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. Dr. Berge Minassian became the Division Chief in 2016, and the Division has been a part of the Department of Pediatrics since 2006.

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 ten 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 in the autism spectrum
- Neuroimaging studies in pediatric movement disorders
- Functional Neurological Disorders
- Clinical Trials in Pediatric Migraine
- Concussion Recovery utilizing cerebrovascular functioning

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:

- a “Categorical” Child Neurology Residency pathway (5-year program, with 2-years preliminary UT Southwestern Pediatrics residency, followed by 3-years of Child Neurology residency),
- a “Categorical” Child Neurology Basic Neuroscience Research pathway (5-year program, with 1-year preliminary UT Southwestern Pediatrics residency, 1 year of ABPN-approved Research, followed by 3-years of Child Neurology residency),
- a “Reserved” Child Neurology pathway (3 years of Child Neurology residency after completion of 2-3 years of ACGME Pediatrics residency training)
- a “Categorical” Neurodevelopmental Disabilities Residency Program (6-year program, with 2-years preliminary UT Southwestern Pediatrics residency, followed by 4-years of Neurodevelopmental Disabilities residency),
- fellowship training in many pediatric neurology subspecialties, including 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. 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. Castro and 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 that 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 Pediatric Headache Fellowship Director, Dr. Tonia Sabo and is now training its third Pediatric Headache fellow, with a newly developed Sports Neurology track. Dr. Eric Remster, the first graduate of the Pediatric Headache Fellowship, recently joined the Pediatric Headache faculty and 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 29 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.
Wilmot Bonnet, M.D.
Assistant Professor, Pediatrics and Neurology & Therapeutics

**B.A.**
- UT Austin (With Honors), 2010

**M.D.**
- UT Medical School, Houston, 2014

**Postdoctoral Training**
- Residency, Pediatrics
  - Children’s Health Pediatrics Residency,
    UT Southwestern, 2014 – 2016
- Residency, Child Neurology Residency,
  UT Southwestern, 2016 – 2019
- Fellowship, Pediatric Vascular Neurology,
  UT Southwestern, 2019 – 2020

**Interests**
Acute intervention and management of cerebrovascular disease in children, cardioembolic stroke in children with congenital heart disease, genetic and inflammatory arteriopathies, stroke recovery and rehabilitation, quality improvement in patient and family education

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Sharmistha Mitra, Ph.D.
Assistant Instructor, Pediatrics

**B.S.**
- University of Calcutta, Kolkata, India 2006
- 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**

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Felix Nitschke, Ph.D.
Assistant Instructor, Pediatrics and Biochemistry

**M.S.**
- University of Potsdam, Germany 2008

**Ph.D.**
- University of Potsdam (Magnum Cum Laude), Germany 2013

**Postdoctoral Training**
- Research Fellow, Genetics and Genome Biology
  The Hospital for Sick Children Research Institute, Toronto, Canada, 2013 – 2017

**Interests**
Regulatory mechanisms impacting structure, function and location of glycogen; Molecular consequences of glycogen storage diseases; Development of treatment strategies on molecular basis of the disease pathogenesis; Engineering proteins for improved Gene Therapy.
Afsaneh Talai, MD
Assistant Professor, Pediatrics

B.S.
Northwestern University, Evanston, IL, 2010

M.D.
University of Oklahoma College of Medicine, Oklahoma City, OK, 2014

Postdoctoral Training
Residency, Pediatrics
University of Chicago Medical Center, 2014-2016
Residency, Child Neurology Residency,
University of Chicago Medical Center, 2016-2019
Fellowship, Pediatric Epilepsy Fellowship,
Children’s Hospital Colorado, 2019 – 2020

Interests
Psychogenic non-epileptic seizures, Medically refractory epilepsy, Epilepsy surgery, Magnetoencephalography

Honors / Awards

Best Pediatric Specialists, D Magazine
- Susan Arnold
- Michelle Christie
- Michael Dowling
- Kimberly Goods speed
- Susan Iannaccone
- Rana Said
- Mathew Stokes
- Jeff Waugh

Texas Super Doctor, Texas Monthly
- Susan Iannaccone

Diana Castro
- Promoted to Associate Professor

Kim Goodspeed
- Dedman Family Scholars in Clinical Care

Rana Said
- 1st recipient of the Excellence in GME award, UT Southwestern Medical Center
Invited Lectures

Rachel Bailey
- SLC13A5 Deficiency 4th Annual Research Roundtable, Teleconferenced, March 2020
  - Presentation, “Gene therapy for SLC13A5 Deficiency”

