The Center for the Treatment of Pediatric Neurodegenerative Disease (CTPND), formerly UT Mitochondrial Center of Excellence, received its official Center designation in the Fall of 2020. Since the establishment of UT Mitochondrial Center of Excellence in 2007, the program has grown to include other sub-specialties, including Movement Disorders, and our Clinical Research Program has undergone exponential expansion. CTPND is the umbrella descriptor for all that we do, including, but not limited to: Leigh clinic, Mitochondrial Disease Clinic, Ataxia-Telangiectasia Clinic, and Clinical Research.

WELCOME WILLIAM GUERRA, SENIOR PROGRAM MANAGER-CLINICAL TRIALS PROGRAM

This year, the most notable change in staffing was the addition of William Guerra to oversee our Clinical Trials Program, a position that has been vacant since Fall 2020.

RESEARCH CORNER by William Guerra

The future of Research looks bright for the CTPND and Department of Pediatrics at UTHealth Houston. With eleven (11) Research Studies at different stages of development, we continue to provide cutting edge therapeutic investigational options to our patients.

2022 has been a year of tremendous growth for research activities in the CTPND. I joined the team January 2022 as Research Program Manager for the Division of Child and Adolescent Neurology. I have been able to participate in the meaningful and innovative work performed by CTPND team of investigators and research staff. Not only have we been able to continue enrolling subjects at a higher pace than other academic centers, but we have also successfully activated seven new clinical trials since January.

In addition, the Department of Pediatrics at UTHealth, has adopted our clinical research operations model in order to centralize efforts by utilizing our resources for the benefit of patients.

Continued on page 11
In June, members of the CTPND traveled to the UMDF annual symposium in Phoenix, Arizona. Dr. Koenig, Dr. Russo, Research Director William, Social worker Patricia, and Research Nurse Lindsey were all in attendance. CTPND members were able to network with other providers and industry sponsors, share our research findings, provide patient support, and even learn a thing or two!

Research Director William and Drs. Koenig and Russo met with industry sponsors to discuss upcoming clinical trials, as well as trials that are already in progress. They were able to provide feedback that we have received from study participants, with the overall goal of improving the research subject experience. They were also able to give industry sponsors a glimpse into the everyday operations of our well-oiled machine. We are looking forward to seeing how this feedback is translated into upcoming clinical trials. Stay tuned!

Dr. Koenig participated in two different LEIGH syndrome talks, LEIGH/Management of Care and LEIGH/ How to Support your Leigh Syndrome Patient. Two “The Doc Is In” sessions, where patients were able to sit down with her one on one and ask questions. Dr. Koenig also participated in the adult “Ask the Mito Doc Panel.”

On Display were two abstract posters co-authored by Dr. Koenig, Dr. Russo, and Lindsey. They highlighted the positive outcomes seen with the use of the investigational drug Elamipretide, under the Expanded Access Program (EAP). Elamipretide, is also being used in another clinical trial at the CTPND called SPIMI-D for patients with primary mitochondrial disease associated with nuclear DNA mutations.

Our wonderful social worker Patricia participated in a special session for newly diagnosed patients.

We look forward to sharing more big things next year in Charlotte, NC at the 2023 UMDF symposium!
Dr. Stuart Fraser, former co-Chief Resident of the Child & Adolescent Neurology Residency Program at UTHealth (with our own Dr. Russo), joined our Division faculty in July 2022, after completion of his one-year ACGME-accredited Vascular Neurology Fellowship at UTHealth. As newly-minted Director of our institution’s Pediatric Stroke Program, he is one of only a handful of pediatric vascular neurologists in the country. Primarily, his time is allocated to research, though he sees pediatric patients in UT clinic on Wednesdays and patients with cerebrovascular disease through the pediatric stroke consultation service.

