Division Introduction

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
- 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 supply high-quality faculty for many years to come. Together with the Department of Neurology & Neurotherapeutics, the division offers:

- a three-year Child Neurology Residency Program (including a “Categorical” 5-year program, with 2-years preliminary UT Southwestern Pediatrics residency),
- a four-year Neurodevelopmental Disabilities Residency Program (including a “Categorical” 6-year program, with 2-years preliminary UT Southwestern Pediatrics 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

Each year, in collaboration with the Departments of Pediatrics and Neurology and Neurotherapeutics 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. The Symposium is directed by Dr. Iannaccone, 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. The Symposium is directed by Drs. Castro, Golla and Sirsi.
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, Tonia Sabo, MD and is now training its third Pediatric Headache fellow, with a newly developed Sports Neurology track. Eric Remster, MD, 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 which includes a Quality Initiative Care Pathway for Headache Treatment (which includes clinical templates for orders and published hospital policy) 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 22 faculty members, four 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 2018.

Rachel Bailey, Ph.D.
Assistant Professor, Pediatrics

B.S.
Rensselaer Polytechnic Institute, Troy, NY, 2008

Ph.D.
Ph.D. Candidate, Molecular Neuroscience
Mayo Clinic, Rochester, MN, 2008-2010
Ph.D., Neuroscience (Interdisciplinary Program)
University of Florida, Gainesville, FL, 2010-2013

Interests
Dr. Bailey has a broad background in studying protein aggregation in neurodegenerative disorders, modeling neurological diseases in rodents and developing gene therapies to treat such disorders. Her research interests involve designing novel AAV-based gene therapies for neurological disorders, optimizing their delivery to the nervous system, and facilitating the translation of these therapies into human testing. Her work in this area has included the development of AAV gene therapies for Giant Axonal Neuropathy, Multiple Sulfatase Deficiency (MSD) and Charcot Marie Tooth Neuropathy Type 4J (CMT4J). Her previous work contributed to the Phase 1 clinical trial for Giant Axonal Neuropathy, which is ongoing at the NIH, and she is currently working with other Division faculty to initiate phase 1 clinical trials to test gene therapies for MSD and CMT4J here at UTSW. Dr. Bailey is also developing gene therapies for the rare pediatric disorders SLC13A5 deficiency and NGLY1 deficiency and for adult tau-related disorders, such as Alzheimer’s disease.
Xin Chen, Ph.D.
Instructor, Pediatrics

M.D.
North China Coal Medical College, Tangshan, China, 1986
M.S., Pathophysiology
Hengyang Medical College, China, 1989
Ph.D., Biosignal Pathophysiology
Kobe University, Japan, 2000

Interests
Dr. Chen has extensive experience in using up-to-date knowledge, techniques, and skills to design and conduct experiments, to troubleshoot problems, to interpret data, and to present and publish results. He has been successfully training personnel at different levels. The year of 2014 was his turning point when Dr. Chen started to develop and employ AAV vectors to treat inherited diseases in animal models. His current research interests include designing novel AAV9-based gene therapies, testing these AAV vectors in Aspartylglucosaminuria (AGU) and CLN7 mice, and translating these therapies into human testing. The year of 2019 should be a very exciting year when Dr. Chen will be collaborating with experts from both within and outside UT Southwestern to initiate clinical trials in which AAV9 vectors will be tested for both AGU and CLN7 patients here at UT Southwestern. Dr. Chen is also very interested in developing novel AAV capsids with superior efficiency and specificity.

Kimberly Goodspeed, M.D.
Assistant Professor, Pediatrics, Neurology & Neurotherapeutics, and Psychiatry

B.S.
Texas Christian University, Fort Worth, TX, 2008
M.D.
UT Health Science Center, Houston, TX, 2012

Postdoctoral Training
Residency, Pediatrics
UT Southwestern/Children’s, 2012-2014
Residency, Neurodevelopmental Disabilities
UT Southwestern, 2014-2018

Interests
Dr. Goodspeed has experience in caring for medically complex patients with intellectual disability and autism spectrum disorders. She has a clinical research interest in rare neurogenetic disorders and is working closely with other Division faculty on a number of translational gene transfer therapy projects including Aspartylglucosaminuria (AGU), Multiple Sulfatase Deficiency (MSD), and Krabbe Disease. Dr. Goodspeed is currently board certified in Pediatrics in addition to board eligible for Neurology with a Special Qualification in Child Neurology and Neurodevelopmental Disabilities.

NEW FELLOWSHIPS APPROVED IN 2018

In 2018 new fellowship programs were approved for Fetal & Neonatal Neurology and Pediatric Headache Medicine. A complement increase was also approved in the Vascular Neurology fellowship for a Pediatric Vascular Neurology fellow.
Cynthia Wang, M.D.
Assistant Professor, Pediatrics, Neurology & Neurotherapeutics

B.A.
Stanford University, Stanford, CA, 2006

M.D.
UT Southwestern, 2011

Postdoctoral Training
- Residency, Pediatrics
  University of Michigan Health System/Mott Children’s Hospital, Ann Arbor, MI, 2011-2013
- Residency, Neurology/Child Neurology
  University of Michigan Health System/Mott Children’s Hospital, Ann Arbor, MI, 2013-2016
- Fellowship, Rare Neuroimmunological Diseases, UT Southwestern, 2016-2018

Interests
Dr. Wang is experienced in caring for patients with inflammatory disorders of the central nervous system including Acute Disseminated Encephalomyelitis (ADEM), Multiple Sclerosis, Neuromyelitis Optica Spectrum Disorder, and Autoimmune Encephalitis. Her clinical research interests include improving the diagnosis and treatment of antibody-negative autoimmune encephalitis and studying the long-term neuropsychological outcomes in children with autoimmune brain disorders. She is board certified in Child Neurology.