Diana Castro
- Cure SMA Summit of Strength, Houston, TX, January 2020
  - “SMA Clinical Trials and Research Update”
- American Academy of Neurology Annual Meeting - Virtual Meeting, May 2020
  - “Nusinersen in Infantile-onset Spinal Muscular Atrophy: Results From Longer-term Treatment From the Open-label SHINE Extension Study”
- Cure SMA Annual Conference – Virtual Meeting, June 2020
  - “Motor Function Change Over Time Among Nusinersen-Treated Participants With Infantile-onset SMA in the ENDEAR/SHINE Study Who Met the Permanent Ventilation (PV) Definition”
- 6th Congress of the European Academy of Neurology - 1st Virtual Congress, June 2020
  - “Motor Function Change Over Time Among Nusinersen Treated Participants with Infantile-onset SMA in the ENDEAR-SHINE Study Who Met the PV Definition”
- Physical Medicine and Rehabilitation Symposium, Dallas, TX, August 2020
  - “New Landscapes in SMA”
- Academia Mexicana de Neurologia, Virtual, November 2020
  - “Miastenia Gravis en Ninos y Diagnostico deferencial”
- Nuevo Landscape en Atrofia Muscular Espinal, Santiago de Chile, Chile, November 2020
  - “EVELAM: Escuela de Verano Euro Latin Americana de Miologia”
- WORKAME 2020 Atrofia Muscular Espinal. Situación actual: Las Nuevas terapias, Buenos Aires, Argentina, Virtual, November 2020
  - “SOLANE: Sociedad Latinoamericana de Enfermedades Neuromusculares”
- 15th Brazilian Congress of Child Neurology, Virtual, November 2020
  - “Repairing the DNA: the state of the art of new therapies”

Xin Chen
- (2020). (Virtual) Boston, MA
  - “Preclinical results in rodents strongly support clinical evaluation of scAAV9/MFSD8as a potential gene therapy for CLN7 patients”

Kim Goodspeed
- TESS Research Foundation Family Meeting, March 2020
  - “The Nuts and Bolts of a Natural History Study”
- National Genetic Counselors Society Annual Meeting (virtual), November 2020
  - “Educational Breakout Session B03: “An update on epilepsy therapeutics: Exploring antisense oligonucleotides, viral vectors, and other precision medicine approaches in the genetic epilepsies”

Steven Gray
  - Co-Organizer, Invited Speaker, And Panelist: “AAV manufacture via triple transfection in HEK293 cells”
  - INVITED TALK: “AAV-mediated gene therapy for neurological diseases”
Michelle Machie

- 41st Western Perinatal Research Conference, Indian Wells, CA, January 2020
  - “Full Throttle-Implementation of a Process to Improve MR Spectroscopy in a Single NICU”

Berge Minassian

- Molecular and Human Genetics Seminar Series, Baylor Houston, TX, February 2020
  - “Gene Therapy: The Unavoidable Medicine of the Future.”

Sharmistha Mitra

- Glycobiology Interest group, Dallas, TX, April 2020
  - “Role of protein post-translational modifications in fatal glycogen storage disorders”

Rana Said

- Student Interest Group in Neurology (SIGN) Virtual Residency Webinar, American Academy of Neurology, 2020
  - Webinar and Panel Discussion
- 2nd Annual Pediatric Rehabilitation Symposium, Virtual, UTSW, August 2020
  - “CBD Oil Use in Cerebral Palsy”
- Johns Hopkins University, Virtual, 2020
  - Visiting Virtual Professor for Johns Hopkins Patient Virtual Rounds course: “Neurology Chats online teaching session for MS2, MS3 and MS4 students”

Sarah Sinnett

- Baylor College of Medicine, Virtual, August 2020
  - “A new approach for designing a feedback-enabled AAV genome improves therapeutic outcomes of miniMeCP2 gene transfer in mice modeling Rett syndrome (RTT).”

Deepa Sirsi

- 2020 Texas Pediatric Neurology Network Conference, March 2020
  - “Epileptic Encephalopathies – Controversies”

Jennifer Thomas

- Texas Neurological Society Annual Winter Conference, Austin, TX, January 2020
  - Imposter Syndrome: “The Confidence Gap”

Cynthia Wang

- World Encephalitis Day, Dallas, TX, February 2020
  - “Pediatric Autoimmune Encephalitis”

Jeff Waugh

- Le Bonheur Children’s Hospital Pediatric Neurology Symposium, Memphis, TN
  - “The Brain Circuits Behind Dystonia, How to Recognize Abnormal Movements in Children”
Conference Presentations

American Society for Gene and Cell Therapy 23nd Annual Meeting, Virtual, May 2020

Platform Presentation, “Direct vagus nerve injection of AAV9 as a treatment approach for autonomic dysfunction in Giant Axonal Neuropathy”

Poster Presentation, “Preclinical Gene Replacement Therapy with a New scAAV9/SUMF1Viral Vector for the Treatment of Multiple Sulfatase Deficiency.”

