Cleveland Clinic
Genomic Medicine Institute

# The PTEN Study

## Information for Prospective Patients

**Principal Investigator: Charis Eng, MD, PhD, FACP**

**Study Coordinator: Beth Crouser**

### What is the PTEN study?

Our study collects blood and/or DNA samples and medical information from persons with characteristics that have been associated with PTEN gene mutations. Researchers study DNA, RNA, and protein from the PTEN gene and related pathways. Among the many goals of this study, we hope to determine the minimal number of features associated with some likelihood of a PTEN alteration. We are also studying the frequency of particular characteristics, such as cancer diagnoses, among those with PTEN alterations. We ultimately hope that this research will lead to the development of targeted molecular therapies and an improved understanding of the natural history of PTEN Hamartoma Tumor Syndrome.

### What is PTEN?

PTEN is one of the body's many tumor suppressor 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 Cowden Syndrome, Bannayan-Riley-Ruvalcaba Syndrome, and a few other genetic conditions have been found to have PTEN gene mutations as the cause of their medical concerns. These conditions as a group are referred to as PTEN Hamartoma Tumor Syndrome.

### What is the Enrollment process?

Your referring healthcare provider will determine if you are eligible and contact the Cleveland Clinic's Genomic Medicine Institute with your contact information and basic clinical information. The research staff will contact you to discuss enrollment, complete the informed consent process, and facilitate sending your blood or DNA sample to Cleveland.

**Biography: Charis Eng, MD, PhD**

Dr. Charis Eng grew up in Singapore and Bristol, UK and entered the University of Chicago at the age of 16. After completing an MD and PhD at Pritzker School of Medicine, she specialized in internal medicine at Beth Israel Hospital, Boston and trained in medical oncology at Harvard's Dana-Farber Cancer Institute. She was formally trained in clinical cancer genetics at the University of Cambridge and the Royal Marsden NHS Trust, UK, and in laboratory-based human cancer. She was Assistant Professor of Medicine at the Dana-Farber Cancer Institute and Harvard Medical School, and after three years was recruited to a tenured position at The Ohio State University in 1999 as Director of the Clinical Cancer Genetics Program and Director of the Division of Human Genetics. She was recruited to the Cleveland Clinic in September 2005 where she is chair and founder of the Genomic Medicine Institute and its clinical component, the Center for Personalized Genetic Healthcare. She currently holds the Sondra J. and Stephen R. Hardis Endowed Chair of Cancer Genomic Medicine, and was recently selected as an American Cancer Society Professor. Dr. Eng led the research team which in 1997 discovered the causative relationship between the PTEN gene and Cowden syndrome. Since then, she and her team have dedicated themselves to characterizing the gene and how it results in the many clinical facets so that personalization of health care can be achieved with these patients and their families.

*Enrollment must be facilitated by a healthcare provider (genetic counselor, doctor, nurse, etc.) able to take responsibility for receiving study results.*

## What Type of Sample Is Taken?

A blood sample is preferred. Turn around time for results is usually 6 months but may be longer depending on laboratory workload.

## Study Results

Your healthcare provider will be informed if a deleterious mutation, single nucleotide polymorphism, or variant of uncertain significance is identified. Depending on your medical institution, your test result may or may not be placed in your medical records.

### BENEFITS OF PARTICIPATION

- You can elect to receive clinically relevant research results.
- Research testing costs are not charged to your insurance or to you.
- If a mutation is identified, the cost of confirmatory site-specific clinical testing is substantially lower than full PTEN gene analysis.
- Your participation in this study helps the translation of scientific knowledge into clinical patient care.

If you elect to receive results, they will be reported to your referring healthcare provider via the Cleveland Clinic secured email system. Your referring provider will contact you with your research study results. Please be aware that we will not release your results to your healthcare provider until all required study documents have been completed and received.

If a mutation is identified, clinical confirmation in a CLIA-certified lab is recommended to confirm research findings. Per our institutional IRB requirements, the exact nomenclature (name of gene alteration) cannot be disclosed to your referring provider. However, if a deleterious mutation is identified, we will share the exact mutation nomenclature with the clinical lab of choice for site-specific mutation confirmation.

## Can My Relatives Participate In the PTEN Study?

Maybe. Your relatives should ask their healthcare provider if they qualify. Sometimes, evaluation by a genetics professional is necessary to determine study eligibility. If you have an identified PTEN mutation, your first-degree relatives (parents, siblings, and children) automatically qualify for study participation. If they wish to participate, they need to enroll through their own healthcare provider.

*Thank you for your interest in our research study!*

## How Do I Find a Medical Geneticist/Genetic Counselor?

If you live in or can travel to the greater Cleveland area, we would be glad to see you for a clinical genetics evaluation in the Cleveland Clinic Center for Personalized Genetic Healthcare (CPGH). To learn more about CPGH's comprehensive clinical genetics services and its staff, visit www.clevelandclinic.org/genetics.

To locate a genetic counselor outside the Northeastern Ohio area, visit the National Society for Genetic Counseling Website at: www.nsgc.org and click on the "Find a Counselor" link on the left-hand side of the page.