Fun facts about Dr. Stuart Fraser:
- Attended UT McGovern Medical School on a full academic scholarship, after completing his undergraduate at NYU with a major in biochemistry, a minor in mathematics, as well as receiving an associate’s degree in liberal studies.
- Dr. Fraser is the UT McGovern Principal Investigator for the International Pediatric Stroke Study, having become the first fellow to be a site PI for the study in 2020. The International Pediatric Stroke Study consists of more than 150 participating children’s hospitals worldwide who collaborate in order to improve understanding and treatment of children with cerebrovascular disease.
- Supported by a NIH grant through the Department of Anesthesia, Dr. Fraser is trying to develop new therapies for children who have suffered from cerebrovascular disease.
- Dr. Fraser still sees adult stroke patients through the wide-reaching UT tele-stroke service and on the inpatient stroke service in the renowned Memorial Hermann stroke service, but his passion is for improving recovery and development in our youngest stroke survivors.

Cerebrovascular Nurse Joins Pediatric Stroke Program

Melika Abrahams, RN, BSN, joined the CTPND as Dr. Fraser’s clinical and research nurse. Melika graduated from Texas State University with a Bachelor of Science in Nursing. She comes to us with 8 years of experience at Children’s Memorial Hermann Hospital, and was a graduate of their Nursing Fellowship Program. As the new pediatric cerebrovascular nurse, Melika’s role is to act as a resource to our families, faculty and staff. Within the pediatric stroke team, Melika helps provide personalized care to ensure timely access to care and interventions as well as provide education and support to our patients and families.
What’s harder than living with or loving someone who has a chronic illness? Doing so while trying to navigate a healthcare system designed to support profit and not people. The coordination of complex medical care consumes an amount of energy equivalent to a full-time job: scheduling and attending appointments, managing benefits and treatment recommendations, and preparing for future concerns are just a few tasks on the compulsory to-do list. Overwhelmed by so many logistical demands, any attention to emotional welfare is usually deemed a low priority – for everyone involved. Balancing the need to act with the destabilization of grief; fighting between having too much to qualify and not enough to make it; waiting on lists until it’s your turn to get what you need; piecemealing a life plan together without many reliable systems of community support... to add any meaningful processing of emotions is just too much for most.

The consequent denial and frustration we often encounter at the CTPND are more than understandable – they are expected and protective reactions against significant, life-altering threat. It is reasonable to find anger where a person recognizes that by their relationship to disease, they’ve been unwillingly cast in a role they never anticipated and cheated of the chance to just simply be and exist without intricacy. But ‘normal’ only counts as a setting on your washing machine, and you didn’t miss your copy of How to Disability in our Ableist World... because no such manual exists. The human ego - too fragile to withstand any recognition of mortality – catered our world to idolize the seemingly invulnerable and able-bodied. Assumptions of health disregard the experiences of persons affected by mitochondrial disease and their families – despite estimates that every 30 minutes a child is born who will develop a mitochondrial disorder by age 10 (source). I want to provide reassurance that you are not alone in your struggle, despite the isolation of having ‘special’ needs. I want to acknowledge that even the use of the word ‘special’ as a qualifier can be attributed to society’s compulsive “othering” of certain human needs, and how I wish society would instead seek to understand and embrace need as a natural part of every person’s life. I want to recognize that your needs neither diminish nor increase your value and worthiness, and are only a component of all that is YOU. I want to encourage you and support your efforts to identify, express, and address your needs – both seen and unseen.

I want these things for many reasons, but primarily because your emotional health has a direct consequence on both your individual well-being and the welfare of your entire family. Caring for someone who has complex health care needs inevitably causes extreme stress, especially if that someone is yourself! When that kind of stress is left unmitigated, the resulting anxiety can negatively affect the health outcomes of the entire family system – and can be particularly harmful to the person who needs specialized health care support. By denying what we need as caregivers in the name of giving more care, we, in fact, harm our care receivers. The emotional welfare and mindset of a person engaged in health care influences their treatment outcomes – for their betterment or to their detriment. By prioritizing medical needs and leaving feelings for future follow-up, we neglect the only patient we’ve got – the patient that is here and now. Denying the importance of emotions when faced with physical pain does not, ultimately, have the desired effect of lessening discomfort – it gives grief an opportunity to build self-interest.

