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The Neurotransmitter

UT Health Austin Pediatric Neurosciences at Dell Children's

Promoting Discovery and Innovation in the Pediatric Neurosciences

dell children's
Ascension

The University of Texas at Austin
UT Health Austin

Dear Colleagues:

Welcome to UT Health Austin Pediatric Neurosciences at Dell Children's. In this issue, we highlight new faculty and staff; national and international advocacy and engagement efforts; and several recent accomplishments and events.

As our program continues expanding its impact in Central Texas and beyond, we remain dedicated to offering outstanding educational opportunities, promoting research and scholarly activity, and delivering exceptional multidisciplinary clinical care for all children with neurological disorders.

E. Steve Roach, MD
Chief of Pediatric Neurosciences
roache@austin.utexas.edu

EDMOND MEETS WITH LAWMAKERS FOR AAO MID-YEAR FORUM

Jane Edmond, MD, advocating at Capitol Hill

From April 17 to 20, pediatric neuro-ophthalmologist **Jane Edmond, MD**, president of the American Academy of Ophthalmology, joined nearly 500 ophthalmologists from across the country for the AAO's 2024 Mid-Year Forum and Congressional Advocacy Day in Washington, D.C., to advocate on behalf of the patients they serve and the profession. Edmond met with Congressional members to strengthen relationships crucial to the academy's advocacy efforts and to inform health care policies that impact ophthalmology research, practice, and care.

This year, Edmond and AAO members engaged Congress on topics such as pediatric access to eye care, prior authorization reform, increased research funding, artificial intelligence, sustainability, and engaging the next generation of ophthalmologists. Edmond also presented several legislators with the Visionary Award on behalf of the academy for their work in advancing high-quality eye care.

PEDIATRIC NEUROSCIENCES HOSTS FIRST ANNUAL IMPRES CONFERENCE

IMPRES panel discussion moderated by Dave Clarke, MD, with panelists (L to R) Ahmed Abdelmoity, MD, Priscilla Duong, PhD, and Sandi Lam, MD, MBA

The Comprehensive Pediatric Epilepsy Center, in collaboration with Lurie Children's Hospital and Corewell Health, presented the first Innovative Minds: Pediatric Research in Epilepsy Surgery (IMPRES) Conference held February 16-18 at the Hotel Van Zandt in downtown Austin.

The IMPRES program focused on evidence-based research on pediatric epilepsy surgery and highlighted collaboration between pediatric epilepsy centers and programs. The national conference featured six sessions and 25 presentations on topics including the process of identifying surgical patients, surgical techniques, medical and dietary management, and adult transition. Each session concluded with moderated panel discussions. Rick Boop, MD, emeritus chair of the Department of Neurosurgery at the University of Tennessee Health Science Center, former chief of Pediatric Neurosurgery at St. Jude Children's Research Hospital, and a member of the St. Jude Global Program, delivered the keynote lecture on the history of epilepsy surgery.

This year's conference organizers were pediatric neurosciences faculty members **Dave Clarke, MD**, and **Elizabeth Tyler-Kabara, MD, PhD**; Sandi Lam, MD, MBA, of Lurie Children's Hospital and Northwestern University Feinberg School of Medicine; and Daniel Arndt, MD, of Corewell Health. Planning is now underway for the second IMPRES conference.

M. Omar Iqbal, MD, Rick Boop, MD, and Sandi Lam, MD, MBA, at an IMPRES reception

A SECOND OPINION

This 12-year-old boy was evaluated for episodic headaches associated with neurological dysfunction. His headaches began around five years of age and initially occurred about once per year. Some of his earlier attacks were associated with a blind spot, and he was diagnosed with migraine. Around age 11, however, his headaches became more frequent and began to feature more troublesome neurological deficits. He was evaluated multiple times in an emergency department due to headaches that were at various times associated with visual disturbance, hemiplegia, hemisensory loss, and dysarthria.

Several cranial CTs and MRI scans were unremarkable. An echocardiogram revealed a patent foramen ovale (PFO), and he had already undergone PFO closure either to prevent suspected transient ischemic attacks or to minimize the severity of migraine. Unfortunately, his episodes continued after his PFO closure.