Sinnett SE, Boyle E, Lyons C, Gray SJ
“A new approach for designing a feedbackenabled AAV genome improves therapeutic outcomes of miniMeCP2 gene transfer in mice modeling Rett syndrome (RTT).”

Child Neurology Society and International Child Neurology Association Annual Meeting, San Diego, CA (Virtual), October 2020

Bain J, Green Snyder L, Helbig K, Cooper DD, Chung W, Goodspeed K
Poster Presentation – Poster 31 – “Comparison between Caregiver-Reported Simons Searchlight Data and Provider-Reported Published Data in SLC6A1-Related Disorders”

Rashid-Khan T, Wang C
Poster presentation, “Two cases of anti-Myelin Oligodendrocyte Glycoprotein (MOG) antibody disease with atypical presentations”.

MS 8th joint ACTRIMS/ECTRIMS meeting, Virtual, September 2020.

Khan T, Wang C
MS 8th joint ACTRIMS/ECTRIMS meeting, Virtual, September 2020.
Poster presentation, “Autologous hematopoietic stem cell transplantation in a pediatric patient with aquaporin-4 neuromyelitis optica spectrum disorder”.

Lorenzo J, Gonzalez Caldito N, Wang C
Poster presentation, “Acute disseminated encephalomyelitis in a pediatric patient in the setting of SARS-CoV2 with histopathology.”

Salinas V, Lorenzo J, Wang C
Poster presentation, “Myelin Oligodendrocyte Glycoprotein-antibody positive transverse myelitis presenting as acute flaccid paralysis in a child with enterovirus infection”.

Rhem B, Castro D, Wang C
Poster presentation, “Recurrent CNS Demyelination in a Pediatric Patient with History of Guillain-Barre Syndrome variant Being Treated with IVIG.”
Other Conferences

Bonnet W
International Stroke Conference, Los Angeles, CA, February 2020
Poster Presentation, “Early v Radiographic Recurrence or Extension Predicts Late Clinical Recurrence in Pediatric AIS”

Bunt S, Stokes M, Olson D, Vargas B, Hicks C, Heinzellmann M, Cullum CM
American Academy of Neurology Sports Concussion Virtual Conference, 2020
“Exploratory Use of Pupillometry in Patients with History of Headache/Migraine and Concussion”

Goodspeed K, Shaffo F, Minassian B, Gray S
Symposium: Towards Targeted Therapies for Neurodevelopmental Disorders, Children’s Hospital of Philadelphia (Virtual), July 2020
Poster Presentation, “Gene Transfer Therapy for Neurodevelopmental Disorders”

Stokes M
7th Annual PRiSM Meeting, Glendale, AZ, January 2020
Poster Presentation, “How do learning disorders impact clinical measures following concussion”

Stokes M, Bunt S
Southwest Athletic Trainers Association Virtual Convention
“Concussion Research to Practice for Athletic Trainers Timing of Youth Sports Participation and Concussion: Boots on the Ground”

Stokes M, Zynda A, Chung J, Silver C, Cullum CM, Miller S
American Academy of Neurology Sports Concussion Virtual Conference
“Do Learning Disorders Impact Clinical Measures Following Concussion?”

Presley C, Case T, Tarkenton T, Stokes M, Miller S, Bell K, Batjer H, Cullum CM
National Academy of Neuropsychology Annual Conference Chicago, IL, Virtual, October, 2020
“Post-Concussion Symptoms in Adolescent Athletes with Premorbid Psychiatric History: A Matched Case-Control Study”

Waugh JL, Bruggemann N, Sharma, N, Brieter HC, Blood AJ
Movement Disorders Society, Pan-American Section, Virtual, September 2020
“Striatal injury in early X-linked Dystonia Parkinsonism affects both matrix and striosomes.”

