

## About the Brain Care Institute

The Brain Care Institute (BCI) at Children's Hospital of Pittsburgh of UPMC is dedicated to developing innovative treatments and approaches for infants, children, and teens with disorders of and injuries to the brain, spinal cord, muscles, and nerves. The Brain Care Institute is home to more than 200 physicians, nurses, and staff who treat more than 14,000 patients every year — patients who come to us from across the country and around the world. The BCI team of world-class specialists and physician-scientists continually strive to break new ground in the diagnosis, treatment, and research of pediatric neurological disorders, and at the same time train the next generation of specialists to accomplish the same goals.

Affiliated with the University of Pittsburgh School of Medicine and ranked among the nation's best children's hospitals by *U.S. News & World Report*.

## Diagnosing Complex Conditions: The Neurogenetics Clinic



The Neurogenetics Clinic seeks to provide diagnostics and clinical care to children and families with suspected neurogenetic disorders. **Deepa Rajan, MD**, co-directs a multidisciplinary program, which includes aspects of neurology, neurogenetics, and genetic counseling for children and their families wrestling with undiagnosed and complex neurological and neurodevelopmental disorders.

The Neurogenetics Clinic at first operated once a month seeing patients and families; however, over the intervening two years, the scope and frequency of clinics has increased substantially, now with one clinic per week.

"The clinic's goal is to leverage technological advances in genomics to help with finding possible diagnoses and to direct individualized clinical management for patients and families who have complex, often undiagnosed neurological disorders. Many of these families have often spent many months or years negotiating a diagnostic odyssey in an effort to find a unifying diagnosis," says Dr. Rajan.

Dr. Rajan and colleagues meet as a team each week prior to clinic to discuss the incoming cases and prepare for what will be needed in terms of evaluation, follow-up, testing, and counseling. This approach facilitates a more efficient clinic process, and it allows the clinical team the ability to discuss aspects of the case in advance.

For some patients, a clear-cut diagnosis is possible. These individuals can then be provided or directed to the right resources. For others, genetic findings may not be so definitive, or they may point to a rare condition with no current standards of care. This is where the second aspect of the Neurogenetics Clinic comes in to view.

### Neurogenetics Clinic Team Members

Deepa Rajan, MD  
 Uta Lichter-Konecki, MD, PhD  
 Alexis Franks, MD  
 Jenna M. Gaesser, MD  
 Michelle Morrow, PhD, LCGC  
 Joshua Barch, MS  
 Jodie M. Vento, MGC, LCGC

For these cases, Dr. Rajan and her clinic colleagues are able to connect patients and families to researchers and clinicians with expertise, or potential research opportunities, in rare disorders, as well as referrals into many of the cutting-edge translational pathways that exist at Children's Hospital and the University of Pittsburgh. "Even if we do not have a specific treatment available, our job in the clinic is to walk with our patients and families through the journey, providing whatever support and services we can, regardless of the diagnosis or prognosis," says Dr. Rajan.

# The EEG Technologist Program at Children's Hospital

A national shortage exists of trained, accredited EEG technologists. In past years, there was a critical shortage in western Pennsylvania, where Children's Hospital resides. The shortage was, and is, mainly the product of a lack of schools able or willing to offer EEG technology training programs. The discipline itself is also not as well-known as other medical technologies.



The EEG Technologist Program operated by Children's Hospital was conceived and began operation in 2007 specifically to address the ongoing shortage of available EEG

technologists. **Russell Phillips, MA, REEGT**, clinical coordinator of the Electroneurodiagnostic Technology Program coordinates and manages the EEG Technologist Program training, and has done so since its inception. Through the program, Mr. Phillips, who is an accredited EEG technologist himself with decades of experience, has trained nearly 30 individuals, most of whom have remained at one of the many hospitals within the UPMC system.

## Program Details and Training Curriculum

The EEG Technologist Program is a one-year, full-time program in which students work in a hands-on manner alongside their teachers and the neurologists and neurosurgeons from the Children's Hospital Brain Care Institute. The program functions in an apprenticeship-type manner, in which students work full-time and learn the skills and expertise needed to successfully become accredited EEG technologists. The program is able to train up to four individuals in any given year.

"What sets our program apart is that students are on-site, working eight hours

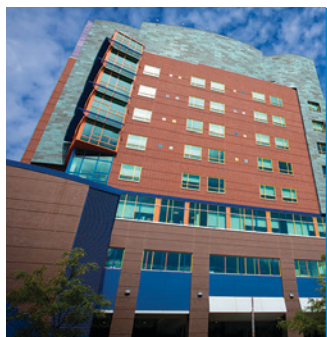
a day, participating in EEGs virtually from the moment they begin training. Students shadow our accredited technologists, learning as they go in a real-world work environment. Didactic learning is part of the program, of course, and occurs twice a week, but the integrated training with our experienced technologists and epilepsy specialists is where our program stands out amongst others. There are few places in the country where EEG trainees can work alongside the clinicians of an internationally respected hospital such as we have here in Pittsburgh," says Mr. Phillips.