His mother and both of his grandmothers have headaches. Around age nine, his mother began having severe headaches associated with a blind spot and vomiting. As a young adult, her episodes became more frequent and began to feature extremity numbness and, on at least one occasion, inability to speak. Various symptomatic and prophylactic migraine medications were ineffective, so she underwent closure of a PFO to prevent presumed transient ischemic attacks.

When seen between attacks, he was asymptomatic and his examination was normal.

Initial Diagnosis

1. Complicated migraine
2. Rule out familial hemiplegic migraine

Are there additional diagnoses to consider? What additional diagnostic studies might be useful?

See below for additional discussion.

BRUMBACK ARTICLE SUMMARIZES NINDS RESEARCH WORKSHOP

Audrey Brumback, MD, PhD

Audrey Brumback, MD, PhD, pediatric neurologist and assistant professor of neurology, was the lead author of "Catalyzing Communities of Research Rigour Champions," recently published in *Brain Communications*. The article summarizes the discussions from a workshop of the same name sponsored by the National Institute of Neurological Disorders and Stroke (NINDS).

The authors describe how "rigor champions" within diverse areas of biomedical research are promoting rigorous, transparent research practices and provide a primer that encourages the broader research community to prioritize such practices. The article outlines recommendations to incorporate rigorous research practices into training and degree requirements.

The NINDS workshop was held last year at the National Institutes of Health campus in Bethesda, Maryland. Brumback was one of 50 attendees invited to discuss obstacles to research reform and develop initiatives for catalyzing change.

Read the article at https://doi.org/10.1093/braincomms/fcae120.

PEDIATRIC NEUROSCIENCES WELCOMES NEW COLLEAGUES

UT Health Austin Pediatric Neurosciences at Dell Children's has grown dramatically since it was founded in 2019. As of this summer, the multidisciplinary program features 20 child neurologists, 20 advanced practice providers, three pediatric neurosurgeons, three pediatric physical medicine and rehabilitation specialists, eight pediatric neuropsychologists, two genetic counselors, and a pediatric neuro-ophthalmologist. We recently welcomed new colleagues to the program.

Lindsey Elliott, PhD, is a pediatric psychologist who joins our program as an assistant professor in the departments of Neurology and Psychiatry and Behavioral Sciences. She specializes in treating children and adolescents with a wide range of functional disorders and will support our Psychogenic Nonepileptic Events Clinic.

Elliott earned her bachelor's degree in psychology from William & Mary in Williamsburg, Virginia, and her doctorate in clinical psychology from the University of Alabama at Birmingham. She completed an internship in pediatric psychology at Nationwide Children's Hospital in Columbus, Ohio, and a fellowship in pediatric psychology with a specialty in consultation-liaison at UT Health Austin's Texas Child Study Center and Dell Children's Medical Center. A member of the American Psychological Association and the Society of Pediatric Psychology, she is engaged in multisite research projects aimed at providing effective treatment to individuals with functional neurological disorders.

Avia Sutton, MPH, MS, is a genetic counselor in the Pediatric Neurogenetics Program. She earned a Bachelor of Science in behavioral neuroscience from Nova Southeastern University in Fort Lauderdale, Florida, a Master of Science in public health from Tuskegee University in Alabama, and a master's in genetic counseling from the University of North Carolina Greensboro. She then completed an internship at the Research Triangle Institute in North Carolina. Her interests include neurodevelopmental disability, public health genetics and education, and patient advocacy.

MacKenzie A. Howard, PhD, an assistant professor of neurology and neuroscience, recently joined our program. He received his bachelor's degree in biology from Lewis and Clark College in Portland, Oregon, and his PhD in physiology and biophysics from the University of Washington. He completed two postdoctoral fellowships at the University of California, San Francisco that focused on the cellular and circuit mechanisms of synaptic plasticity and connectivity in epilepsy and neurodevelopmental disease.

Howard's research has delved into ion channel function, neurotransmitter receptors, molecular signaling, learning and memory, and neural development and plasticity. In the Howard Neuro Lab, he uses his extensive background in cellular neurophysiology to understand mechanisms of genetic epilepsies.