Waugh JL
International FND Symposium, Dec. 4, 2020
“Reasons For Inadequate Communication In Fnd”

Education and Training

Dr. Rana Said is the Associate Director of Education Affairs 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 16.5 years, the program has grown considerably. Along with the growth of the program, Dr. Lauren Dengle has joined as the Associate Program Director of the Child Neurology Residency Program. The program offers an ACGME-accredited 3-year training program, including a combined 5-year “Categorical” program with 2 years of ACGME-
accredited Pediatrics training and 3 years of ACGME-accredited Child Neurology training based at Children’s Health™ Dallas/UT Southwestern Medical Center. We also offer a “Categorical” Child Neurology Basic Neuroscience Research pathway (5-year program, with 1 year preliminary UT Southwestern Pediatrics residency, 1 year of ABPN-approved Research, followed by 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 complement to five Child Neurology residents each year, making it one of the larger 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 6-year “Categorical” program with 2 years of ACGME-accredited training and 4 years of ACGME-accredited Neurodevelopmental Disabilities training based at Children’s Health™ 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 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 programs accepts 2 fellow per year. Graduates are eligible for boards 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, which accepts one 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 United Council for Neurologic Subspecialties (UCNS)-accredited program. This program accepts one fellow per year. 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 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.
Research Activities

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

- Rachel Bailey is the Principal Investigator and Co-Investigator of several research projects funded by private foundations and industry. The main research areas are rare pediatric disorders, such as SLC13A5 deficiency and Giant Axonal Neuropathy, and age-related neurodegenerative disorders including Alzheimer’s disease and Frontotemporal Dementia. Dr. Bailey is working with clinicians within the Department of Pediatrics to initiate a natural history study for SLC13A5 deficiency for clinical trial readiness for a gene therapy that she is developing for this disorder. She is also developing unfunded projects in other areas, including collaborating with the Department of Radiology to develop the use of focused-ultrasound to target viral based gene therapies to select regions of the brain.

- 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 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, CLN7 Batten disease, SPG50 disease, SPG52 disease, SLC52A2 deficiency disease, and SRD5A3 deficiency disease. He is currently working to facilitate the initiation of gene therapy clinical trials for Aspartylglucosaminuria and CLN7 at UTSW. 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 Dengle 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.

• Dr. 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, neurodevelopmental outcomes in children after extra-corporeal membrane oxygenation, and mechanisms of Fragile X Syndrome cognitive deficits.

• Kimberley Goodspeed is involved in clinical research of rare neurogenetic and neurodevelopmental disorders including Phelan-McDermid Syndrome and Pitt-Hopkins 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 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 mucopolysaccaradosis 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. Concurrently, she is completing a graduate certificate program in Clinical Sciences through the UTSW Center for Translation Medicine.

• Steven Gray is the PI on several grant-funded projects. These include an NIH R01 grant to develop novel AAV capsids for CNS gene transfer, an NIH R01 grant to understand and treat Giant Axonal Neuropathy, NIH funding to test a gene therapy treatment for Krabbe disease, a sponsored research agreement from Abeona Therapeutics to support the initiation of a Phase I clinical trial for CLN1 Batten disease, and a sponsored research agreement from Neurogene to support the initiation of a Phase I clinical trial for Aspartylglucosaminuria. He also has funding from several small foundations to support the development of gene therapy treatments for CLN7 Batten disease, Multiple Sulfatase Deficiency, Rett syndrome, Angelman Syndrome, ATP1A3 Alternating Hemiplegia of Childhood, SLC6A1 disease, Mucolipidosis type II, SPG50 disease, SLC52A2 disease, and Leigh Syndrome. Dr. Gray is actively working to create a working core facility for research and clinical AAV manufacture, and also facilitate the initiation of gene therapy clinical trials for multiple rare neurological diseases.

• 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 is involved in various research projects funded by industry and federal grants. Saima is acting in capacity of Principal Investigator and co-investigator of these research projects. The main research interests are rare neurodegenerative disorders and development of gene therapy programs for these disorders. These disorders include various neuro-metabolic disorders including energy metabolism disorders, lysosomal storage disorders and peroxisomal biogenesis disorders.

• 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.

• 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. Rana 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.

- Deepa Sirsi is involved in a range of clinical research studies concerning EEG, EEG as a predictor of neuroimaging and outcome in pediatric ECMO, Genetic epilepsy syndromes and 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.
- Peter Tsai Dr. 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 R01 and 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 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.
Clinical Activities

The Pediatric Neurology multidisciplinary teams include board-certified pediatric neurologists, pediatric nurse practitioners, physician assistants, genetic counselor, 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. She directs the Phelan McDermid Syndrome and Pitt Hopkins Syndrome clinics, 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. The physicians divide their time between patient care, epilepsy surgical procedures and interpretation of neurophysiology studies. 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 sees outpatients at three locations. The majority are seen in the CHST clinic in downtown Dallas, with a smaller number seen at the CHST clinic at Texas Heath Presbyterian Hospital, and at the newly opened UTSW Pediatric Epilepsy Clinic in Frisco. Patient demand remained high in 2020 with 2105 new patient referrals to the CHST Epilepsy Center, and additional referrals accepted in Frisco. With the addition of Dr Afsaneh Talai to the team in 2020 there are now
8 epilepsy physicians, which has helped maintain access for new patient appointments despite demand for more physician time devoted to neurophysiology procedures in 2020. The Epilepsy Center physicians led the Pediatric Department in rapid transition to telemedicine at the onset of the pandemic, and completed more than 6000 outpatient visits in 2020.

