

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

We are enrolling individuals between the ages of 3 and 21 years old 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

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)

Who is eligible to participate?

Individuals between the ages of 3 and 21 years are eligible to participate if they have been diagnosed with PTEN hamartoma tumor syndrome or autism spectrum disorder with macrocephaly.

What will we do?

The study involves five visits over a two year period. Three of the visits occur on-site at a study location. The other two visits occur as phone calls. The on-site visits include 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 rilibove@stanford.edu or 650-736-1235.