Honors / Awards

Best Pediatric Specialists, D Magazine
- Susan Arnold
- Diana Castro
- Michael Dowling
- Susan Iannaccone
- Juan Pascual
- Rana Said

Texas Super Doctor, Texas Monthly Magazine
- Susan Iannaccone
- Sailaja Golla

Susan Arnold
- Castle Connolly Exceptional Women in Medicine Award
- Castle Connolly Top Doctors Award

Michael Dowling
- Pediatric Neurology Resident Teaching Award “Golden Boots”

Rachel Bailey and Kimberly Goodspeed
- Finalist in Swim with the Sharks! – A Team Science Pilot Grant Competition, UT Southwestern

Kim Goodspeed
- NeuroNEX 2018-2019 Early Stage Investigator

Drew Thodeson
- Promotion to Assistant Professor
Invited Lectures

Susan Arnold

- Texas NeuroDiagnostic Society Annual Meeting, Grapevine, TX, November 2018
  - “Pediatric Epilepsy Syndromes”

Rachel Bailey

- Krabbe Translational Research Network 7th Annual Meeting, Clearwater, FL, March 2018
  - “AAV Vector Design to Address Peripheral Nerve Disease”
- Charcot-Marie-Tooth Association (CMTA) Gene Therapy Meeting, Baltimore, MD, July 2018
  - “AAV Targeting of Peripheral Neuropathy in Giant Axonal Neuropathy”
- SLC13A5 Deficiency Annual Research Roundtable, Palo Alto, CA, August 2018
  - “Gene Therapy for SLC13A5 Deficiency”

Diana Castro

- Myotonic Dystrophy Foundation Dallas Patient Group, Dallas, TX, January 2018
  - “Depression and Anxiety in Myotonic Dystrophy”
- Mexican Foundation of GBS and CIDP, Mexico City, Mexico, February 2018
  - “Guillain-Barre Syndrome and CIDP in Children and Teenagers”

Michael Dowling

- Annual Meeting of the Texas Neurologic Society, Austin, TX, February 2018
  - “Update on Pediatric Stroke”
- SickKids Pediatric Stroke Symposium in Honor of Gabrielle deVeber, MD, Toronto, Canada, April 2018
  - Debate vs. Adam Kirton, MD: “Childhood Stroke: Protocolized vs. Precision Treatment” (Position: Protocolized)
  - “Gabrielle deVeber and the International Pediatric Stroke Study”
- 1st Annual Aspen Conference on Pediatric Cerebrovascular Disease and Stroke, Aspen, CO, July 2018
  - “Acute Stroke in the ED and Inpatient Setting”
  - Mini Symposium: “Overview of Sickle Cell Trials”
  - “Neurologic Risk Factors in PHACES”
  - “Perinatal Stroke: Identification and Management”
  - “Importance of Family History in Cerebrovascular Disorders”
  - “Brain AVMs in Hereditary Hemorrhagic Telangiectasia”

Sailaja Golla

- Pitt Hopkins Scientific Symposium and Parent Conference – Minneapolis, June 2018
  - “Pitt Hopkins Clinic and Case Series”
- Rajiv Gandhi Institute of Health Sciences, VIMS, India, July 2018
  - “Assessment of Developmental Delay, Autism and ADHD”
- Invited Radio Interview/Health Show, Radio Caravan, November 2018
  - Community Education Topic, “Autism and learning disabilities”

Steven Gray

- AAV Gene Therapy Symposium, Houston, TX, December 2018
  - “Platform AAV-based Gene Transfer Approaches to Treat the Nervous System”
- Department of Neurology & Neurotherapeutics Grand Rounds, UT Southwestern, Dallas, TX, October 2018
  - “Viral Vectors in Neurologic Treatment”
• 7th ATP1A3 in Disease Symposium, Chicago, IL, October 2018
  - “AAV-based Gene Transfer to the Nervous System”
• 2018 NIH & FDA Gene Therapy Workshop, Bethesda, MD, August 2018.
  - “Patient Advocacy Driving Forward First-in-human Clinical Trials”
• American Society of Cell and Gene Therapy, Chicago, IL, May 2018
• 15th Annual World Congress for SBMT2, Los Angeles, CA, April 2018
  - “First-in-human Intrathecal AAV9 Gene Transfer for the Treatment of Nervous System Diseases”
• Cracking CLN7, Boston, MA, April 2018
  - “Platform Translational Nervous System Gene Transfer, Applied to CLN7”
• Gene Therapy for CMT: Opportunities and Challenges, Baltimore, MD, July 2018
  - “Gene Therapy for CMT: Challenges and Practical Considerations”
• 15th International Symposium on MPS and Related Diseases, San Diego, CA, August 2018
  - “Translating Gene Therapy from Bench to Bedside for Neurological Disorders”
• It Will Take a Village to Cure Rett Syndrome Conference, Boston, MA, May 2018
  - “Limitations and Needs of AAV-mediated MeCP2 Gene Transfer for the Treatment of Rett Syndrome”
• 2018 Family Conference on Pediatric Hereditary Spastic Paraparesis (HSP), Dallas, TX, April 2018
• Women’s Health Symposium, UT Southwestern, Dallas, TX, March 2018

Susan Iannaccone

• Inaugural Dr. Iannaccone Child Neurology Research Day, UT Southwestern, Dallas TX, June 2018
  - Keynote Speaker, “From Rochester to Dallas; From Therapeutic Nihilism to Cure. How a Child Neurologist Changes her Tune”
• Department of Neurology & Neurotherapeutics Grand Rounds, UT Southwestern, Dallas, TX, September 2018
  - “Clinical Aspects of DMD”
• First Annual End Duchenne Tour (PPMD) Meeting, Dallas, TX, October 2018
  - “DMD Research Overview”
• Annual American Academy of Pediatrics (AAP) Meeting, Orlando FL, November 2018
  - “Educating SMA Patients and Families on New Therapies”

