

Natural History Study of Individuals with Autism and Germline Heterozygous PTEN Mutations

We are enrolling individuals 18 months and older with PTEN hamartoma tumor syndrome for a new study. The goal of this study is to gain a better understanding of PTEN mutation syndromes to identify early markers and ultimately effective interventions for autism spectrum disorder.

Where is this study taking place?

Boston Children's Hospital, Cleveland Clinic, University of California at Los Angeles, Stanford University Medical Center, Cincinnati Children's Hospital Medical Center

Who are the Principal Investigators?

Mustafa Sahin, MD, PhD (Boston), Charis Eng, MD, PhD (Cleveland), Julian Martinez, MD, PhD (Los Angeles), Antonio Hardan, MD (Stanford), Darcy Krueger, MD, PhD and Jamie Capal, MD (Cincinnati)

Who is eligible to participate?

Individuals 18 months and older are eligible to participate if they have been diagnosed with PTEN hamartoma tumor syndrome or autism spectrum disorder with macrocephaly.

What will we do?

New enrollees will have three on-site visits over a two year period. For previously enrolled subjects, the study will involve at least two on-site visits over a two-year period. Each visit includes a blood draw, physical/neurological exams and behavioral testing.

What is the time commitment/cost to participate?

There is no fee to participate in this study, and there will be no financial compensation for participation in this study.

What results are provided?

Summary score reports of behavioral testing will be provided.

Contact Information

If you are interested in learning more about this study, please contact Robin Libove at rlibove@stanford.edu or (650) 736-1235.

Get Involved!

Join our RDCRN Contact Registry to learn more about new and ongoing research studies. Visit: www.rdcrn.org/dsc/registry to sign up and learn more.