ELECTED & SELECTED

Epilepsy Program Faculty Member & Student Receive Ken Shine Award

Liberty Hamilton, PhD, was one of four recipients of the 2024 Ken Shine Award for Excellence in Research and Research Mentorship. Hamilton was nominated for the category of Research Mentorship. **Arpan Patel**, a third-year MD/MSE candidate and a student in Hamilton's lab, was nominated for his project "Decoding Audiovisual Stimuli From Intracranial Recordings of Pediatric Epilepsy Patients." The annual research awards, made possible from a donation by Kenneth Shine, MD, recognize the achievements of students and trainees and mentorship by Dell Medical School faculty.

Brumback Mentee Awarded Alpha Omega Alpha Fellowship

Audrey Brumback, MD, PhD, is collaborating with Dell Medical School student **Matthew Taing** on his research project, "Investigating the Co-Occurrence of Neurodevelopmental Disorders and Atopic Diseases in U.S. Children: Implications for Healthcare Access and Quality," for which he received the 2024 Alpha Omega Alpha Carolyn L. Kuckein Student Research Fellowship. The national fellowship fosters the development of the next generation of medical researchers by providing financial support for research projects. Only one candidate from each medical school may be nominated by their Alpha Omega Alpha chapter.

DeLeon Serves on NAEC Guidelines Panel

Rosario DeLeon, PhD, assistant professor of neurology, was one of two neuropsychologists selected to serve on the panel of experts that developed the 2023 Guidelines for Specialized Epilepsy Centers: Report of the National Association of Epilepsy Centers Guideline Panel, published earlier this year. The panel drafted 52 evidence- and consensus-based recommendations on topics relevant to specialized epilepsy centers, including epilepsy monitoring unit care, neuropsychology, neuroimaging, surgery, and genetics. The executive summary of the guidelines, coauthored by **Dave Clarke, MD**, was published in *Neurology* and includes the complete guidelines in an appendix.

Gettig Awarded Texas Global Faculty Research Seed Grant

Kudos to **Kelly Gettig, DNP, APRN**, assistant professor of neurology and director of the Traumatic Brain Injury/Concussion Clinic, whose proposal "Exploring the Burden of Traumatic Brain Injury in Young Children in Northern Tanzania" was awarded a Faculty Research Seed Grant from The University of Texas at Austin's Texas Global Engagement project. Her project was selected based on its potential for long-term academic and societal impact, including its alignment with sustainable development goals and its potential to save lives and reduce inequality.

Texas Global supports the global engagement efforts of UT Austin faculty, students, and alumni. The seed grants are awarded semiannually to faculty whose collaborations with organizations and institutions abroad help address critical worldwide challenges. Gettig, who has partnered with Kilimanjaro Christian Medical University College in Tanzania, was one of 12 University faculty members to receive the spring 2024 grant.

Clarke Organizes Epilepsy Session for UN Conference

Dave Clarke, MD, chief of the Dell Children's Comprehensive Pediatric Epilepsy Center, organized and moderated the session "Addressing Deficits, Deficiencies, and Inequities in Managing Epilepsy in Small Island Developing States" at the United Nations' fourth International Conference on Small Island Developing States, held on May 27 at the American University of Antigua.

The conference brings international communities together to propose solutions that facilitate sustainable development and address the unique challenges these island nations face due to their geography. People living with epilepsy in these regions are disproportionately affected by such challenges and consequently experience many barriers to care. The epilepsy session originated from a UN proposal that calls for an approach to reducing these barriers to care based on the World Health Organization's Intersectoral Global Action Plan on epilepsy and other neurological disorders. Read more about the conference and session.

Chinthaparthi Receives 2024 Resident Teaching Award

Sireesha Chinthaparthi, MD, assistant professor of neurology, was recently honored with the Dell Children's 2024 Neurology Resident Teaching Award. Chinthaparthi was selected by Dell Medical School's pediatric and adult neurology residents. Congratulations, Dr. Chinthaparthi!

Faculty Members Receive Advising Award

Audrey Brumback, MD, PhD, and **Daniel Freedman, DO**, were honored with the annual Excellence in Specialty Advising Award by Dell Medical School's Career Advising and Mentoring Program. Brumback and Freedman, both C.A.M.P. Specialty Advisors, were selected for their dedication to offering students guidance and support throughout medical school.