Berge Minassian

• 46th SENP Meeting, European Society of Pediatric Neurology, Barcelona, Spain, April 2018
  - “Lafora and NCL: Cures at Last?”
• Italian Annual Journal Club, Study Days on Neuronal Excitability, San Terenziano, Italy, May 2018
  - “Epileptogenesis in Lafora Disease”
• Italian Lafora Disease Association, Instituto G. Gaslini, Genova, Italy, May 2018
  - “Update on Lafora disease”
• Lafora Workshop, San Diego, CA, August/September 2018
  - “Update on Lafora Disease Therapy”
• German Congress of Neurology, Berlin, Germany, October/November 2018
  - “The Lafora Type of Progressive Myoclonic Epilepsy – Soon to Be a Curable Condition?”
• SLC6A1Connect Roundtable Symposium, New Orleans, LA, November 2018
  - “The Epilepsy of GAT1 Deficiency”
• American University of Beirut, Beirut, Lebanon, December 2018
  - “Pathogenesis of Lafora Disease”
Tonia Sabo

- Childress Foundation-KiDS and PROS Clinic, Southern Methodist University, July 2018
  - “What is a concussion?”
- Women’s Dallas Auxiliary Group, Children’s Health Foundation, Dallas, Texas, February 2018
  - “Pediatric Headache and Concussion”
- WFAA Facebook Live, Dallas, TX, Recorded January 26, 2018
  - “Pediatric Headache”

Rana Said

- Caregiver Education and Networking Night for IDD Council of Tarrant County, Richland Hills, TX, September 2018
  - “Cannabinoid and Compassionate Use”
- Texas NeuroDiagnostic Society’s Annual Meeting, Grapevine, TX, November 2018
  - “Alternative Therapies for Epilepsy”

Jeff Waugh

- Cape University Biomedical Imaging Center, University of Cape Town School of Medicine, South Africa, February 2018
  - “Quantitative Assessment of Probabilistic Tractography: The Value of Colocalization”
- American Academy of Neurology Webinar Series, February 2018
  - “Getting Paid for Your Time, All of the Time”
- North Carolina Neurological Society (NCNS), Charlotte, NC, February 2018
  - Plenary Speaker, “Bulletproof Notes: How to Defend your Documentation Against Audits”
- 41st Annual Paediatric Refresher Course, University of Cape Town, South Africa, February 2018
  - Invited Lecturer, Clinical Consultant:
    - “Recognizing Dystonia in Children”
    - “Mechanisms of Injury in Cerebral Palsy: The Impact of Location and Etiology”
    - “Functional Neurological Disorders in Children: Towards a Data Driven Practice”
- 3rd Movement Disorders Society-PAS School for Neurology Residents, Grapevine, TX, March 2018
  - “Pediatric Movement Disorders: Case Presentations”
- South Shore Hospital’s Pediatric Summit, Weymouth MA, May 2018
  - “Recognizing Abnormal Movements in Children”

Conference Presentations

**American Academy of Neurology, Los Angeles, CA, April 2018**


Husari K, Labiner K, Said R
Said, R
Invited Course Director, Moderator and Speaker, Clerkship Director/Program Director Leadership Course Lecture, “Negotiation Skills/Conflict Resolution”

Invited Speaker, Medical Student Symposium: Careers in Education Session Lecture, “Neurology in Pop Culture”

Wang C, Conger D, Greenberg B
Poster, “Myelin oligodendrocyte glycoprotein positive cohort of 15 patients: Clinical, imaging, and optical coherence tomography characteristics.”

Wang C, Harder L, Greenberg B
Poster, “Case Series of Four High School Football Players with Multiple Sclerosis.”

Plumb P, Wang C, Greenberg B
Poster, “Acute Flaccid Myelitis: Treatment Outcomes from a Tertiary Referral Center.”

American Society for Gene and Cell Therapy, Chicago, IL, May 2018

Bailey R, Hooper A, Sanford L, Chen D, Gray S
Poster, “AAV Targeting of the Peripheral Nervous System”

Oral Presentation, “Review of CSF and Peripheral Immune Responses Following Intrathecal Gene Transfer for Giant Axonal Neuropathy”

Chen X, Snanoudji-Verber S, Pollard L, Cathey S, Gray S
Poster, “The Efficacy and Safety of scAAV9/AGA Gene Therapy in Aspartylglucosaminuria Mice”

Child Neurology Society Annual Meeting, Chicago, IL, October 2018

Golla S, Ren J, Malloy C, Pascual J
Poster, “Intramyocellular Lipid Excess in the Mitochondrial Disorder MELAS”

Kayani S, Dolce A
Poster, Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay: Expanding the Phenotypic Spectrum from a Clinician’s Perspective”

Machie M, Snyder M, Rollins N, Kayani S
Poster, “Diffusion Tensor Imaging Abnormalities in Patients with Developmental Delay”

Wang C, Miles D, Narayan R, Greenberg B
Poster, “Fulminant Acute Disseminated Encephalomyelitis Associated with Increased Intracranial Pressure and anti-MOG antibodies: Presentation of Four Cases.”

Waugh J
Invited Lecturer, “Practice Management I: Coding for Complex and Chronic Patients”
Course Director and Lecturer, “Inherited Movement Disorders in Children” and “Childhood Inherited Dystonias”
American Epilepsy Society Annual Meeting, New Orleans, LA, December 2018

Fox J, Thodeson D, Dolce A
Poster, “Epilepsy and Neuronal Nicotinic Acetylcholine Receptor Gene Variants-is Nicotine a Precision Therapy?”