Students work daylight shifts during the first six months of the program and then transition to second and third shift work, as the hospital requires EEG coverage 24 hours a day. Initially, students shadow techs until they obtain enough training and knowledge to start head measurements, the application of electrodes, and running tests independently. Students need to have a clear understanding of what montages to run and what procedures to perform to best bring out electrical activity that will help the electroencephalographers to adequately interpret test results.

## What Makes for a Good EEG Technologist?



While most of the students in the program have a science or neuroscience background or degree, the program is open to anyone who is interested in becoming an EEG technologist. Individuals with science backgrounds are preferred because many of the concepts and basic scientific knowledge has already been acquired during their previous education. However, science degrees are not a mandatory prerequisite for interested candidates. Both Mr. Phillips and **Christina Patterson, MD**, director of epilepsy and the epilepsy monitoring unit at Children's Hospital, emphasize that there are two characteristics apart from all others that can predict someone's success, not only in being accepted in the program, but excelling as a technologist. Those characteristics are empathy and compassion. The ability to translate both of these attributes to the dynamics of working with patients and their families is a critical skill. "Students who are pursuing a career as an EEG technologist because they intrinsically want to help care for other people are going to be most successful. Especially with children, that level of connection and empathy is critical. We look for it when screening candidates, and we also help to teach and reinforce it while they are in training," says Dr. Patterson.



## About Children's Hospital of Pittsburgh of UPMC

Regionally, nationally, and globally, Children's Hospital of Pittsburgh of UPMC is a leader in the treatment of childhood conditions and diseases, a pioneer in the development of new and improved therapies, and a top educator of the next generation of pediatricians and pediatric subspecialists. With generous community support, Children's Hospital has fulfilled this mission since its founding in 1890. Children's is named consistently to several elite lists of pediatric hospitals, including ranking No. 9 in the prestigious *U.S. News & World Report* annual Honor Roll of America's Best Children's Hospitals for 2017–2018 and ranking 10th among children's hospitals and schools of medicine in funding for pediatric research provided by the National Institutes of Health (FY2016).

## New Faculty Profile: Jenna M. Gaesser, MD



**Jenna M. Gaesser, MD**, assistant professor of pediatrics, joined the Division of Neurology at Children's Hospital in July 2017. A fellowship-trained pediatric neurologist with specialties in neurogenetics and white matter neurodevelopmental disorders, Dr. Gaesser completed both her pediatric residency (2013) and her neurology fellowship (2017) at Children's Hospital after obtaining her medical degree from SUNY Upstate Medical University in Rochester, New York.

### Clinical Responsibilities

Dr. Gaesser's current clinical responsibilities include coverage in the Brain Care Institute's Neurofibromatosis Clinic, the multidisciplinary Spasticity Clinic, and the Neurogenetics Clinic, among other clinical duties that include following NICU graduates who may have white matter injuries due to issues of prematurity, and that may be portend future spasticity or neurodevelopmental delays or disorders. "NICU graduates are at significant risk for underlying neurodevelopmental disabilities, especially those born after less than 30-weeks gestation. While advances in neonatology have greatly increased survival rates for these infants, there exist high levels of neurologic morbidity and risk for structural white matter injury," says Dr. Gaesser.

Working in the Neurogenetics Clinic co-directed by Deepa Rajan, MD, Dr. Gaesser sees children who have global developmental delays manifesting in a range of deficits but who do not have a definitive diagnosis for their condition. "The goal of the Neurogenetics Clinic is to uncover a basis for these individuals' conditions through metabolic and genetic testing in order to target treatments and counsel patients and families on what the findings are. An offshoot of this is my work in the Neurofibromatosis Clinic. As neurofibromatosis is a condition with a neurogenetic basis, and also one that affects white matter in the brain by overproducing myelin, it is a natural fit for me to see patients here because

these are two areas I have a concentration in," says Dr. Gaesser. Children with neurofibromatosis are at risk for gliomas, abnormal myelin formation, or dysmyelination, which can lead to severe developmental disabilities, autism, and epilepsy. "Being part of this clinic and working alongside the other practitioners, nurses, and social workers, I'm involved in treating many of the learning disabilities, behavioral challenges, issues with sleep, and other factors that these patients and their families must negotiate, both at home and at school."

