

⌘ Is there a PHTS test?

The diagnosis of PHTS is established by identification of a germline, or inherited, PTEN mutation, which is passed from parent to child, on molecular genetic testing.

⌘ How do I find a Medical Geneticist or Genetic Counselor?

To locate a genetic counselor, visit the National Society of Genetic Counselors website at www.nsgc.org and click on the “Find a Genetic Counselor” link.

⌘ What is the Cleveland Clinic PTEN/Cowden syndrome multi-disciplinary clinic?

Some patients with PHTS, Cowden syndrome, or Bannayan-Riley-Ruvalcaba syndrome have health needs for which coordinated visits with multiple subspecialists are helpful. Our clinical team includes healthcare providers who have seen and treated other patients with PHTS / Cowden syndrome / Bannayan-Riley-Ruvalcaba syndrome and are knowledgeable regarding these uncommon syndromes. If you are interested in learning more about our PTEN multi-disciplinary clinic, please contact our genetic counselor and clinic coordinator or visit our website at: <https://my.clevelandclinic.org/departments/genomics/specialties/pten-clinic#overview-tab>

PTEN Genetic Counselor Coordinator

216-636-5535 or pten@ccf.org

The PTEN Hamartoma Tumor Syndrome Foundation was founded to find treatments or therapies for PTEN Syndromes by funding research, providing PHTS education, supporting patients, and by raising awareness.

PTEN Hamartoma
Tumor
Syndrome
Foundation

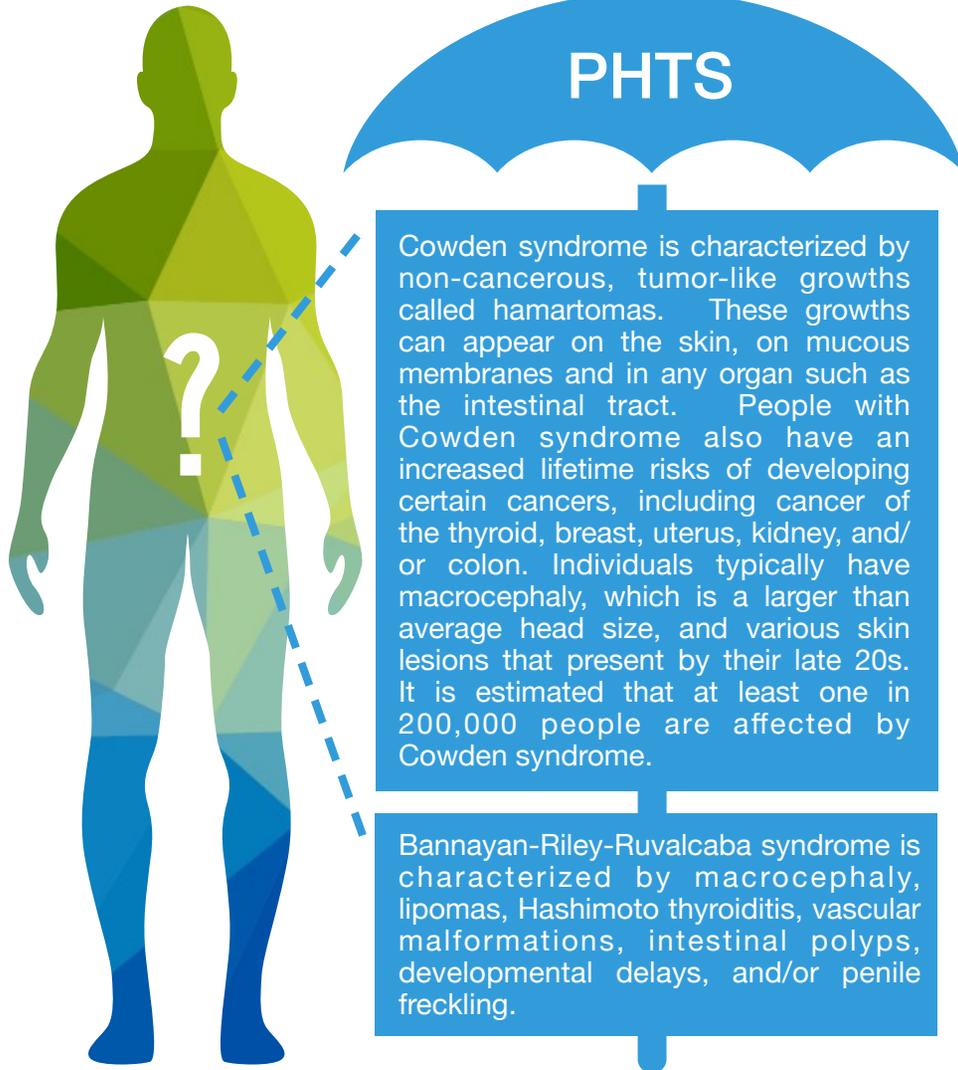


In collaboration with the Cleveland Clinic

www.ptenfoundation.org

Do I have PHTS?

PTEN hamartoma tumor syndrome (PHTS) is an umbrella term describing individuals with any clinical condition who also carry a PTEN gene alteration (mutation). PHTS includes those subsets of Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome (BRRS), PTEN-related Proteus syndrome (PS), and Proteus-like syndrome that have PTEN mutations.



It is important to point out that not every individual will develop all features of Cowden syndrome or Bannayan-Riley-Ruvalcaba syndrome.

What is PTEN?

PTEN is one of the body's many tumor suppressor (cancer fighting) genes. When they work properly, tumor suppressor genes help to control cell growth. When they are not functioning properly, cells can grow out of control and turn into either benign or malignant tumors. Many people with PTEN syndromes, Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, Cowden-like syndrome, Proteus syndrome, and autism have been found to have PTEN gene mutations as the cause of their medical concerns.

Does PHTS have a cure?

At this time, PHTS does not have a cure. Hope can be found in several ongoing research studies at the Cleveland Clinic on PTEN-related disorders. Combining clinical patient information with DNA, RNA, and protein studies of the PTEN gene and related pathways, these studies focus on frequency of particular characteristics (i.e. cancer diagnoses among those with PTEN alterations). The goals of our research are to better define cancer and other risks associated with PTEN and other gene mutations, to inform genetic counseling and medical management, such as clinical screening and prevention (for example, preventive mastectomy). We also hope that this research will lead to the development of targeted molecular therapies, for treatment as well as prevention, for persons with PTEN alterations. If you have questions about eligibility, please contact our genetic counselor coordinator.



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