**Epilepsy Surgery Program**

The Comprehensive Epilepsy Center was successfully recertified in 2020 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 will further enhance our ability to offer epilepsy surgery to complex epilepsy patients. On the Childrens Medical Center Dallas campus the center expects to begin offering the new therapeutic options of responsive neurostimulation and deep brain stimulation in 2021. 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 but went through a dramatic expansion in late 2017 with the addition of robot assisted stereotactic surgical procedures. The Covid-19 pandemic caused cancellation of surgeries and inpatient procedures for several months in 2020, however surgical volume increased towards the end of the year and is expected to continue to recover slowly in 2021. 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 clinic.

**Neurophysiology services**

Neurophysiology services have remained stable overall. A decrease in outpatient EEGs and Epilepsy Monitoring Unit admissions due to the pandemic was offset by an increase in inpatient video EEG monitoring studies. The center also added EEG interpretation at Parkland NICU in 2020, as well as continuing to provide NICU services at Texas Health Presbyterian Hospital Dallas and Clements University Hospital. The continued growth in ICU monitoring follows a national trend, and
represents a challenge to the Epilepsy Center physicians. Active recruitment efforts are aimed at hiring at least one additional faculty member in 2021 to help meet this need.

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.
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 Epilepsy Health Learning System in 2020 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
- 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 medical director, is a past board member of the National Association of Epilepsy Centers, and currently chairs their accreditation committee. She also served on an International League Against Epilepsy task force in 2019 for establishing international standards for accreditation of Epilepsy Centers.

Outreach:

The faculty and staff of the Epilepsy Center partner with the Epilepsy Foundation of Texas to provide medical staff to two epilepsy camps each year, provide educational programs and run family and teen support groups. Center physicians and staff meet monthly with the Epilepsy Foundation and with patient family partners in the community as part of ongoing Quality Improvement activities related to the Epilepsy Health Learning System.

Research and Education

In 2020 the Epilepsy Center physicians were site principal investigators for 4 clinical trials for new anti-seizure medications, and for a multicenter study of pharmacokinetics of antiseizure medication sponsored by the NICHD. Center physicians also provided EEG interpretation studies for a study of triheptanoin for Glut-1 deficiency, and for several natural history studies for genetic epilepsies which are overseen by other Pediatric Neurology faculty. Four faculty members presented research at national meetings in 2019, and four spoke at regional or national symposiums. Four faculty authored or co-authored a total of 7 papers in peer reviewed journals in 2020.

The Pediatric Epilepsy and Neurophysiology Fellowship programs continue to attract talented applicants and the majority of fellows go on to university faculty positions. In 2020 the program expanded to three available fellowship positions each year.
Current Grant/Contract Support

Rachel Bailey

**Contractor:** King Foundation  
**Title of Project:** Alzheimer’s Disease Gene Therapy Project  
**Role:** Principal Investigator  
**Dates:** 9/2019 – 8/2020

**Contractor:** G.D. Broughton, Jr. Foundation  
**Title of Project:** Alzheimer’s Disease Research Projects – Tau Gene Therapy Sub-project  
**Role:** Co-Investigator  
**Dates:** 1/2019 – 12/2022

**Contractor:** David M. Crowley Foundation  
**Title of Project:** Gene Therapy for Alzheimer’s Disease  
**Role:** Principal Investigator  
**Dates:** 2/2019 – 1/2020

**Contractor:** NIH-National Institute of Neuro Disorders & Stroke  
**Title of Project:** 4th International SLC13A5 Deficiency Research Roundtable  
**Role:** Principal Investigator  
**Dates:** 3/2020 – 2/2021

**Contractor:** TESS Research Foundation  
**Title of Project:** Gene Therapy for SLC13A5 Deficiency  
**Role:** Principal Investigator  
**Dates:** 11/2018 - 12/2020

**Contractor:** United MSD Foundation  
**Title of Project:** Multiple Sulfatase Deficiency Gene Therapy Using AAV  
**Role:** Principal Investigator  
**Dates:** 01/2018 – 3/2021

