Quarterly Newsletter

PTEN Hamartoma Tumor Syndrome Foundation

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TRAVEL SCHOLARSHIPS AVAILABLE THROUGH THE PTEN FOUNDATION!

The PTEN Hamartoma Tumor Syndrome Foundation's mission is to educate about PTEN syndromes, provide financial support to patients, and support scientific research. Your financial contribution will help provide hope to those battling this rare disease, promote awareness, and find a cure.

support the FIGHT against RARE DISEASE

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Launch of a New Clinical Trial in PTEN

The Developmental Synaptopapthies Consortium led by Dr. Mustafa Sahin at Boston Children's Hospital has launched a randomized double-blind controlled trial of everolimus in children and adolescents with a PTEN mutation. The main goals of the study are to evaluate the safety of everolimus in patients with a PTEN mutation, and to determine the efficacy of the medication on neurocognition. The neurocognitive and neurobehavioral deficits often seen in patients with PTEN are associated with very high comorbidity, health care cost, and impact the quality of life of patients and their family. Currently, there are no approved medications that target these neurocognitive and social deficits, so exploration into potentially effective treatments is crucial in order to better the short and long-term outcomes of individuals affected by PTEN.

You may be eligible for the study if you are between the ages of 6 and 21, and have a confirmed PTEN mutation. You can enroll at one of three participating sites, which include Boston Children's Hospital, Stanford University, and Cleveland Clinic. Participants will be randomized to receive either everolimus or placebo. For participants receiving placebo, there will be an optional follow-up phase to receive everolimus. The study consists of eight visits, six of which are onsite, and two are check-in phone calls. Funding is available for costs related to participating in the study. If you are interested in participating in the study and want more information please contact Mia Diplock by phone at 617-919-1476 or email at amelia.diplock@childrens.harvard.edu.

Developmental Synaptopathies Consortium

Update on Natural History Study in PTEN

In our Natural History Study of Individuals with Autism and Germline Heterozygous PTEN Mutations, we have currently enrolled 83 participants with even more scheduled to come in soon! By comparing information about medical history, performance on developmental testing, treatment and interventions received (i.e. speech/language therapies, current and past medications), biochemical markers in the blood and neurological biomarkers through neurological exams, we hope to learn and develop a more effective, standardized care for each one of the three groups enrolled in this study. Individuals ages 3-21 with PTEN and Autism Spectrum Disorder, PTEN without Autism Spectrum Disorder, or Autism Spectrum Disorder and Macrocephaly (without PTEN) are all eligible to enroll and be a part of this study! Enrollment is still open but will be closing soon. If you are eligible and interested or you know of someone who is, please contact Sarah Mischianti by email at Sarah.Mischianti@childrens.harvard.edu or by phone at (617) 919-3499.

PTEN Survey

Boston Children's Hospital (BCH) is working to better understand the parent perspective of patients with PTEN. BCH is looking to conduct a survey of the parents enrolled in our Contact Registry who have children from the ages of 3-17 with a self-reported diagnosis of a PTEN mutation to assess their family's experience with and access to care. If you are eligible and interested in becoming a part of our Contact Registry, please notify Sarah Mischianti (See contact information above).