Genomic screening can help detect thyroid cancer, finds study

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Population-based genomic screening can facilitate early detection of medullary thyroid cancer (MTC) in patients with variants in the RET gene, a new Geisinger study found.

MTC accounts for 2% to 5% of all thyroid cancers and is responsible for 13% of all deaths related to thyroid cancer. About one-quarter of MTC

cases are hereditary and can be attributed to variants in the RET gene.

The Geisinger study evaluated 75 patients who were found to have RET gene variants as part of the MyCode Community Health Initiative. None of the patients had any symptoms of thyroid cancer prior to receiving these genetic results.

Twenty of these patients chose to have a thyroidectomy after learning of the RET variant; 13 also had lymph nodes removed. Of the 20 patients who chose to have surgery, cancer was detected in 12. Two additional patients had C-cell hyperplasia, a known precursor to MTC. The results were published in *JAMA Otolaryngology—Head and Neck Surgery*.

While genomic screening can provide opportunities for early detection and treatment of MTC, more work is needed to ensure patients have the education and resources they need to understand the condition and make decisions about their care based on their genetic risk factors, the research team said.

"It is possible that some patients in the study chose not to have surgery due to an absence of symptoms at the time of detection, or were hesitant to act upon a potential risk," said Nicholas Purdy, D.O., FACS, director of head and neck surgery in Geisinger's department of otolaryngology and a lead author of the study. "It is important to further identify barriers to care and make sure that all patients receive the information they need to make informed decisions about their care."

"Genomic screening through MyCode allows us to find previously unrecognized risks for cancer and other serious diseases and intervene early," said Adam Buchanan, M.S., MPH, chair of Geisinger's Department of Genomic Health and a co-author of the study.

"But it also compels us to support patients and clinicians throughout the

process and conduct research that improves the likelihood of positive health outcomes for patients and families."

More information: Priscilla F. A. Pichardo et al, Thyroidectomy Outcomes in Patients Identified With RET Pathogenic Variants Through a Population Genomic Screening Program, *JAMA Otolaryngology–Head & Neck Surgery* (2023). DOI: 10.1001/jamaoto.2022.4195

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