**Contractor:** Taysha Gene Therapies Inc.  
**Title of Project:** Tau Reduction Therapy  
**Role:** Principal Investigator  
**Dates:** 6/2020 – 5/2022

**Contractor:** Taysha Gene Therapies Inc.  
**Title of Project:** SLC13A5 SRA  
**Role:** Principal Investigator  
**Dates:** 6/2020 – 5/2022

**Contractor:** Applied Genetic Technologies Corporation  
**Title of Project:** Evaluate tolerability and efficacy of two vectors in the progranulin knock-out mouse model  
**Role:** Principal Investigator  
**Dates:** 2/2020 - Present
Diana Castro

Contractor: Cure SMA
Title of Project: Site Capacity Funding for SMA Patients
Role: Site Principal Investigator
Dates: 02/2018 – 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: 03/2015 – present

Contractor: Syneos Health LLC
Title of Project: An Open-label Extension Study for Patients with Spinal Muscular Atrophy Who Previously Participated in Investigational Studies of ISIS 396443
Role: Site Principal Investigator
Dates: 02/2016 – 02/2021

Contractor: Fibrogen Inc.
Title of Project: Trial of FG-3019, a Monoclonal Antibody to Connective
Role: Principal Investigator
Dates: 11/2016 – 11/2021

Contractor: RevenaGen BioPharma Inc.
Title of Project: A phase 2a open, 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: Sarepta Therapeutics
Title of Project: An Open-Label Extension Study for Patients with Duchenne Muscular Dystrophy Who Participated in Studies of SRP-5051
Role: Site Principal Investigator
Dates: 01/2019 – 01/2024

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 ofSRK-015 in Patients With Type 2 or Type 3 Spinal Muscular Atrophy(TOPAZ)
Role: Site Principal Investigator
Dates: 2019 – present

Contractor: Sarepta
Title of Project: A study to evaluate the safety, tolerability, and pharmacokinetics of a single dose of SRP-5051 in participants with DMD
Role: Site Principal Investigator
Dates: 2017 - 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: AveXis  
**Title of Project:** Gene replacement therapy clinical trial for patients with Spinal Muscular Atrophy Type 1 (STRIVE)  
**Role:** Site Principal Investigator  
**Dates:** 2017 - present  

Contractor: AveXis  
**Title of Project:** Study of Intrathecal Administration of AVXS-101 for Spinal Muscular Atrophy (STRONG)  
**Role:** Site Principal Investigator  
**Dates:** 2017 - present  

Contractor: Sarepta  
**Title of Project:** Study of SRP-4045 and SRP-4053 in DMD Patients (ESSENCE)  
**Role:** Site Principal Investigator  
**Dates:** 2017 - present  

Contractor: Sarepta  
**Title of Project:** Study of Eteplirsen in patients with DMD amenable to Exon 51 Skipping  
**Role:** Site Principal Investigator  
**Dates:** 2015 - present  

Contractor: PTC Pharmaceutical  
**Title of Project:** An open label safety study for previously treated Ataluren (PTC124) patients with nonsense mutation dystrophinopathy  
**Role:** Site Principal Investigator  
**Dates:** 2016 - present  

**Xin Chen**  
Contractor: Sappani Family Foundation  
**Title of Project:** SRD5A3 Gene Therapy  
**Role:** Co-Investigator  
**Dates:** 02/2020 – 01/2022  

Contractor: CURESPG50  
**Title of Project:** SPG50 Gene Therapy  
**Role:** Co-Investigator  
**Dates:** 02/2020 – 07/2021  

Contractor: Taysha Gene Therapies  
**Title of Project:** DDX3X Gene Therapy  
**Role:** Investigator  
**Dates:** 12/2020 - 11/2022
Lauren Dengle

**Contractor:** Phoenix Nest  
**Title of Project:** A Retrospective Natural History study of Mucopolysaccharidosis type IIIC  
**Role:** Principal Investigator  
**Dates:** 11/2020 – present

Alison Dolce

**Grantor:** Texas Institute of Brain Injury and Repair  
**Title of Project:** North Texas Sports Concussion Network (CON-TEX)  
**Role:** Prospective Registry  
**Dates:** 2019 - Present

Michael Dowling

**Grantor:** 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

Patricia Evans

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

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  
**Contractor:** Neurogene  
**Title of Project:** A Natural History and Outcome Measure Discovery Study of Aspartylglucosaminuria  
**Role:** Principal Investigator  
**Dates:** 10/2018 to 02/2020
Steven Gray

**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 – 06/2021

**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 – 03/2022

**Contractor:** Taysha Gene Therapies  
**Title of Project:** SLC6A1 Gene Therapy  
**Role:** Principal Investigator  
**Dates:** 04/2020 – 03/2022