Nine Faculty Members Named *Austin Monthly*'s Top Doctors of 2024

Each year *Austin Monthly* magazine solicits peer nominations from the Austin area, asking each doctor to nominate up to three physicians in their field whom they would trust with the medical care of their own family. The top-rated physicians from over 50 medical fields are then verified to be in good standing with the appropriate medical boards.

We are pleased that nine pediatric neuroscience physicians are featured in *Austin Monthly*'s most recent Top Doctors roster. We are honored that these nine are so highly regarded, although we think all our doctors are special.

Glendaliz Bosques, MD
Pediatric rehabilitation

Dave Clarke, MD
Pediatric epilepsy

Jane Edmond, MD
Pediatric neuro-ophthalmology

Daniel Freedman, DO
Epilepsy and functional disorders

Louisa Keith, MD
General neurology, NICU and cardiac follow-up

Sara Pavitt, MD
Headache in children

E. Steve Roach, MD
Genetic disorders, stroke

Elizabeth Tyler-Kabara, MD, PhD
Pediatric neurosurgery

Veda Vedanarayanan, MD
Neuromuscular disorders

Verma Awarded Resident Teacher of the Year

Khushboo Verma, MBBS, at the neurology residency graduation ceremony with pediatric neuroscience faculty mentors Manikum Moodley, MD (L) and Stephen Deputy, MD (R)

Former child neurology resident **Khushboo Verma, MBBS**, was named Resident Teacher of the Year at Dell Medical School's neurology residency graduation ceremony. The award is based on medical students' evaluations of the residents who taught them during their neurology rotation. Verma was chief resident and complemented her child neurology training with additional training in adult behavioral neurology. She is now a behavioral neurology fellow at Stanford University.

FIREFLY FUND KICKS OFF CENTER FOR RARE DISEASE

The Andrews family

The Firefly Fund recently made an initial donation to Dell Medical School to launch the Center for Rare Disease, the first of its kind in the Austin area. The center will facilitate research and provide support and therapies for patients and families affected by rare disease.

Established in 2017 by Pam and Chris Andrews, the Firefly Fund is an Austin-based nonprofit whose mission is to fund and support the research and education needed to accelerate cures for rare neurodegenerative genetic diseases that affect children. The gift honors the Andrews' two daughters who have Niemann-Pick type C.

The developing center is led by Stephen Ekker, PhD, associate dean of innovation and entrepreneurship at Dell Medical School and associate vice president for research translation at UT. It will be housed within the Dell Pediatrics Research Institute, directed by Department of Pediatrics chair Z. Leah Harris, MD, who has been part of the Andrews' care team since 2020. The Pediatric Neurosciences Program, which was involved in conceptualizing the center with Harris, will remain involved in the planning stages and beyond. The Pediatric Neurogenetics Center will play a key role in the center's collaborative care and research initiatives, including clinical trials.

"The Center for Rare Disease will provide hope to children and parents for years to come," said Dell Medical School dean Claudia Lucchinetti, MD. "By investing in cutting-edge research, we aim to support patients and their families as we find new ways to treat and, one day, hope to cure many of these diseases."

DELL CHILDREN'S HOSTS FIFTH ANNUAL PRACTICAL PEDIATRIC NEUROSCIENCE SYMPOSIUM

Leah Ferrante, MD, co-director of the Dell Children's Neuro NICU program, presenting on neurodevelopmental outcomes and management in prematurity

The fifth annual Practical Pediatric Neuroscience Symposium was held on May 18 at Dell Children's Medical Center. The hybrid event featured presentations by 11 pediatric neurosciences faculty members, with topics ranging from concussion to emerging gene therapies for neuromuscular disease. Each interactive presentation provided techniques and evidence-based recommendations for diagnosis and management, including the roles of interprofessional teams in improving quality of life and treatment outcomes. The symposium is designed to enhance knowledge of pediatric neurological conditions among general pediatric practitioners. The presentations are available to watch online.

PROGRAM HOSTS RENOWNED NEUROSURGEON DOMINIC THOMPSON AS VISITING PROFESSOR

Dominic Thompson, MBBS BSc FRCS, presenting a grand rounds lecture

This February, the Department of Neurosurgery hosted visiting professor Dominic Thompson, MBBS BSc FRCS, senior pediatric neurosurgery consultant and honorary senior lecturer at the Great Ormond Street Institute of Child Health in London. During his professorship, Thompson presented a fascinating Neurosurgery Grand Rounds lecture on the psychology, science, and dogma of spinal cord lipomas.