Husari K, Labiner K, Said R
Poster, “Outcomes of Refractory Febrile Status Epilepticus”
Poster, “Time to Immunotherapy: A New Concept in Pediatric New Onset Refractory Status Epilepticus”

New Frontiers in Pediatric Neurology, Dallas, TX June 2018

Gray S
Invited Keynote Lecturer, “AAV-mediated Gene Therapy for Nervous System Disorders”

Iannaccone S
Invited Lecturer, “Desperately Seeking Early Diagnosis: NBS for Neuromuscular Disease”

Waugh J
Invited Lecturer, “Recognizing and Treating Movements Disorders in Children”

Other Conferences

Hurd, A, Remster, E., Sabo, T
American Headache Society, Scottsdale, AZ, July 2018
Poster, “Establishment of a Migraine Pathway QI in the Pediatric Emergency Room”

Purkayastha S, Frantz J, Hynan L, Sabo T, Sorond F, Bell KR
4th Federal Interagency Conference on Traumatic Brain Injury, Washington DC, June 2018
Poster, “Cerebral Vasoreactivity is Impaired Three Month Following Sports Related Concussion in Collegiate Athletes”

North American Brain Injury Society (NABIS) 14th Annual Conference, Houston, TX, March 2018
Oral Presentation, “Dynamic Cerebral Autoregulation Impairment at rest and During Physical Stress Persists Despite Symptom Resolution in C”

Texas Chapter of American College of Sports Medicine (ACSM), Austin, TX, March 2018
Poster, Cardiac Vagal Tone Impairment is Associated with Reduced Cerebral Blood Flow in Collegiate Athletes Following Concussion

Wang CS, Greenberg BM
American Neurological Association Meeting, Atlanta, GA, October 2018
Poster, “Newly Diagnosed Anti-Myelin Oligodendrocyte Glycoprotein Syndromes in the Inpatient Setting: Six-month Experience at a Tertiary Pediatric Center.”

Waugh, J
Multidisciplinary Treatment of Children with Complex Motor Disorders, Ft. Worth, TX, May 2018
Plenary speaker, “What Neuroimaging Can Teach Us About Movement Disorders in Cerebral Palsy”
Cerebral Palsy: Prep for Life 2016 Symposium, Dallas, TX, August 2018
Platform Presentation and Panel Discussion, “Current and Future Management of Movement Disorders in Cerebral Palsy”

Waugh JL, Makris N, Sharma N, Breiter HC, Blood AJ
American Neurological Association, Atlanta, GA, October 2018

Education and Training

Dr. Patricia Evans is the Director of the Neurodevelopmental Disabilities (NDD) residency program. The NDD residency is a four-year residency program, of which there are only 8 nationally. The program accepts one NDD resident per year. The program offers an ACGME-accredited 4-year program in Neurodevelopmental Disabilities, including a combined 6-year “Categorical” program with 2 years of ACGME-accredited Pediatrics training and 4 years of ACGME-accredited Neurodevelopmental Disabilities training, based at Children’s Health™ Dallas/UT Southwestern Medical Center. Graduates are eligible for 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.

Dr. Sailaja Golla serves as the Associate Program Director for the Neurodevelopmental Disabilities Residency Program. She also co-directs the Annual CME event “New Frontiers in Pediatric Neurology.”

The Neuromuscular Medicine program is an ACGME-accredited program, that accepts 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. 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 1 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.

Dr. Rana Said is the Director of Pediatric Neurology Education Programs for the division of Pediatric Neurology. 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 Director of the Child Neurology Residency Program (core program) for the past 15 years, the program has grown considerably. 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. The Program was approved by the ACGME in 2018 to increase complement to four Child Neurology residents each year, making it one of the larger Child Neurology training programs in the nation. The program received “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. Deepa Sirsi is the Associate Program Director of the Clinical Neurophysiology and Epilepsy fellowship programs. She is the director of the Pediatric tracks of these fellowships. Each ACGME program accepts 1 Pediatric subspecialty fellow per year. Graduates are eligible for the American Board of Psychiatry and Neurology in Clinical Neurophysiology or 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 1 fellow per year, who has completed prior training in either Child Neurology or Neonatal 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 SLC13A5 deficiency, Multiple Sulfatase Deficiency, and Charcot Marie Tooth disease type 4J. Dr. Bailey is working to facilitate the initiation of gene therapy clinical trials for Multiple Sulfatase Deficiency and Charcot Marie Tooth disease type 4J at UT SW. She is also developing unfunded projects in other areas, including the development of gene therapies for Alzheimer’s disease and for Frontotemporal Dementia.
- Diana Castro serves as the Principal Investigator and Co-Investigator of several research projects funded by industry and federal grants. The main research areas are Spinal Muscular Atrophy and Duchenne Muscular Dystrophy. She is also developing unfunded projects in other areas including Guillain Barre Syndrome, RYR1 myopathies and Myasthenia Gravis.
- Xin Chen is the co-investigator of several research projects funded by private foundations and industry to Steven Gray. Dr. Chen’s main research areas are Aspartylglucosaminuria and CLN7 Batten disease and is currently working to facilitate the initiation of gene therapy clinical trials for Aspartylglucosaminuria and CLN7 at UT SW. He is also helping in developing novel AAV capsids with superior efficacy, higher specificity, and less toxicity and mentoring laboratory personnel.
- Michael Dowling is involved in clinical research in the areas of stroke in children, Sturge-Weber syndrome, and neurologic complications of sickle cell disease.
- Patricia Evans is an active participant in translational research, both for the disorders of autism spectrum disorders and fragile X syndrome. She is participating 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.
Sailaja Golla is involved with multiple grants including Industry funded, federal funded and unfunded projects. She is the Site PI for the NIH Neuronext Trial for Fragile X Syndrome She also serves as Co PI for Phelan McDermid Syndrome funding by Nocarts and Rare Disease Network. She is involved in multiple studies looking at long term neurodevelopmental outcomes in ECMO patients, neurodevelopmental and neurobehavioral sequela in patients with dystrophinopathies.

Kimberly 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 aspartylglucosaminuria, multiple sulfatase deficiency, Krabbe Disease, and SLC6A1-related neurodevelopmental disorders.

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, and a sponsored research agreement from Abeona Therapeutics to support the initiation of a Phase I clinical trial for CLN1 Batten disease. He has additional Sponsored Research Agreements to initiate Phase I clinical trials for Aspartylglucosaminuria and Charcot-Marie-Tooth disease type 4J. He also has funding from several small foundations to support the development of gene therapy treatments for CLN7 Batten disease, Krabbe disease, Multiple Sulfatase Deficiency, Rett syndrome, Angelman Syndrome, Pitt-Hopkins disease, ATP1A3 Alternating Hemplegia of Childhood, SLC6A1 disease, Mucolipidosis type II, 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 clinical 12 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 and acting the Principal Investigator and co-investigator of several research projects funded by industry and federal grants. The main research interests are rare neurodegenerative disorders and development of gene therapy programs of these disorders. These disorders include various 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 which 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. She is currently the Co-Principal Investigator working with Lauren Dengle, Daniel Bowers (Neuro-oncology), Elizabeth Maher (Internal Medicine), Rebekah Clarke (Neuroradiology) and Robert Bachoo (Neurology & Neurotherapeutics) in a 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.