Continued on page 11
**Expanded Access Program (EAP)**

**by: Lindsey Miller, LVN**

An EAP is a way for a patient, with serious or life-threatening disease, to gain access to investigational medications. The patient must not qualify for an ongoing clinical trial nor have any comparable treatment options available. Investigational drugs have not (yet) been approved by the FDA for the specific use necessary for the patient. Expanded access can be used for individual patients, including for emergency use, an intermediate size group of patients, or widespread treatment.

**How does an EAP get started?**

First, a patient and physician discuss if the investigational medication could have potential to be effective. The physician must ask the drug company for permission to use the drug. The drug company then provides a letter of authorization to the physician. At this point we are ready to present our case to the Food and Drug Administration (FDA) and to the Internal Review Board (IRB). These are the governing bodies that give the green light, indicating “your plan to conduct research and give this medication is safe and ethical.” After we receive the “all clear” from the FDA and the IRB, it’s go time.

The patient will then sign a consent stating that they understand all the risk and benefits of the medication, who to contact in an emergency, and other legal stuff. The drug company will then provide the drug at NO COST to the patient, but the physician’s, research nurse, and study coordinator’s time is not covered. The medical staff donates their time to file all necessary paperwork, working with the patients, scheduling drug shipments, etc. A way we try to offset cost is by scheduling the patient to be seen in clinic during a regularly scheduled appointment. This way the cost of the exam and any other test needing to be performed for safety reasons will be covered by the patient’s insurance, with the hopes the patient will not have to pay out of pocket. From start to first enrollment, it can take approximately 1-3 months to receive approval to start in EAP.

**What if there is an emergent need to begin the investigational drug?**

If an EAP is needed in an emergency, we can obtain approval in as little as 72 hours, depending on the status of the patient. This is an “all hands-on deck” situation. Communications start flying between members of the CTPND, the drug company (sponsor), the FDA and IRB. We work around the clock communicating to keep the process moving. Once a patient is enrolled the research coordinator/research nurse continue to work with the patient, submitting reports and documents through the lifetime of the study.

The CTPND absorbs most of the costs of EAPs (in staff and supplies), but the potential success of the treatment for our patients, and progress in medical science, make it worthwhile.

--

In a medical team like ours, that is engaged in clinical practice, provides consult service to a hospital system, and runs clinical research trials, work in our office has light moments, but also has some very intense moments as well. In November, all of our members (with the lone exception of our valued Clinical Coordinator/lead nurse, LaKeesha Minor) gathered for a light-hearted gathering outside of work. Thankful for this team, who brings their best to the table each and every day, to help our patients and advance medical science.

#CTPND #UTHEALTH

Anyone wanting to support these or other CTPND efforts can do so by clicking on Donate to CTPND.

IN 2022, CTPND RECEIVED 5,000 POUNDS OF MEDICAL SUPPLIES, INCLUDING STUDY MEDICATIONS.
As the first dedicated full-time occupational therapist (OT) working in the CTPND, I want to introduce myself, as well as the profession of occupational therapy. Even after being a licensed occupational therapist for 9 years, when I first tell someone, “I am an OT,” many people have no idea what it is that I do. While a physical therapist may get you moving, an OT focuses on function, independence, and what is meaningful for someone to be able to do in their day-to-day lives.

My role in the CTPND, just like the profession of Occupational Therapy, is unique. Not only do I get to work with the other clinicians and patients in the clinic, but I also play a distinct role in the research process. From a research standpoint, I am the rater who is trained to perform scales for our studies. “Scales” is the word we use to describe assessments that are administered as outcome measures for a study. Many times, a physical assessment is the primary outcome measure to determine whether a drug or other intervention has a successful outcome, so it is critical that the administration of the assessment is consistent, accurate, and reliable.

Additionally, I will have the opportunity to provide 1:1 OT intervention to patients who participate in upcoming studies that will explore the efficacy of using medical devices/intervention in combination with therapy services for upper extremity recovery after pediatric stroke. In a clinical setting, I work with our physicians, performing assessments/screenings for functional and physical abilities, to assisting with decisions regarding therapy, equipment, and orthotic needs. Although my specialty is occupational therapy, I am also an advocate for all of the rehabilitation professions, including physical and speech therapy. Just like the CTPND works as a team to ensure the best outcomes for the patients that we serve, the therapy profession must also work as an interdisciplinary team in order for every patient to live their lives to the fullest. My goal is that all patients served by our Center are able to overcome any barriers that may stand in the way of them getting the rehabilitative services that they need.

