



Developmental  
Synaptopathies  
Consortium

# We Are Seeking Volunteers for a PHTS Research Study

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

The goal of this study is to better understand the symptoms and characteristics of PTEN Hamartoma Tumor Syndrome (PHTS) and how these change over time. This information could help develop future treatments

### WHAT WILL THE RESEARCH INVOLVE?

The study will involve up to three visits over three years. Each visit includes a physical exam, medical history questions, a blood draw, and assessments and questionnaires to assess development, behavior, and thinking skills. **Virtual visits are an option.**

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

### WHAT ARE THE POSSIBLE BENEFITS OF TAKING PART?

There are no direct benefits to you or your child for taking part in the study. If you take part, you will receive summary scores of your child's behavioral testing. You are also making a valuable contribution to medical research and helping us learn more about PHTS and autism spectrum disorder.

## LOCATIONS AND CONTACTS

### **Boston Children's Hospital**

Principal Investigator: Mustafa Sahin, MD, PhD

Contact: Emine Arcasoy, emine.arcasoy@childrens.harvard.edu

### **Cleveland Clinic**

Principal Investigator: Charis Eng, MD, PhD

Contact: Beth Crouser, crouseb2@ccf.org

### **University of California at Los Angeles**

Principal Investigator: Julian Martinez, MD, PhD

Contact: Jeff Anderson, jtanderson@mednet.ucla.edu

### **Stanford University Medical Center**

Principal Investigator: Antonio Hardan, MD

Contact: Robin Libove, rlibove@stanford.edu

### **Cincinnati Children's Hospital Medical Center**

Principal Investigator: David Ritter, MD, PhD

Contact: Adrienne Victory, adrienne.victory@cchmc.org

### WHO CAN TAKE PART?

Participants should be **18 months or older** and have been diagnosed with a **PTEN genetic mutation** either with or without autism spectrum disorder (ASD).