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HUNTSVILLE, AL, UNITED STATES,
October 23, 2024 /EINPresswire.com/ -Today marks International PTEN
Hamartoma Tumor Syndrome
Awareness Day. The PTEN Hamartoma
Tumor Syndrome Foundation is
hosting an event in Huntsville,
Alabama, bringing together patients,
families, and esteemed PTEN clinicians
and researchers. This gathering aims to
celebrate PTEN families while sharing



essential information about PHTS care and research.

This is a significant occasion as participants will raise awareness about PTEN, emphasize the importance of clinical trials and treatments, and honor the memory and remarkable life of Dr. Charis Eng, a PTEN community champion and a global patient advocate. PTEN Hamartoma Tumor syndrome (PHTS) is a rare genetic condition that causes an increased risk for certain cancers, benign growths, and neurodevelopmental disorders. PHTS describes any person found to have a change or mutation in the PTEN gene; some may also carry diagnoses of Cowden syndrome or Bannayan-Riley-Ruvalcaba syndrome based on their medical history and the features they have developed. One of PTEN's roles in the body is as a tumor suppressor gene, which means that when it works correctly, PTEN helps suppress the growth of any cells trying to grow out of control and become tumors. All our genes come in pairs; persons without PHTS have two working PTEN gene copies in each cell. In people with PHTS, one of these PTEN gene copies has a change that makes it not work in each of the body cells. As you can imagine, this puts people with PHTS at increased risk of developing tumors.

Fortunately, most tumors that develop in persons with PHTS are benign, meaning they will not turn into cancer that can then metastasize or spread to other body parts. Persons with PHTS commonly develop benign growths, most of which are small in the skin, tongue, and gums by adulthood. It has recently been discovered that colon polyps of various types, most of which have a low potential to develop into a malignancy, are seen in most adults with an endoscopy (colonoscopy or upper). Benign breast lumps, thyroid nodules/goiter, and uterine fibroids are common. Vascular malformations needing surgical intervention may also occur. A benign tumor of the cerebellum called Lhermitte-Duclos disease develops in a minority of adults with PHTS. Macrocephaly (larger than average head size) is common, and some children with PHTS are identified due to the presence of developmental delays and autism spectrum disorders. However, many persons with PHTS have no developmental challenges early in life and have successful careers as doctors, lawyers, or whatever other path they wish to follow.

For a person with PHTS, three different studies have found increased lifetime risks for specific cancer types. The study with the most significant number of patients found the following cancer risks (to age 70): breast (85%), thyroid (35%), kidney (34%), uterus (28%), colorectal (9%), and melanoma (6%). (https://pubmed.ncbi.nlm.nih.gov/22252256/) (https://ptenfoundation.org/whatis-pten/)

This event is essential. Families and Clinicians will gather to raise PTEN awareness, learn about new research and care guidelines, and celebrate the memory and life of the community's champion, Dr. Charis Eng.

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