Deepa Sarsi is involved in a range of clinical research studies concerning EEG & autism, yield of EEG & imaging in complex febrile seizures, genetic epilepsy syndromes and industry funded anti-seizure medication studies. She collaborates with other clinicians and basic scientists in research involving treatment of sodium channelopathies and other genetic causes of epilepsy such as GLUT1 disorder.
• Drew Thodeson has collaborated with the Hsieh laboratory in designing an in vitro model of genetic epilepsy using induced pluripotent stem cells specifically looking at ARX and CHD2 gene mutations. He also has a research collaboration with Jason Park in the Department of Pathology evaluating the clinical diagnostic testing of childhood epilepsy using clinically available next generation sequencing.
• 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.
• Cynthia Wang is involved in a research project studying the longitudinal and neuropsychological outcomes of patients with acute disseminated encephalomyelitis (ADEM). She is also involved in a study of biomarkers in autoimmune brain 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
• 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 which 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. Sailaja Golla, Associate NDD Residency Program Director, is an expert in rare autism variants and directs the Phelan McDermid Syndrome and Pitt Hopkins Syndrome clinics and also participates in clinical trials for these rare syndromes. Dr. Kimberly Goodspeed recently joined the group upon completion of training and is developing a multidisciplinary specialty clinic for Pitt-Hopkins Syndrome, incorporating experts from Gastroenterology, Pulmonology & Sleep Medicine, and Physical Medicine and Rehabilitation in addition to the services provided by CADD. 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 straightforward and complex conditions, promotion of research and education, and community outreach to families and health care providers. In 2018 the epilepsy surgery program experienced dramatic growth and is becoming one of the largest pediatric surgery centers in the country and a leader in the use of new surgical modalities in children.

Access
The Epilepsy Center continues to see patients at three locations including CHST clinics in downtown Dallas and Plano, and also at Texas Heath Presbyterian Hospital in Dallas. A fourth location in Frisco is planned when UTSW opens specialty clinics there. Patient demand remains high with 2530 new patient referrals received in 2018. A seventh pediatric epilepsy physician was successfully recruited and will join the group in 2019.
Neurophysiology Services
Neurophysiology services continue to grow, with admissions to the Epilepsy monitoring unit increasing by 23% in 2018. Physicians continue to provide EEG interpretation services at two off-site NICU locations, Texas Health Presbyterian Hospital Dallas and Clements University Hospital in Dallas and expect to begin interpreting Parkland NICU studies in 2019.
**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. 2018 saw an expansion in use of stereo EEG monitoring, a new technology that reduces morbidity from invasive monitoring and offers surgical evaluation to patients with poorly defined or multifocal epileptic zones. The epilepsy surgery program is also a pioneer in the use of laser interstitial thermoablation therapy for corpus callosotomy. This minimally invasive surgical approach reduces surgical complications and allows for much faster recovery time. With the growth in 2018, UTSW now has one of the largest pediatric epilepsy surgery programs in the country. Addition of MEG source localization in 2019 is anticipated to further enhance our ability to evaluate complex epilepsy patients. 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.

**Epilepsy Disease Specific Certification**

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. The program was commended for its educational resources and for services aimed at helping teens with epilepsy transition to adulthood. In 2017 the center began screening all teens for depression, which is common in individuals with epilepsy, allowing the center to provide higher quality comprehensive care to this vulnerable population. The preliminary data from this program was presented at the American Epilepsy Society national meeting and has received media attention as an important study of large scale depression screening in an Epilepsy clinic.

**Epilepsy Center Outreach**

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. 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. The center partners with the Epilepsy Foundation of Texas to provide transition education and support services to teens with epilepsy and their parents. 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.

**Epilepsy Fellowship Training**

The pediatric Epilepsy and Neurophysiology Fellowship programs continue to attract talented applicants and the majority of fellows go on to university faculty positions. The UT Southwestern Epilepsy Fellowship Program was one of the first seven epilepsy training programs in the country to receive AGME accreditation and was reaccredited after a successful site visit in 2016. Three fellows presented original research at national meetings in 2018.

**Current Grant/Contract Support**

**Susan Arnold**

- **Grantor:** Duke Clinical Research Institute, funded by National Institute of Child Health and Human Development  
  **Title of Project:** Pharmacokinetics of Antiepileptic Drugs in Obese Children, NICHD-2015-AED01  
  **Role:** Site Principal Investigator  
  **Dates:** 2016 – present

- **Grantor:** 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:** Site Principal Investigator (Co-Site Investigator: Rana Said)  
  **Dates:** 2017 – present

- **Grantor:** UCB Pharma  
  **Title of Project:** Protocol EP0060 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 (STU 112015-045)  
  **Role:** Site Principal Investigator (Co-Site Investigators: Rana Said, Deepa Sirsi)  
  **Dates:** 2015 – present

- **Grantor:** UCB Pharma  
  **Title of Project:** A Multicenter, Double-blind, Randomized, Placebo-controlled, Parallel-group 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:** Site Principal Investigator (Co-Site Investigators: Rana Said, Deepa Sirsi)  
  **Dates:** 2015 – present

**Rachel Bailey**

- **Contractor:** TESS Research Foundation  
  **Title of Project:** Gene Therapy for SLC13A5 Deficiency  
  **Role:** Principal Investigator  
  **Dates:** 2018 – 2019
**Contractor:** United MSD Foundation  
**Title of Project:** Multiple Sulfatase Deficiency Gene Therapy Using AAV  
**Role:** Principal Investigator  
**Dates:** 2018 – 2020

