

Herscot Center for TSC

at Massachusetts General Hospital

Community Newsletter

Vol. 1 No. 1, 2022

A Warm Welcome from Carol Herscot

When my child was growing up, I dreamt that one day there would be one place where Tuberous Sclerosis could be diagnosed and treated. Each individual with TSC may have different symptoms and many different specialists are needed.

Today I am excited to share with you that we now have a world-renowned center where children, teenagers, and adults are treated.

This is our first newsletter and we hope you will enjoy meeting our team: neurologists, social workers, dieticians, nephrologists, pulmonologists, dermatologists, and our wonderful staff. I welcome all of you to our first newsletter.

Warmly,
Carol Herscot

What's New at the Herscot Center?

A SNEAK PEEK INTO THE NEW HERSCOT CENTER CLINIC SPACE

The new Herscot Center clinic space will be up and running in early 2022! The new space will be located at 175 Cambridge Street on the third floor. Validated parking will conveniently be located adjacent to the building in the Charles River Plaza parking lot. The new clinic will be equipped for pre-visit vitals, phlebotomy, social work services, and more. We would like to extend a huge thank you to Carol and James Herscot for helping us with these renovations. It is our hope that these updates, will improve the comfort and functionality of the clinic space, as well as improve our overall patient experience.



Updated entryway with natural elements



New clinic room with technology that allows individuals and specialists to join the appointment virtually





Children's waiting area with coloring wall



Adult waiting area with comfortable chairs and natural light for relaxation while waiting

TSC Research Updates:

EXPLORING GENE THERAPY FOR TUBEROUS SCLEROSIS

By Xandra O. Breakefield, Ph.D.

Professor and Geneticist, Harvard Medical School and Massachusetts General Hospital

Tuberous sclerosis complex (TSC) is what is called by geneticists a “tumor suppressor syndrome” in which an affected individual inherits one defective copy of a pair of *TSC* genes. Tumor suppressor genes help to slow down cell division, repair DNA mistakes, and initiate programmed cell death. When the normal copy of the tumor suppressor gene is disrupted in susceptible cells in the body during development or later in life, tissue overgrowths occur in multiple tissues and organs that can compromise bodily functions and even be life-threatening.

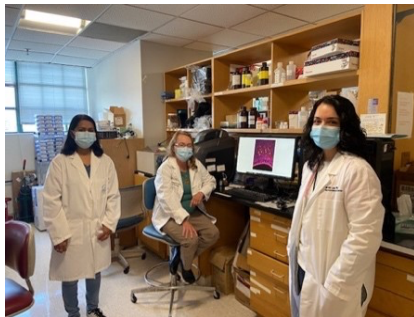
I became aware of such tumor suppressor genes at an early age, as my younger sister inherited a similar syndrome, called neurofibromatosis type 1. At that time, the gene was unknown, and the course of the disease could not be predicted. I devoted the early part of my career to finding neurologic disease genes and then to developing gene therapy to replace lost genes.

A couple years ago, I attended a talk on tuberous sclerosis at Harvard Medical School (HMS) sponsored by the Tuberous Sclerosis

Alliance. The audience was told that under someone’s chair was taped a book. It turned out to be under my chair and it was an autobiography by a young woman with lymphangiomatosis (overgrowth of lymphatic vessels), a disorder that occurs in association with TSC. It was a very moving story, and I subsequently shared the book with medical students.

My questions about TSC were answered by the many amazing scientists and physicians who study TSC in our HMS community – including Elizabeth Thiele, Vijaya Ramesh, David Kwiatkowski, Lisa Henske, Brendan Manning and Mustafa Sahin, and they have all contributed to our work.

