PTEN Hamartoma Tumor Syndrome

PHTS is a rare but under-diagnosed genetic syndrome which affects about 1 in 200,000 people. It is caused by a PTEN gene not working properly, and this raises the risk for developing benign growths and certain cancers. For some people, it can affect learning and development.

Our Mission

- Connect and support PHTS patients worldwide
- Work with clinicians who have expert knowledge of PHTS to develop guidelines for Clinical Centers of Excellence
- Work with researchers to find treatments, and offer patients opportunities to be engaged in this process
- Collaborate with partners who share our vision for supporting patients

Connect * Collaborate * Cure

PTEN Hamartoma Tumor Syndrome Foundation

in collaboration with

PTEN Italia

ptenfoundation.org

ptensyndromefoundation@gmail.com

The content of this leaflet is provided for education and information purposes only. For advice about diagnoses, treatments and health screenings, please consult qualified medical professionals.

The National Comprehensive Cancer Network's screening guidelines for PHTS are regularly reviewed. Please check their website for the latest information about screenings.

PTEN Hamartoma Tumor Syndrome Foundation

PTEN Hamartoma Tumor Syndrome

Managing Health Risks

CONNECT COLLABORATE CURE

www.ptenfoundation.org

The main health risk of PTEN hamartoma tumor syndrome (PHTS) is cancer, but being born with a PTEN mutation does not mean you will get cancer, or have other health or learning problems associated with this mutation. People can have exactly the same type of PTEN mutation yet be affected very differently. A combination of diet, lifestyle, environment and overall genetic makeup may be the reason for this. Currently, routine enhanced screenings offer the best chance of finding cancer early, and preventative procedures may help to stop some cancers from developing in the first place.

For Women

From age 18

- breast self-exam: monthly

From age 25

- breast exam by a doctor: every 6 to 12 months

From age 30-35

- Yearly Mammogram (consider digital breast Tomosynthesis) and Yearly breast MRI beginning at age 35 (or ten years earlier than the first relative that had breast cancer in the family)
- Report any abnormal bleeding and consider an endometrial biopsy every 1-2 years or discuss annual trans-vaginal ultrasound with your physician.

Connect * Collaborate * Cure

For All Adults

Yearly

- physical exam including urinalysis by cytospin for blood
- skin check
- thyroid ultrasound

From age 35

- colonoscopy: every 5 years or more often if needed

From age 40

- Kidney Ultrasound or CT if advised: yearly or every other year

Ask your doctors about:

- ✳ the common warning signs of cancer.
- ✳ Recommended risk reducing surgical prevention.
- ✳ The best plan for managing your health risk: If you have symptoms and/or a family history of cancer, you may be asked to start screenings earlier and have other screening tests.

Additional Considerations

Under 18 years

- Annual physical exam
- Annual skin check
- thyroid ultrasound: start at age 7; annual if advised.

If advised:

- learning and development checks
- assessment for autism
- GI screening recommended
- Baseline Brain MRI (Annual if symptomatic and recommended by your physician)

Please note that guidelines vary in some countries.

Join our efforts to help accelerate pten research by signing up for our IRB approved patient registry at: www.ptenregistry.org

Connect * Collaborate * Cure

This leaflet was created by patients for patients and approved by an advisor with expert knowledge of PHTS

Sources: National Comprehensive Cancer Network Guidelines for Detection, Prevention and Risk Reduction: Genetic/Familial High-Risk Assessment: Breast and Ovarian: Cowden Syndrome/PHTS: nccn.org
Gene Reviews (© 1993-2018 University of Washington): PTEN Hamartoma Tumor Syndrome: ncbi.nlm.nih.gov/books/NBK1488/