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ABOUT TGEN

Translational Genomics Research Institute (TGen) is a Phoenix, Arizona-based non-profit organization dedicated to conducting groundbreaking research with life changing results. TGen is focused on helping patients with neurological disorders, cancer, diabetes, and infectious diseases, through cutting edge translational research (the process of rapidly moving research towards patient benefit). TGen physicians and scientists work to unravel the genetic components of both common and rare complex diseases in adults and children. Working with collaborators in the scientific and medical communities literally worldwide, TGen makes a substantial contribution to help our patients through efficiency and effectiveness of the translational process. TGen is affiliated with City of Hope, a world-renowned independent research and cancer and diabetes treatment center. This precision medicine affiliation enables both institutes to complement each other in research and patient care, with City of Hope providing a significant clinical setting to advance scientific discoveries made by TGen. **www.tgen.org**

IRB Conditions Satisfied at the Study Level
Apr 24, 2019

Genetic Modifiers of Disease Severity in

TUBEROUS SCLEROSIS COMPLEX

Clinical research study conducted by TGen's Center for Rare Childhood Disorders

tgen
AN AFFILIATE OF City of Hope

WHO WE ARE

Translational Genomics Research Institute (TGen) is a non-profit organization in Phoenix, Arizona. **Dr. Vinodh Narayanan** is the Medical Director of TGen's Center for Rare Childhood Disorders (C4RCD). The Center uses next generation sequencing to diagnose and treat children with rare disorders.

OUR STUDY

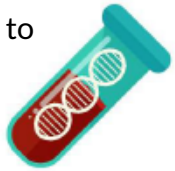
Our center is currently recruiting and enrolling families into our Tuberous Sclerosis Family Study.

We are looking for families where the parent has a mild form of the disease and the child has a severe neurological disease. In our study, we are searching for genetic modifiers of disease severity that could account for the variability in symptoms observed within single families, where affected members have the same TSC1 or TSC2 mutation. After completing a consent form, participating families will provide DNA samples (blood, cheek swab or skin biopsy, if possible) which will undergo genomic testing.

We will also collect and examine medical records and school records for each participant. Participants will be assessed by a clinical psychologist, in person or on the phone. Age-appropriate cognitive, behavioral, developmental scores will be assigned.

OUR GOAL: EARLY DIAGNOSIS

By identifying genetic differences, we hope to develop **a blood test that could predict disease severity,** so we could start treatment earlier.



WHO CAN ENROLL?

We are now enrolling parents and children of all ages that have been diagnosed with TSC. All participants must be living in the United States. Children must be severely affected with seizures, cognitive impairment, developmental delay, and/or autism. Parent must present with a mild phenotype. Unaffected parents are also encouraged to enroll, but their participation is not mandatory.

CONTACT US

For more information, please contact our clinical research coordinators at TGen's Center for Rare Childhood Disorders:

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