Shilpa Prabhakar, M.S. in my laboratory has focused on mouse models of TSC1 and TSC2 for over 10 years, joined by an AAV vector expert, Casey Maguire, Ph.D. and two talented postdoctoral fellows – Pike See Cheah and Sevda Lule. (See photo of Shilpa, Xandra and Sevda viewing the distribution of a gene delivered to a mouse brain after injection of an AAV vector into the blood). We have shown in these models that gene therapy replacement of hamartin (TSC1) and tuberin (TSC2) can extend the lifespan of these mice to almost normal periods and reduce the size of lesions in the brain. These pre-clinical studies have been funded by the Department of Defense, NINDS and a biotechnology company. In a few years, we hope to fulfill federal requirements to carry out a clinical trial in TSC patients. Following the model of other clinical trials in the gene therapy field, we hope to reduce lesions throughout the body in TSC patients by a single injection of the vector into the blood.



Shilpa, Xandra and Sevda viewing distribution of a gene delivered to mouse brain after injection of an AAV vector into the blood.

We have been so moved and inspired by e-mails from the TSC community, patients, and their family members (Thank you!), and we are dedicated to better treatment for these patients.

Get to Know Our Clinicians!

CLINICIAN SPOTLIGHT SERIES: JAN PAOLINI, R.N.

Hello and welcome to our Clinician Spotlight Series! We would like to introduce you to Jan who has been a nurse with the Herscot Center for almost 17 years. Outside of work, Jan likes to play with her two rescue dogs, Channing, a chihuahua and jack-russle mix, and Kaia, a lab and pit mix. When the weather is a bit warmer out, you might find Jan in her backyard either playing with her pups or tending

to the her garden, which is filled with many different types of flowers and vegetables.

Like most people, Jan has picked up an array of hobbies and crafts during the course of the pandemic. She has enjoyed sewing, sudoku, and puzzles. Jan also loves to spend time with her family. One of her favorite activities to do with her family is to play board games together.

If you see Jan at the Herscot Center be sure to say hello!

THANK YOU DR. LAWRENCE SELTER



We would like to thank Dr. Lawrence Selter for his commitment to providing exceptional care in psychiatry to patients at the Herscot Center and greater MGH community. This fall, Dr. Selter will be retiring after over 40 years at MGH. Dr. Selter joined the Herscot Center clinic in 2015 and since then he and

his role in psychiatry have had a tremendous impact on the care of patients. Psychiatry has played an invaluable role at the Herscot Center, and we are grateful for this interdisciplinary teamwork and Dr. Selter's contribution to making this possible.

We wish him all the best and look forward to hearing all about his adventures in Italy!

Want to get involved?

THRIVE IS THRIVING

Despite the challenges that came with the pandemic, Thrive has been in full operation, serving families across New England and beyond. Thrive is run by the Herscot Center and Pediatric Epilepsy Program to promote the social and emotional wellbeing of patients and their families affected by epilepsy and other related disorders. In the past year, the switch to connecting virtually, has given many individuals the opportunity to come together who otherwise might not have met.

This year, the Thrive team has run several virtual groups catered towards different populations, including parents, kids, teens, and adults. Some of our most popular groups include the TSC Adult Group, and the Girls Hangout Group which have served as spaces for individuals to socialize, share, and connect! These groups are supervised and facilitated by many of the staff in the Herscot Center community including Leigh Horne-Mebel, LICSW, and Emma Bauman, LICSW, and Jan Paolini, RN. Other active groups include, the Middle School Boys Group, Parent Education and Share Group, the Sibling Group and the Diet Therapy Group for

Families. Furthermore, we are so excited to announce the addition of our two newest groups: a group for teens affected by TSC and a group for parents' of young children with TSC !

A few of Thrive's favorite memories have included a haunted mansion tour for Ketoween this year and painting positive affirmations on rocks with the Girls Hangout Group!

Lastly, Thrive continues to have a strong social media presence that is both interactive and engaging for those connected. On Facebook and Instagram, you can find our weekly Wellness Series that promotes self-care actives for kids and families. You will also find other content such as Tip Sheets written by Leigh Horne-Mebel, LICSW, that give advice for parents when handling pandemic related situations such as teaching little ones how to wear a mask!

Thrive is looking forward to many more events and the cultivation of new friendships! To stay updated or if you are interested in getting involved- give us a follow on social media!