Seeing patients in the Spasticity and Movement Disorders Clinic is a multidisciplinary affair. Dr. Gaesser is working alongside pediatric neurosurgeon Elizabeth Tyler-Kabara, MD, PhD, and Ozgur Dede, MD, who is a pediatric orthopaedic surgeon, as well as members from physical medicine and rehabilitation and social work, in a medical home-type model of care for, among others, neurologically complex children who have spasticity as a major concern secondary to developmental brain injuries and traumatic brain injuries, and those children with brain malformations.

### Research Priorities and Interests

From a research perspective, Dr. Gaesser is currently involved in translational studies related to neurodevelopmental outcomes in children with congenital heart disease, where she is working with colleagues Ashok Panigrahy, MD, and Cecilia Lo, MD, in the study of cilia in relation to the genetic underpinnings of congenital heart disease and related neurodevelopmental disorders. "Cilia in the brain help to circulate cerebral

spinal fluid, and studies are also implicating them in their ability to help neurons migrate and travel to their final destinations throughout the development of a child's brain. Abnormal cilia in a developing fetus may contribute to abnormal heart and brain formation. The studies I'm working on with Drs. Panigrahy and Lo, and others here at the Brain Care Institute involve the use of radiologic biomarkers and MRI to assess both structure and connectivity to determine if there is a link between abnormal findings and neurodevelopmental outcomes," says Dr. Gaesser.

### References and Further Reading

Below are references to research and past work of Dr. Gaesser discussed in this article.

- 1 Panigrahy A, Lee V, Ceschin R, et al. Brain Dysplasia Associated With Ciliary Dysfunction in Infants With Congenital Heart Disease. *J Pediatr*. 2016; 178: 141-148.
- 2 Gaesser JM, Fyffe-Maricich SL. Intracellular Signalling Pathway Regulation of Myelination and Remyelination in the CNS. *Exp Neurol*. 2016 Sep; 283(Pt B): 501-11. doi: 10.1016/j.expneurol.2016.03.008. Epub 2016 Mar 5.
- 3 Gaesser J, Votava-Smith J, Reynolds WT, Lee V, Lorenzi L, O'Neil S, Paquette L, Schmithorst VJ, Lo CW; Panigrahy A. Term Congenital Heart Disease Infants With Brain Dysplasia Show Subcortical-Related Altered White Matter Connectivity and Poor Short-Term Neurodevelopmental Outcome. Poster Presentation, Pediatric Academic Society May 2017.

## Diagnosing Complex Conditions: The Neurogenetics Clinic *(Continued from Page 1)*

### Types of Cases

The clinic sees a broad spectrum of diseases including, but not limited to, genetic epilepsies, developmental disorders, movement disorders, and mitochondrial disorders. These cases are largely ones with either atypical manifestations for which an underlying genetic issue may be contributing to, or typical manifestations of a rare disorder in an age group or clinical background for which there is a high level of suspicion for a genetic cause.

### Genetic Counseling: A Foundational Element of the Neurogenetics Clinic



**Jodie Vento, MGC, LCGC**, manager, Center for Rare Disease Therapy, the Brain Care

Crucially important to the diagnostic process and care within the clinic are the roles and responsibilities of the genetic counselors. "Pre- and post-test counseling are vitally important," says

Institute, and genetic counseling supervisor, Laboratory Services. "Helping families understand the complexities in front of them, the options available with testing, what the testing may or may not uncover, and understanding from them what and how much they actually want to know must be done in a compassionate and clear manner." **Michelle Morrow, PhD, LCGC**, was recently hired to coordinate the neurogenetics clinical and research programs. "As a genetic counselor with a doctorate in Neuroscience, Michelle is uniquely poised to help to lead this effort."

Beyond the frontline genetic testing, when moving into large multigene panel tests or whole exome sequencing, these tests can uncover findings that may or may not be related to the neurological condition, or that were completely unexpected, or that have implications for the broader family, immediate or otherwise. For example, whole exome sequencing may uncover disease-causing BRCA gene variants related to breast cancer, or genetic findings related to an inheritable

cardiac condition. The counseling aspect with family members in this regard, both in the pre- and post-test environment, is supported by referrals for follow-up testing, helping families assess the risk and consequences, and generally serving as an advocate for the patient and family.

As the clinic has expanded services and grown in frequency, more resources have been added to fill the growing clinical and counseling needs. This will likely continue to occur as the science and technology of genetic testing advances at a pace that sometimes exceeds the ability of clinicians and researchers to interpret and understand the findings and associations uncovered.

### Referrals

For physician referrals, please contact the Neurogenetics Clinic at **412-692-5520**.