**Contractor:** MRCG-HRB Joint Funding Scheme  
**Title of Project:** Toxicology Study to Support a Phase I/II Gene Therapy Clinical Trial for Multiple Sulfatase Deficiency  
**Role:** Co-Investigator  
**Dates:** 2018 – 2023

**Contractor:** Neurogene  
**Title of Project:** CMT4J Nonclinical Project  
**Role:** Co-Investigator  
**Dates:** 2018 – 2023

**Diana Castro**

**Grantor:** NIH/NINDS  
**Title of Project:** NeuroNEXT SMA Biomarker Protocol Development  
**Role:** Site Principal Investigator (Ohio State University); Susan Iannaccone, Co-Principal Investigator  
**Dates:** 2012 - present

**Contractor:** Sarepta Therapeutics  
**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:** Figrogen  
**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  
**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 Pharmaceutical  
**Title of Project:** EMBRACE: A Study to Assess the Safety and Tolerability of ISIS 396443 (ISIS SMNRx) in Participants with Spinal Muscular Atrophy  
**Role:** Site Principal Investigator  
**Dates:** 2015 – present

**Contractor:** Biogen 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: AveXis  
**Title of Project:** Gene Replacement Therapy Clinical Trial for Patients with Spinal Muscular Atrophy Type 1 (STRIVE)  
**Role:** Site Co-Principal Investigator  
**Dates:** 2017 – present

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

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

Contractor: Sarepta Therapeutics  
**Title of Project:** Study of Eteplirsen in patients with DMD amenable to Exon 51 Skipping  
**Role:** Site Co-Principal Investigator  
**Dates:** 2017 – present

Contractor: PTC Therapeutics  
**Title of Project:** A Phase 3 Efficacy and Safety Study of PTC124 in Subjects with Nonsense-Mutation-Mediated Duchenne and Becker Muscular Dystrophy  
**Role:** Site Co-Principal Investigator  
**Dates:** 2013 – present (extension study)

Contractor: Sarepta Therapeutics  
**Title of Project:** A Phase 2 Safety Study of Eteplirsen to Treat Early Stage Duchenne Muscular Dystrophy  
**Role:** Site Co-Principal Investigator  
**Dates:** 2015 – present

Contractor: Eli Lilly and Company  
**Title of Project:** A Phase 3 Randomized, Double-Blind, Placebo-Controlled, Trial of Tadalafil for Duchenne Muscular Dystrophy  
**Role:** Site Co-Principal Investigator  
**Dates:** 2013 – present

Contractor: ISIS Pharmaceuticals  
**Title of Project:** An Open-label Safety and Tolerability Study of ISIS SMNRx (CS12) in Patients with Spinal Muscular Atrophy Who Previously Participated in ISIS SMNRx-CS2 or ISIS SMNRx-CS10  
**Role:** Site Co-Principal Investigator  
**Dates:** 2012 – present

Contractor: ISIS Pharmaceuticals  
**Title of Project:** A Phase 3 Study to Assess the Efficacy and Safety of ISIS-SMN Rx (CS4) in Patients with Later-onset Spinal Muscular Atrophy  
**Role:** Site Co-Principal Investigator  
**Dates:** 2015 – present
Contractor: ISIS Pharmaceuticals  
**Title of Project:** A Phase 3 Study to Assess the Efficacy and Safety of ISIS-SMN Rx (CS3b) in Infants with Spinal Muscular Atrophy  
**Role:** Site Co-Principal Investigator  
**Dates:** 2015 – present

Contractor: Quintiles/Biogen Idec  
**Title of Project:** Phase 2 Study of ISIS 396443 (BIIB058) for Spinal Muscular Atrophy  
**Role:** Principal Investigator  
**Dates:** 2015 – present

Michael Dowling

**Grantor:** Seattle Children’s Hospital  
**Title of Project:** TIPSTERS: Thrombolysis in Pediatric Stroke Extended Results  
**Role:** Site Principal Investigator  
**Dates:** 2017 – 2018

**Grantor:** NIH  
**Title of Project:** PumpKIN: Pumps for Kids, Infants, and Neonates Clinical Trial  
**Role:** Neurologist  
**Dates:** 2017 - 2021

**Grantor:** NIH/NINDS  
**Title of Project:** SPRING: Primary Prevention of Stroke in Children with SCD in Sub-Saharan Africa II  
**Role:** Neurologist, Member of Data Safety Monitoring Board  
**Dates:** 2015 - 2021

**Grantor:** NIH/NINDS – R56  
**Title of Project:** VIPS2: Vascular Effects of Infection in Pediatric Stroke II  
**Role:** Site Principal Investigator  
**Dates:** 2016 – 2018

**Grantor:** NIH/NINDS – R01  
**Title of Project:** VIPS2: Vascular Effects of Infection in Pediatric Stroke II  
**Role:** Site Principal Investigator  
**Dates:** 2017 - 2018

**Grantor:** Pediatric Epilepsy Research Foundation  
**Title of Project:** SIPSII: Seizures in Pediatric Stroke  
**Role:** Site Principal Investigator  
**Dates:** 2015 – 2019

**Grantor:** Syncardia, Inc.  
**Title of Project:** Ventricular Assist Device Clinical Trial  
**Role:** Site Neurologist  
**Dates:** 2016 - 2018

Patricia Evans

**Grantor:** Fragile X Foundation  
**Title of Project:** Fragile X Clinical and Research Consortium Registry and Database Study  
**Role:** Site Principal Investigator  
**Dates:** 2012-present
Steven Gray

**Contractor:** Angelman Syndrome Foundation  
**Title of Project:** Angelman Syndrome Gene Therapy  
**Role:** Principal Investigator  
**Dates:** 2017 – 2019

**Contractor:** Mila’s Miracle Foundation to Stop Batten  
**Title of Project:** CLN7 Gene Therapy  
**Role:** Principal Investigator  
**Dates:** 2017 – 2019

**Contractor:** University of Pennsylvania/International Advocate for Glycoprotein Storage Diseases  
**Title of Project:** Evaluation of Adeno-associated Virus Gene Therapy in the Feline Model of Mucolipidosis II  
**Role:** Principal Investigator of the Subcontract  
**Dates:** 2016 – 2018