Thompson was a mentor to Dell Medical School pediatric neurosurgeon **M. Omar Iqbal, MD**, during his residency at Great Ormond Street Hospital for Children (GOSH). Thompson is internationally recognized for his research in abnormalities of the pediatric spinal cord, including spinal dysraphism and craniovertebral junction anomalies. Thompson launched the United Kingdom's first fetal surgery program for spina bifida and leads clinics at GOSH for the management of spinal dysraphism and the evaluation of children with moyamoya disease. He is a fellow of the Royal College of Surgeons and a member of the British Paediatric Neurosurgery Group, the European Society for Pediatric Neurosurgery, the International Society for Pediatric Neurosurgery, and the Society of British Neurological Surgeons. He has been a neurosurgery consultant at GOSH for over 25 years.

A SECOND OPINION: EPILOGUE

PFOs occur in an estimated 25% of the population. While cerebrovascular complications can result from paradoxical embolism, the vast majority of individuals with a PFO remain

asymptomatic. In contrast to complicated migraine, transient ischemic attacks are not typically associated with headache, and it would be unusual to experience numerous ischemic attacks without a trace on subsequent MRI scans. Although the evidence is not compelling, some physicians suggest PFO closure to reduce the severity of migraine. This child's continued episodes following his PFO closure are not supportive of either narrative.

The key to this child's diagnosis was his strong family history of headache and his mother's similar episodes as a young woman. Although hemiplegia is seemingly required for the diagnosis of familial hemiplegic migraine, the singular focus on hemiplegia may be counterproductive as affected individuals can experience a variety of other neurological deficits, sometimes in the absence of hemiplegia.

At least three types of familial hemiplegic migraine (commonly designated as FHM1, FHM2, and FHM3) have been linked to three distinct gene loci. FHM1 accounts for about half of the patients with familial hemiplegic migraine; it results from a pathogenic variant of the *CACNA1A* gene coding for an alpha subunit of the calcium channel. FHM2 accounts for another 25% of the patients; it is associated with a pathogenic variant of the *ATP1A2* gene. FHM3 is caused by a pathogenic variant of the *SCN1A* gene.

However, this child's hemiplegic migraine gene panel revealed a pathogenic variant in the *PRRT2* gene on chromosome 16. *PRRT2* mutations classically lead to paroxysmal kinesigenic dyskinesia or benign infantile convulsions. However, *PRRT2* variants are occasionally identified in individuals with episodic ataxia or hemiplegic migraine.

Commonly used migraine prophylactic medications tend to be ineffective in individuals with familial hemiplegic migraine, although daily administration of a calcium channel blocking agent may be useful. Patients with *PRRT2*-related paroxysmal kinesigenic dyskinesia usually respond dramatically to a low daily dose of carbamazepine or oxcarbazepine. These agents can also be effective for the *PRRT2* hemiplegic migraine phenotype.

Final Diagnosis: familial hemiplegic migraine due to a *PRRT2* pathogenic variant

Additional Reading

1. Larrosa D, Ramon C, Alvarez R, et al. No relationship between patent foramen ovale and migraine frequency. *Headache*. 2016;56:1466-1473.
2. Landolfi A, Barone P, Erro R. The spectrum of PRRT2-associated disorders: update on clinical features and pathophysiology. *Front Neurol*. 2021;12:629747.
3. Riant F, Roos C, Roubertie A, et al. Hemiplegic migraine associated with PRRT2 variations: a clinical and genetic study. *Neurology*. 2022;98:e51-e61.
4. Suzuki-Muromoto S, Kosaki R, Kosaki K, et al. Familial hemiplegic migraine with a PRRT2 mutation: phenotypic variations and carbamazepine efficacy. *Brain Dev*. 2020;42:293-297.

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UT Health Austin Pediatric Neurosciences at Dell Children's is a clinical partnership between Dell Children's Medical Center and UT Health Austin, the clinical practice of Dell Medical School at The University of Texas at Austin.

For additional program information:

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