Cleveland Clinic researcher discovers genetic cause of thyroid cancer

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Cleveland Clinic researchers have discovered three genes that increase the risk of thyroid cancer, which is has the largest incidence increase in cancers among both men and women.

Research led by Charis Eng, M.D., Ph.D., Chair and founding Director of the Genomic Medicine Institute of Cleveland Clinic's Lerner Research Institute, included nearly 3,000 patients with Cowden syndrome (CS) or CS-like disease, which is related to an increased risk of breast and thyroid cancer.

Mutations in the PTEN gene are the foundation of Cowden syndrome. PTEN is a tumor suppressor gene, helping to direct the growth and division of cells. Inherited mutations in the PTEN gene have been found in approximately 80 percent of Cowden syndrome patients. These mutations prevent the PTEN protein from effectively regulating cell survival and division, which can lead to the formation of tumors.

"Our investigation into the genetics behind thyroid disease raises important details relevant to diagnosis and treatment," said Dr. Eng. "We hope to promote the earliest diagnosis and most targeted treatment possible."

The conclusions of this research, published in the *Journal of Clinical Endocrinology & Metabolism*, found that all six patients under age 18 had pathogenic PTEN mutations. The researchers recommend that the thyroids of children with PTEN mutation-causing CS-related disease

receive increased surveillance.

Children with thyroid cancer are recommended to have testing for PTEN mutations, which could warrant surveillance for additional cancers or maladies. In contrast, alterations in the SDH and KLLN genes did not associate with thyroid cancer in children.

PTEN gene testing in the setting of genetic counseling is already routinely practiced, and has been a powerful gene-enabled diagnostic test which then personalizes clinical screening and treatment. Once SDH and KLLN findings are independently validated, the tests could be implemented as a clinical routine test as well. Importantly, these three genes belong to different cell pathways so that specific molecular-targeted treatments can be utilized depending on which gene is involved.

Provided by Cleveland Clinic

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