This year has been one of enormous growth for the CTPND. A second mitochondrial specialist, Dr. Nick Russo, joined our team, enabling us to double our patient and research capabilities. I have also accepted the role of President of the Mitochondrial Medicine Society and continue to represent the Leigh syndrome community as a board member for the Leigh Syndrome Roadmap Project. In November I had the opportunity to travel to Australia and meet with mitochondrial clinicians and researchers, reinforcing that although we may be separated by great distance, our world is small and our community is connected. For 2023, I look forward to serving our patients and remind you that collectively, you are not rare. We stand together with you pledging to provide excellent clinical care as we advance research to ease the lives of you and your family.

Happy Holidays!

Dr. Koenig

For the beloved patients of CTPND, whose life journeys have concluded, it was truly our privilege to get to know you and your families, and provide the very best care and support we could offer.
Fall Gathering
Helen Keller is quoted as saying, “Although the world is full of suffering, it is also full of the overcoming of it.” Sometimes it’s like the suffering is all we see, all we experience, all we can expect – and we know mitochondrial disease redefines pain in so many of the lives it touches. The CTPND hopes to be a place where you and your loved ones can witness, experience, and expect the overcoming. While we cannot take any of the feelings away, we can be with you when you’re with them. To best meet you where you are, I have created a survey. Please, take a few more minutes to consider what emotional support means to you and share that VITAL health information with your CTPND care team through your response. With the benefit of your input, I hope to offer new opportunities for community support as we suffer and overcome… so be on the lookout! As your partner on this journey, I wish you and those whom you care for peace as we begin our next lap around the sun. As always, I remain available to provide help in any ways I can.

Lastly, GO ‘STROS!

Patricia Arnold, MSW, LCSW
Senior Social Worker
Patricia.Arnold@uth.tmc.edu
713-500-6205

Some of the highlights of the last 12 months include:

- Successfully obtaining FDA approval and opening an extended access program (EAP) for patients with Ataxia Telangiectasia, who would otherwise not have access to treatment options.

- Becoming the top enrolling institution for an international clinical trial for patients with Neurofibromatosis type 1.

- Expanding our research endeavors to include not only Mitochondrial disease but also other neurological disorders including; Pediatric Stroke, Tourette's syndrome, Autism, Epilepsy, Spinal Muscular Atrophy, and Neurofibromatosis Type 1.

- Growing our research and clinical staff to include a Research Support Assistant, a third Research Coordinator, a full time Occupational Therapist and a Clinical/Research Nurse to support the stroke clinic and research initiatives.

- Expanding our infrastructure to include laboratory equipment for sample processing as well as temperature-controlled chambers for investigational product storage and dispensing.

- Submitted a variety of academic research grants, including the Chan Zuckerberg Initiative (CZI) for Neurocognitive Decline in MELAS; a very extensive and ambitious collaborative grant proposal.

- Developing a “Research Club”; a semi-monthly gathering for faculty, fellows, residents and other research staff to discuss scientific topics of interest and new investigator-initiated project ideas.

- Our faculty has published several scientific articles and abstracts in world renowned medical journals.

- Faculty and research staff have had the opportunity to present case reports, describing the progress of some of our research patients, at medical conferences and symposiums including UMDF’s Mitochondrial Medicine Symposium 2022 in Phoenix, Arizona.

All of these outstanding accomplishments would not be possible without the dedication and passion of our wonderful team of faculty, researchers, nurses, and administrators. We are equally proud of our academic and scientific achievements as we are of the kind, collaborative and spirited nature of our team. It is truly the people that make our center special; a group of intellectually curious and compassionate individuals, united for a common cause.

As we look forward to 2023 and beyond, the CTPND is well-positioned to continue improving the quality and duration of life for pediatric and adult patients suffering from neurodegenerative diseases, through compassionate, innovative, and meaningful research.