

An aerial photograph of the University of Texas Health at Texas Medical Center in Houston, Texas, taken during sunset. The sky is a mix of orange, yellow, and blue. Several tall skyscrapers are visible, including the distinctive cylindrical tower of the UTHealth Tower. In the foreground, there are lower-rise buildings, parking lots, and a curved highway with a few cars. The overall scene is a mix of urban architecture and greenery.

University of Texas Health at Texas Medical Center

TSC Newsletter

Summer 2021

Featuring-

Staff Spotlight

STOP2 Research

Meet Zoey

TSC International Conference

TSC Golf Tournament

International Tuberous Sclerosis Research Conference 2021

Newborn Screening for TSC *Dr. Hope Northrup*

In her presentation at the International Tuberous Sclerosis Research conference this year, Dr. Northrup explored the possibility of including TSC on the newborn screen (NBS). In the United States, all babies receive the NBS at birth via a heel stick. The goal of NBS is early detection, intervention, and treatment. For other conditions on the newborn screen, early diagnosis and intervention can allow for better patient outcomes. During her presentation, Dr. Northrup explained the process for doing newborn screening for TSC, possible hurdles that may be faced adding TSC to the NBS, as well as provide examples of NBS success stories. Dr. Northrup's hopes that all babies born in the United States will one day be screened for TSC using a cost effective, but accurate screening method.

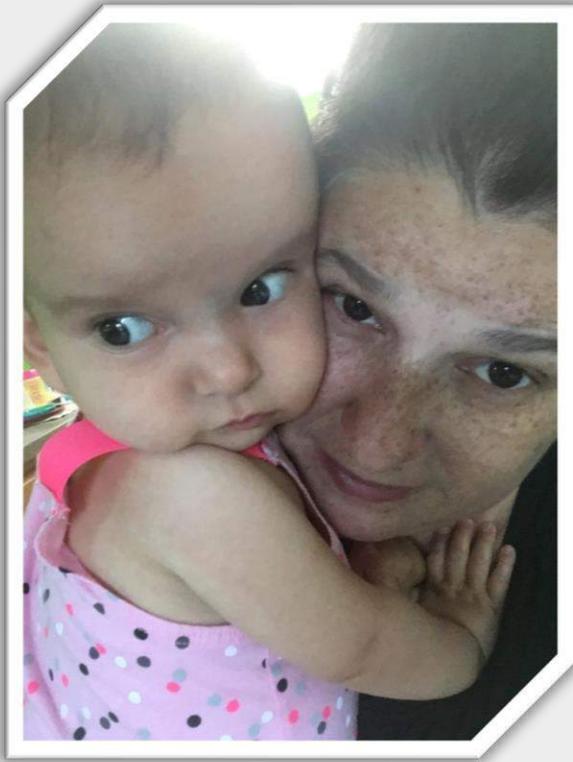
mTOR Inhibitors and Pancreatic Neuroendocrine Tumors *Kate Mowrey, MS, CGC*

Kate Mowrey, a genetic counselor for the UTHealth TSC Center, gave a rapid-fire poster presentation about her research with Dr. David Rodriguez-Buritica (geneticist and endocrinologist) regarding management and treatment of non-functional pancreatic neuroendocrine tumors (PNETs). A PNET is a tumor in the pancreas and a rare clinical symptom of TSC. Data was collected from Cincinnati Children's Hospital TSC Database and the TSC Alliance's Natural History Database regarding PNET characteristics, tracked growth of the PNETs over time, and if an individual was taking or not taking an mTOR inhibitor. She notes that this data provided evidence that mTOR inhibitors reduces the rate of growth for PNETs and may offer a primary treatment method over invasive surgery.