**Contractor:** Cure SPG50 Foundation  
**Title of Project:** AP4M1 gene therapy  
**Role:** Principal Investigator  
**Dates:** 02/2020 – 07/2021

**Contractor:** Sappani Foundation  
**Title of Project:** SRD5A3 gene therapy  
**Role:** Principal Investigator  
**Dates:** 02/2020 – 01/2022

**Contractor:** University of Pennsylvania  
**Title of Project:** Combination Therapy, Biomarkers and Imaging in Canine Krabbe Disease - YR3  
**Role:** Site Principal Investigator  
**Dates:** 06/2018 – 05/2020

**Contractor:** Hannah’s Hope Fund  
**Title of Project:** GAN vagus nerve injection studies  
**Role:** Principal Investigator  
**Dates:** 11/2019 – 4/2021
Contractor: SLC6A1 Connect  
Title of Project: SLC6A1 gene therapy  
Role: Principal Investigator  
Dates: 10/2019 – 10/2020

Contractor: Angelman Syndrome Foundation, Inc.  
Title of Project: Angelman Syndrome Gene Therapy  
Role: Principal Investigator  
Dates: 12/2017 – 08/2020

Contractor: Cure AHC Inc.  
Title of Project: ATP1A3 Gene Therapy  
Role: Principal Investigator  
Dates: 12/2018 – 11/2020

Contractor: NIH/NINDS  
Title of Project: Giant Axonal Neuropathy Gene Therapy  
Role: Principal Investigator  
Dates: 04/2014 – 03/2020

Contractor: Mila’s Miracle Foundation to Stop Batten  
Title of Project: CLN7 Gene Therapy  
Role: Principal Investigator  
Dates: 05/2017 – 04/2020

Contractor: 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 – 06/2020

Contractor: International Rett Syndrome Foundation  
Title of Project: Can non-invasive interventions synergistically enhance the efficacy of MECP2 gene therapy?  
Role: Principal Investigator  
Dates: 01/2019 – 10/2020

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 – 07/2021

Susan Iannaccone  

Contractor: Parent Project Muscular Dystrophy (PPMD)  
Title of Project: Parent Project Muscular Dystrophy Certification  
Role: Co-director (with Diana Castro, MD)  
Dates: 2020-2021

Contractor: Muscular Dystrophy Association  
Title of Project: MDA Care Center  
Role: Co-director (with Diana Castro, MD)  
Dates: 2017 - 2021
Contractor: NIH
Title of Project: Wellstone Muscular Dystrophy Center
Role: Associate Director/co-Chair of Administrative Core (PI Eric Olson)
Dates: 2015-2025

Contractor: NIH, 1U10 NS 077323-01
Title of Project: Network of Excellence in Neuroscience Clinical Trial (NeuroNext)
Role: Site PI
Dates: 2018-2023

Contractor: DoD
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2019-2023

Contractor: Sarepta
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: Sarepta
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: Sarepta
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: Fibrogen
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: ReveraGen
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: ReveraGen
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: PTC Therapeutics
Title of Project: Extracellular RNA biomarkers of myotonic dystrophy
Role: Site PI (sub-contract with Massachusetts General Hospital, PI Thurman Wheeler)
Dates: 2020

Contractor: Biogen
Title of Project: An open label extension study for patients with spinal muscular atrophy who previously participated in investigational studies of ISIS 396443
Role: Co-PI
Dates: 2020
Contractor: AVEXIS
Title of Project: STRONG - Phase I, open-label, dose comparison study of AVXS-101 for sitting but non-ambulatory patients with spinal muscular atrophy
Role: Principle Investigator
Dates: 2020

Contractor: AVEXIS
Title of Project: SPRINT - A global study of a single, one-time dose of AVXS-101 delivered to infants with genetically diagnosed and pre-symptomatic spinal muscular atrophy with multiple copies of SMN2
Role: Principle Investigator
Dates: 2020

Contractor: AVEXIS
Title of Project: A long-term follow-up study of patients in the clinical trials for spinal muscular atrophy receiving AVXS-101
Role: Principle Investigator
Dates: 2020

Contractor: AVEXIS
Title of Project: A prospective, long term REGISTRY of patients with a diagnosis of spinal muscular atrophy
Role: Principle Investigator
Dates: 2020

Contractor: CureSMA
Title of Project: Multicenter Cure SMA Clinical Data Registry
Role: Co- Principle Investigator
Dates: 2020

Contractor: Scholar Rock
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 (TOPAZ)
Role: Co- Principle Investigator
Dates: 2020

