S, Nitschke F, Minassian BA.** [Targeting Gys1 with AAV-SaCas9 Decreases Pathogenic Polyglucosan Bodies and Neuroinflammation in Adult Polyglucosan Body and Lafora Disease Mouse Models.](#) *Neurotherapeutics.* 2021 Apr;18(2):1414-1425. PMID:33830476
30. Haffner DN, **Machie M**, Hone E, **Said RR**, Maitre NL. [Predictors of Neurodevelopmental Impairment After Neonatal Bacterial Meningitis.](#) *J Child Neurol.* 2021 Oct;36(11):968-973. PMID:34256644
31. Heitkotter H, Linderman RE, Cava JA, Woertz EN, Mastey RR, Summerfelt P, Chui TY, Rosen RB, Patterson EJ, Vincent A, Carroll J, **Minassian BA.** [Retinal alterations in patients with Lafora disease.](#) *Am J Ophthalmol Case Rep.* 2021 Sep;23():101146. PMID:34195479
32. **Iannaccone ST**, Paul D, **Castro D**, Weprin B, Swift D. [Delivery of Nusinersen Through an Ommaya Reservoir in Spinal Muscular Atrophy.](#) *J Clin Neuromuscul Dis.* 2021 Mar 1;22(3):129-134. PMID:33595996
33. Israelian L, Nitschke S, Wang P, Zhao X, Perri AM, Lee JPY, Verhalen B, **Nitschke F, Minassian BA.** [Ppp1r3d deficiency preferentially inhibits neuronal and cardiac Lafora body formation in a mouse model of the fatal epilepsy Lafora disease.](#) *J Neurochem.* 2021 Jun;157(6):1897-1910. PMID:32892347
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35. Judson MC, Shyng C, Simon JM, Davis CR, Punt AM, Salmon MT, Miller NW, Ritola KD, Elgersma Y, Amaral DG, **Gray SJ**, Philpot BD. [Dual-isoform hUBE3A gene transfer improves behavioral and seizure outcomes in Angelman syndrome model mice.](#) *JCI Insight.* 2021 Oct 22;6(20):. PMID:34676830
36. Khan TR, Zimmern V, Aquino V, **Wang C.** [Autologous hematopoietic stem cell transplantation in a pediatric patient with aquaporin-4 neuromyelitis optica spectrum disorder.](#) *Mult Scler Relat Disord.* 2021 May;50():102852. PMID:33662857
37. Klimchak AC, Szabo SM, Qian C, Popoff E, **Iannaccone S**, Gooch KL. [Characterizing demographics, comorbidities, and costs of care among populations with Duchenne muscular dystrophy with Medicaid and commercial coverage.](#) *J Manag Care Spec Pharm.* 2021 Oct;27(10):1426-1437. PMID:34595954
38. Kot S, Karumuthil-Melethil S, Woodley E, Zaric V, Thompson P, Chen Z, Lykken E, Keimel JG, Kaemmerer WF, **Gray SJ**, Walia JS. [Investigating Immune Responses to the scAAV9-HEXM Gene Therapy Treatment in Tay-Sachs Disease and Sandhoff Disease Mouse Models.](#) *Int J Mol Sci.* 2021 Jun 23;22(13):. PMID:34201771

39. Kour S, Rajan DS, Fortuna TR, Anderson EN, Ward C, Lee Y, Lee S, Shin YB, Chae JH, Choi M, Siquier K, Cantagrel V, Amiel J, Stolerman ES, Barnett SS, Cousin MA, **Castro D**, McDonald K, Kirmse B, Nemeth AH, Rajasundaram D, Innes AM, Lynch D, Frosk P, Collins A, Gibbons M, Yang M, Desguerre I, Boddaert N, Gitiaux C, Rydning SL, Selmer KK, Urreiziti R, Garcia-Oguiza A, Osorio AN, Verdura E, Pujol A, McCurry HR, Landers JE, Agnihotri S, Andriescu EC, Moody SB, Phornphutkul C, Sacoto MJG, Begtrup A, Houlden H, Kirschner J, Schorling D, Rudnik-Schöneborn S, Strom TM, Leiz S, Juliette K, Richardson R, Yang Y, Zhang Y, Wang M, Wang J, Wang X, Platzer K, Donkervoort S, Bönnemann CG, Wagner M, Issa MY, Elbendary HM, Stanley V, Maroofian R, Gleeson JG, Zaki MS, Senderek J, Pandey UB. [Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder.](#) *Nat Commun.* 2021 May 7;12(1):2558. PMID:33963192
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45. Manouchehri N, Guisso DR, Hussain RZ, **Minassian BA**, Stüve O. [The antioxidant MnTBAP does not effectively downregulate CD4 expression in T cells in vivo.](#) *J Neuroimmunol.* 2021 May 15;354():577544. PMID:33756414
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49. McGetrick ME, Varughese NA, Miles DK, **Wang CX**, McCreary M, Monson NL, Greenberg BM. [Clinical Features, Treatment Strategies, and Outcomes in Hospitalized Children With Immune-Mediated Encephalopathies.](#) *Pediatr Neurol.* 2021 Mar;116():20-26. PMID:33388545
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