The Epilepsy Genetic Risk Model for TSC Patients *Dr. Laura Farach*

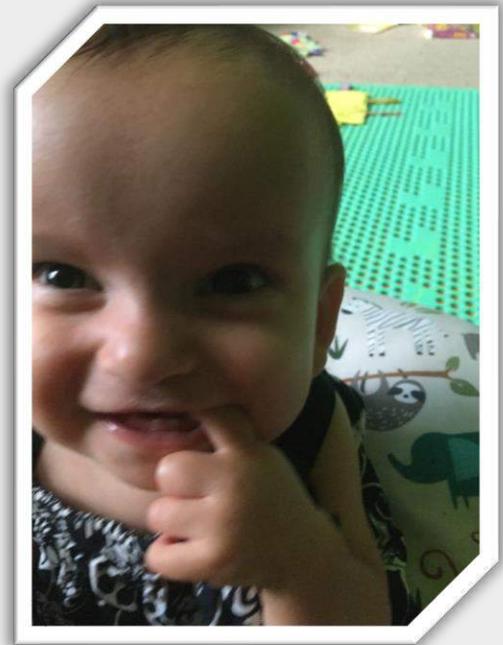
Dr. Laura Farach gave her presentation on her ongoing research project, in which genetic modifier genes are identified for tuberous sclerosis complex patients with epilepsy. She is analyzing genetic data to identify biomarkers that may help predict which patients will develop seizures. Early control of seizures has been shown to provide for better outcomes for patients, and Dr. Farach aims to develop a clinical risk model using the genetic data she received to provide better outcomes for patients moving forward. In doing so, this data may also provide her with information on what kinds of seizure medications work best with which biomarkers.

STOP2 Research & Meet Zoey



Sirolimus (rapamycin) is an mTOR inhibitor that is now widely used to change the course of tuberous sclerosis complex. Sirolimus is used to treat kidney and brain tumors, and now is a primary treatment for epilepsy. Epilepsy affects between 80-90% of individuals with TSC and often start to occur before an individual turns 1 year of age. When seizures are uncontrolled, it can lead to developmental delay. Until recently, sirolimus has only ever been used after an individual experiences a seizure. STOP2 is a research project aimed to proactively prevent seizure activity in infants with TSC using sirolimus, in hopes that this early treatment will be effective for preventing developmental and cognitive delay. Although Phase 1 of this research study is being carried out in Cincinnati, Ohio under Dr. Darcy Krueger, Phase 2 will be taking place at our TSC Center of Excellence under the guidance of our very own Dr. Hope Northrup.

When diagnosed with a disease many have never heard of, it can be easy to fall down the rabbit hole expecting the worst possible outcomes. After Zoey's tuberous sclerosis diagnosis, her parents found the STOP2 study to be a beacon of hope for their daughter and possibly for future children and families diagnosed with TSC. Her mother, Kelly, describes Zoey as being small but mighty! Although she was born at around 5 pounds, Zoey's mother says she gets bigger and stronger every day. At eight weeks old, Zoey and her family made their first visit to Cincinnati where Zoey had an EEG done to determine whether she would be eligible for the STOP2 study. When no seizure activity was found, she was enrolled in the research project and began taking the study medication twice a day. The past year has been hard for all of us in many ways, but the difficulty of traveling during a pandemic with a baby has been made worth it for Zoey's family by their hopes that this study can contribute to future infants diagnosed with TSC. Kelly explains that her decision to enroll her daughter in this study was an effort to be proactive rather than reactive and to provide her daughter with the best possible care. At 12 months old, Zoey has never experienced any side effects from the study drug and her mother reports that she has never had a seizure.



Meet the Staff!

Saba Usmani – Research Coordinator

Saba Usmani joined the TSC Center of Excellence in April of this year. In her role as research coordinator, Saba conducts patient interviews, collects patient data, and performs the administrative and logistical tasks for the TSC Center's research projects. Saba enjoys making a difference in the lives of her patients and their families and is looking forward to getting better acquainted with everyone at UTHealth.

Alma Torres – Medical Assistant

Alma Torres has been with the TSC Center of Excellence since April of 2020. As the center's medical assistant, Alma makes sure that patient needs are met by conducting patient screening, scheduling appointments, and ordering labs. Alma is passionate about facilitating patient care and aims to continue learning and improving within her role at UTHealth.

Claire Sartwell – Nurse Coordinator

Claire Sartwell has been the TSC Center's nurse coordinator for nearly three years now. Claire plays a pivotal role in keeping the TSC clinic running smoothly by confirming information for new patient appointments, organizing patient visits, and ordering patient medication. Claire enjoys interacting with the TSC clinic staff and faculty and is dedicated to improving patient care.



**Join us for the 10th Annual
TSC Golf Tournament on
Friday, October 22, 2021!**