**Contractor:** Pitt-Hopkins Research Foundation  
**Title of Project:** Gene therapy for Pitt-Hopkins Syndrome  
**Role:** Co-Principal Investigator  
**Dates:** 2016 – 2018

**Contractor:** Abeona Therapeutics  
**Title of Project:** Completion of CLN1 Efficacy Studies and Novel CNS Capsid Evaluations  
**Role:** Principal Investigator  
**Dates:** 2016 – 2019

**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:** 2016 – 2021

**Contractor:** NIH/NINDS  
**Title of Project:** Giant Axonal Neuropathy Gene Therapy  
**Role:** Principal Investigator  
**Dates:** 2014 – 2019

**Contractor:** U. Penn/NIH/NINDS (subcontract)  
**Title of Project:** Combination Therapy, Biomarkers, and Imaging in Canine Krabbe Disease  
**Role:** Principal Investigator of the Subcontract  
**Dates:** 2016 – 2021

**Contractor:** Rett Syndrome Research Trust  
**Title of Project:** A Gene Therapy Consortium to Develop and Evaluate Gene Therapy Approaches in Rett syndrome  
**Role:** Co-Principal Investigator (1 of 4)  
**Dates:** 2014 – 2020

**Contractor:** NIH/NINDS  
**Title of Project:** Development of Intravenous AAV Vectors for Intractable Epilepsy  
**Role:** Principal Investigator  
**Dates:** 2013 – 2018
Contractor: Cure AHC  
Title of Project: ATP1A3 gene therapy  
Role: Principal Investigator  
Dates: 2018-2019

Contractor: Neurogene  
Title of Project: Gene therapy for Angelman Syndrome  
Role: Co-Principal Investigator  
Dates: 2018-2020

Contractor: Neurogene  
Title of Project: CLN5 non-clinical studies  
Role: Principal Investigator  
Dates: 2018-2020

Contractor: Neurogene  
Title of Project: IND-enabling Studies for CMT4J to Support the Initiation of an AAV9/Fig4 Gene Transfer Clinical Trial  
Role: Principal Investigator  
Dates: 2018-2023

Contractor: Neurogene  
Title of Project: IND-enabling studies for Aspartylglucosaminuria (AGU) to support the initiation of an AAV9/AGA gene transfer clinical trial  
Role: Principal Investigator  
Dates: 2018-2023

Sailaja Golla

Grantor: Novartis  
Title of Project: Biomarkers associated with Phelan-McDermid (22q13) Syndrome  
Role: Co-Principal Investigator  
Dates: 2012-2018

Grantor: NINDS via Neuronext  
Title of Project: Study: Effects of AFQ056 on Language Learning in Young Children with Fragile X Syndrome (FXS)  
Role: Site Principal Investigator  
Dates: 2017 – 2020

Grantor: Fragile X Foundation  
Title of Project: Fragile X Clinical and Research Consortium Registry and Database Study  
Role: Co-Principal Investigator  
Dates: 2012-present

Kimberly Goodspeed

Contractor: Novartis  
Title of Project: Biomarkers associated with Phelan McDermid (22q13) Syndrome  
Role: Co-Investigator  
Dates: 2012 – 2018
Contractor: Neurogene  
**Title of Project:** A Natural History and Outcome Measure Discovery Study of Aspartylglucosaminuria (AGU)  
**Role:** Principal Investigator  
**Dates:** 2018 – present

Contractor: Neurogene  
**Title of Project:** A Phase I Study of Intrathecal Administration of scAAV9/AGA for the Treatment of Aspartylglucosaminuria  
**Role:** Principal Investigator  
**Dates:** 2018 – present

Contractor: Neurogene  
**Title of Project:** A Natural History and Outcome Measure Discovery Study of Multiple Sulfatase Deficiency (MSD)  
**Role:** Principal Investigator  
**Dates:** 2018 – present

Contractor: Neurogene  
**Title of Project:** A Phase I Study of Intrathecal Administration of scAAV9/SUMF1 for the Treatment of Multiple Sulfatase Deficiency  
**Role:** Principal Investigator  
**Dates:** 2018 – present

Contractor: Neurogene  
**Title of Project:** A Phase I Study of Intrathecal Administration of scAAV9/GALC for the Treatment of Krabbe Disease  
**Role:** Principal Investigator  
**Dates:** 2018 – present

Contractor: NINDS via NeuroNEXT Early Stage Investigator Training Program  
**Title of Project:** A Natural History and Outcome Measure Discovery Study of Aspartylglucosaminuria (AGU)  
**Role:** Principal Investigator  
**Dates:** 2018 – 2019

**Susan Iannaccone**

Contractor: NIH, Wellstone Muscular Dystrophy Center  
**Role:** Associate Director and co-chair of Administrative Core  
**Dates:** 2015–2020

Grantor: NIH, 1U10 NS 077323-01  
**Title of Project:** Network of Excellence in Neuroscience Clinical Trial (NeuroNEXT)  
**Role:** Co-Investigator  
**Dates:** 2011 – 2018

Contractor: PTC Therapeutics  
**Title of Project:** Extension Study of Ataluren in Subjects with Nonsense-Mutation-Mediated Duchenne and Becker Muscular Dystrophy  
**Role:** Site Principal Investigator  
**Dates:** 2011 – Present

Contractor: Sarepta  
**Title of Project:** Essence: double-blind, placebo-controlled, multi-center study with an open-label extension to evaluate the efficacy and safety of SRP-4045 and SRP-4053 in patients with DMD  
**Role:** Site Principal Investigator  
**Dates:** 2015 – Present
Contractor: AveXis  
**Title of Project:** AVXS-101 for participants with SMA, 2017 - present  
**Role:** Site Principal Investigator  
**Dates:** 2017 – Present

Contractor: Fibrogen  
**Title of Project:** FG-3019, phase 2 in DMD  
**Role:** Co-Principal Investigator  
**Dates:** 2017 – Present