Contractor: NEUROGENE
Title of Project: A Natural History and Outcome Measure Discovery Study of Charcot-Marie-Tooth 4J
Role: Co- Principle Investigator
Dates: 2020

Contractor: BIOGEN
Title of Project: Escalating Dose and Randomized, Controlled Study of Nusinersen (BIIB058) in Participants with Spinal Muscular Atrophy
Role: Co- Principle Investigator
Dates: 2020

Contractor: PFIZER
Title of Project: A PHASE 3, MULTICENTER, RANDOMIZED, DOUBLE-BLIND, PLACEBO-CONTROLLED STUDY TO EVALUATE THE SAFETY AND EFFICACY OF PF-06939926 FOR THE TREATMENT OF DUCHENNE MUSCULAR DYSTROPHY
Role: Principle Investigator
Dates: 2020
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 Subjects With Duchenne Muscular Dystrophy (EMBARK)  
**Role:** Co-Principle Investigator  
**Dates:** 2020

Contractor: SAREPTA  
**Title of Project:** A Multi-Center, Longitudinal Study of the Natural History of Subjects with Limb Girdle Muscular Dystrophy (LGMD) Type 2E (LGMD2E/R4), Type 2D (LGMD2D/R3), and Type 2C (LGMD2C/R5)  
**Role:** Co-Principle Investigator  
**Dates:** 2020

**Berge Minassian**

Contractor: University of Kentucky  
**Title of Project:** Genome Editing, mRNA Suppression, and Glycogen Chain Termination as Therapy for Lafora Disease YR3  
**Role:** Site Principal Investigator  
**Dates:** 07/2019 – 06/2021

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

**Eric Remster**

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

Contractor: Dr. Roy Elterman Pediatric Neurology Research Fund  
**Title of Project:** Glymphatic and Meningeal Lymphatic Imaging to Unravel Pediatric Idiopathic Intracranial Hypertension  
**Role:** Principal Investigator  
**Dates:** 09-2020 – 08/2021

**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
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:** FOXG1 Gene Therapy Feasibility and Risk Assessment  
**Role:** Principal Investigator  
**Dates:** 07/2020 – 12/2021

**Grantor:** Taysha Gene Therapies  
**Title of Project:** Pre-clinical assessment of a regulated miniMECP2 vector  
**Role:** Principal Investigator  
**Dates:** 05/2020 – 04/2022

**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 – 04/2022
Grantor: Rettsyndrome.org
Title of Project: Can non-invasive interventions synergistically enhance the efficacy of MECP2 gene therapy?
Role: Principal Investigator
Dates: 06/2018 – 10/2021

Deepta Sirsi

Grantor: Zogenix Inc
Title of Project: A Two-Part Study of ZX008
Role: Principal Investigator
Dates: 10/19/18 – 10/19/2023

Grantor: NNDS
Title of Project: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D)
Role: Co-Investigator
Dates: 07/01/2016 – 07/31/2021

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: 7/1/2016-7/31/2021

Grantor:
Title of Project: Compatibility of C7 with the Ketogenic Diet in Patients Diagnosed with Glucose Transporter Type 1 Deficiency G1D
Role: Co-Investigator
Dates:

Grantor:
Title of Project: A Multicenter, Open-Label, Long-term Extension Study to Investigate the Efficacy and Safety of Lacosamide as Adjunctive Therapy in Pediatric Subjects with Epilepsy with Partial-Onset
Role: Co-Investigator
Dates:

Grantor:
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

Grantor: USB Biosciences
Title of Project: An Open-Label Study to determine safety, tolerability and efficacy of long term lacosamide (LCM) as an adjunctive therapy in children with epilepsy
Role: Co-Investigator
Dates: March 12, 2016-current (still open but closed for enrollment)
Grantor:
Title of Project: A Two Part Study of ZX008 in Children and Adults with Lennox-Gastaut Syndrome (LGS); Part 1: A Randomized, Double-blind, Placebo-controlled Trial of Two Fixed Doses of ZX008 (Fenfluramine Hydrochloride) Oral Solution as Adjunctive Therapy for Seizures
Role: Principal Investigator
Dates: 10/2018 - 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

Jeff Waugh

Grantor: Massachusetts General Hospital
Title of Project: Assessment of presymptomatic stages of XDP
Role: Principal Investigator
Dates: 10/2019 – 09/2020

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 – 01/2021

Peer-Reviewed Publications


38. Striano P, Minassian BA. From Genetic Testing to Precision Medicine in Epilepsy. Neurotherapeutics. 2020 Apr;17(2):609-615. PMID:31981099