Contractor: Regeneron  
**Title of Project:** Vamorolone Phase 2 in DMD  
**Role:** Co-Principal Investigator  
**Dates:** 2017 – Present

Contractor: Biogen  
**Title of Project:** SHINE, CS II, open label extension for Nusinersen in SMA  
**Role:** Site Co-Principal Investigator  
**Dates:** 2017 – Present

Contractor: AveXis  
**Title of Project:** Phase 1, Phase 2b Study of AVXS-101 for participants with SMA Type 1 (STRIVE)  
**Role:** Site Principal Investigator  
**Dates:** 2017 – Present

Contractor: AveXis  
**Title of Project:** Phase 1, Phase 2b study of AVXS-101 for participants with SMA type 2 (STRONG)  
**Role:** Site Principal Investigator  
**Dates:** 2018 – Present

Contractor: AveXis  
**Title of Project:** Phase 1, Phase 2b study of AVXS-101 for participants with asymptomatic SMA (SPRINT)  
**Role:** Site Principal Investigator  
**Dates:** 2018 – Present

Contractor: Capricor  
**Title of Project:** HOPE -2: Phase 2b trial of CAP-1002 in DMD non-ambulatory patients  
**Role:** Site Principal Investigator  
**Dates:** 2017 – Present

**Saima Kayani**

**Grantor:** NIH  
**Title of project:** Study: Dietary treatment of Glucose Transporter Type 1 Deficiency (G1D).  
**Role:** Co- Investigator  
**Dates:** 2017 – present

**Grantor:** NIH  
**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:** 2016 – present
**Grantor:** Nerogene  
**Title of project:** A Natural History and Outcome Measure Discovery Study of Variant Late Infantile Neuronal Ceroid Lipofuscinoses Type 5 (CLN5) and Variant Late Infantile Neuronal Ceroid Lipofuscinoses Type 7 (CLN7)  
**Role:** Principal Investigator  
**Dates:** 2018 – 2020

**Berge Minassian**

**Contractor:** National Institutes of Health  
**Title of Project:** Lafora Epilepsy – Basic Mechanisms to Therapy  
**Role:** Principal Investigator  
**Dates:** 2016 – 2021

**Contractor:** The Ontario Brain Institute  
**Title of Project:** New Approaches to Intractable Epilepsy: Phase II of The Epilepsy Discovery Project  
**Role:** Principal Investigator  
**Dates:** 2014 – 2019

**Contractor:** The Ontario Brain Institute  
**Title of Project:** EpLink: Phase II of The Epilepsy Discovery Project Genetic Database  
**Role:** Principal Investigator  
**Dates:** 2014 – 2019

**Tonia Sabo**

**Contractor:** Texas Institute of Brain Injury and Repair  
**Title of Project:** Texas Institute for Brain Injury and Repair (TIBIR) North Texas Sports Concussion Network (CON-TEX) Prospective Registry  
**Role:** Co-Investigator, Site PI, Children’s Medical Center  
**Dates:** 2015 – present

**Contractor:** Texas Institute of Brain Injury and Repair  
**Title of Project:** Association between Brain Blood Flow Regulation, Cognition and Balance in Concussion  
**Role:** Co-Investigator  
**Dates:** 2016 – present

**Contractor:** Texas Institute of Brain Injury and Repair  
**Title of Project:** Association Between Brain Blood Flow Regulation, Cognition and Balance in Concussion  
**Role:** Co-Investigator  
**Dates:** 2016 – present

**Contractor:** Impax Laboratories and PRA Health Sciences  
**Title of Project:** A Multicenter, Randomized, Double-blind, Placebo-controlled, Crossover Study to Evaluate the Efficacy and Safety of Zolmitriptan Nasal Spray for the Treatment of Acute Migraine in Subjects Ages 6 to 11 Years, with an Open-Label Extension  
**Role:** Principal Investigator  
**Dates:** 2017 – present
Rana Said

**Contractor:** UCB Pharma  
**Title of Project:** Retrospective Study of the Prevalence, Type and Outcome of Electrographic Seizures in Infants and Children with Non-Accidental Abusive Traumatic Brain Injury (STU 052013-056)  
**Role:** Co-Principal Investigator  
**Dates:** 2017 – present

Deepa Sirsi

**Contractor:** Quintiles  
**Title of Project:** Open Label, Long Term, Flexible Dose Study of Clobazam as Adjunctive Therapy in Pediatric Pts 1-16 yrs. with Dravet Syndrome  
**Role:** Principal Investigator  
**Dates:** 2015 – 2020

**Grantor:** NIH  
**Title of Project:** Study: Dietary Treatment of Glucose Transporter Type 1 Deficiency (G1D).  
**Role:** Co-Principal Investigator  
**Dates:** 2017 – present

**Grantor:** NIH  
**Title of Project:** Treatment Development of Triheptanoin (C7) for Glucose Transporter Type I Deficiency (G1D): A Phase I Maximum Tolerable Dose Trial  
**Role:** Co-Principal Investigator  
**Dates:** 2016 – present

**Grantor:** Zogenix  
**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 in Children and Adults with LGS, followed by Part 2: An Open-label Extension to Assess Long-Term Safety of ZX008 in Children and Adults with LGS  
**Role:** Principal investigator  
**Dates:** 2018- present

Jennifer Thomas

**Contractor:** NINDS - 1U01NS092764  
**Title of Project:** HEAL Trial: Randomized, Double-blind, Multi-center, Placebo-controlled Trial Designed to Test the Efficacy of High Dose Erythropoietin for Neuroprotection (Epo) in Term Infants with Hypoxic-ischemic Encephalopathy (HIE)  
**Role:** Site Co-Principal Investigator  
**Dates:** 2017 – 2022

Jeff Waugh

**Contractor:** 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:** 2018 – 2020
Contractor: Collaborative Center for X-linked Dystonia Parkinsonism
Title of Project: Evaluating functional network derangements in XDP using PET imaging
Role: Co-Principal Investigator
Dates: 2018 – 2020

Peer-Reviewed Publications


**Book Sections**
