Genomic variants study points to improved detection of thyroid cancer

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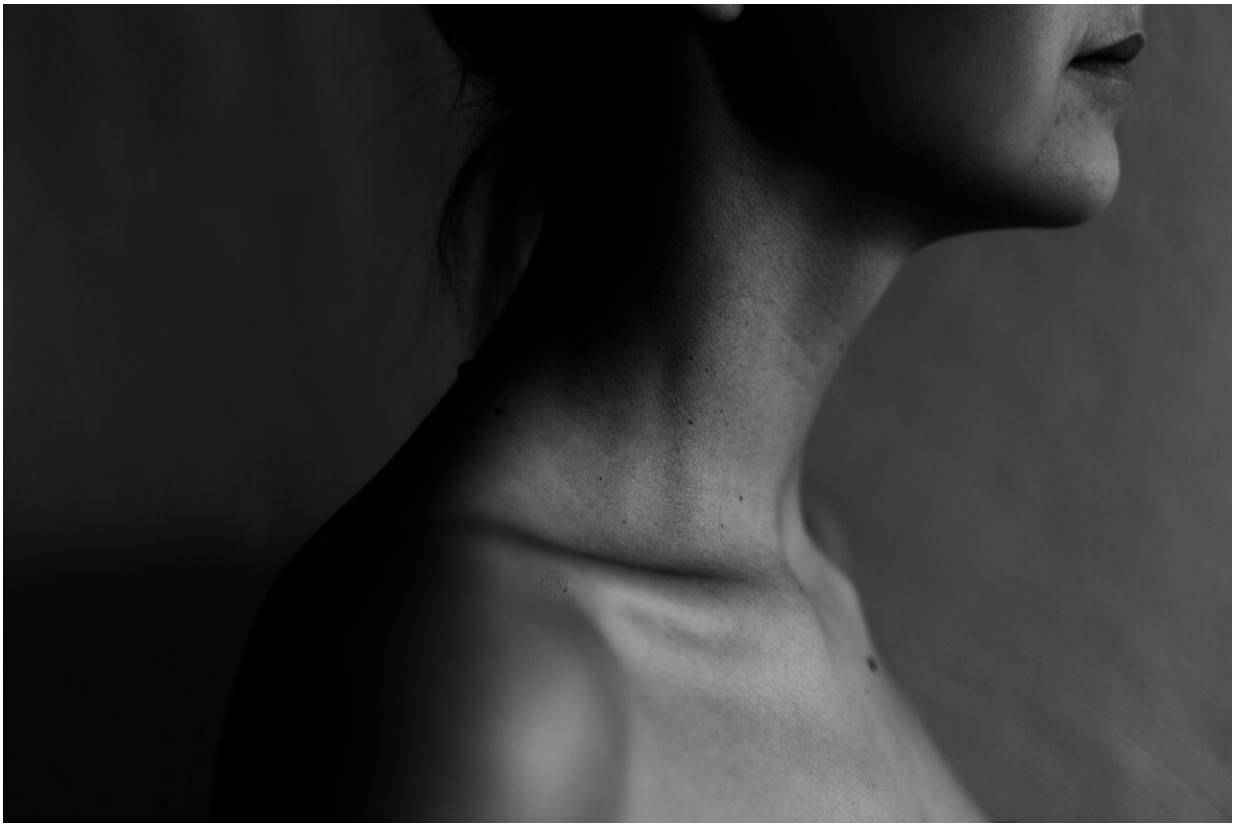

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Researchers from Sinai Health and the University of Toronto have gleaned new insights into how thyroid cancer could be more effectively treated.

The study, which looked at thyroid tumor tissues and thyroid nodule biopsies from 620 patients at Mount Sinai Hospital from 2016 to 2022, examined whether differences in patients' RAS genomic variants were reflected in the status of their tumors. It also investigated the presence of the variant BRAF V600E and TERT promoter variants in the patient's samples.

Researchers ultimately concluded that differences in RAS in combination with BRAF V600E and TERT promoter variants could be used to arrive at more accurate cancer diagnoses in patients with indeterminate thyroid nodules.

"The findings help promote understanding of the interpatient differences in genomic variation among patients who carry the same genetic mutation, thereby facilitating individualized treatment based on the extent of the mutation present in the patient," says Guodong (David) Fu, a researcher at the Lunenfeld-Tanenbaum Research Institute and the Alex and Simona Shnaider Research Laboratory in Molecular Oncology at Mount Sinai Hospital.

Fu adds that researchers developed novel molecular assays for the study using digital polymerase chain reaction, a technique that means they could sensitively quantify the genetic mutation level of the patient materials.

The results were published in *JAMA Network Open*.

Other researchers involved in the study included: Ronald Chazen, also of the Lunenfeld-Tanenbaum Research Institute and the Alex and Simona Shnaider Research Laboratory in Molecular Oncology, and Christina MacMillan, a pathologist at Sinai Health and an assistant professor in the Temerty Faculty of Medicine's department of laboratory medicine and pathobiology, and Ian Witterick, surgeon-in-chief at Sinai Health and a

professor in Temerty Medicine's department of otolaryngology—head and neck surgery.

The paper notes that there has been a sharp increase in papillary thyroid cancer since the 1980s, and that in 30% of cases where a fine-needle aspiration biopsy of a suspected nodule takes place, there is an indeterminate diagnosis that may lead to a diagnostic surgery.

Fu says research that assists with precision thyroid cancer detection is important for many reasons, including that some patients who seek treatment for thyroid tumors end up finding out their tumors are benign after diagnostic surgery. The findings could help medical practitioners differentiate low-risk tumors from high-risk ones, he says, and help avoid unneeded surgical procedures.

"(This finding) enhances the preoperative diagnostic accuracy for patients, in order to avoid unnecessary surgery for benign thyroid nodules," says Fu.

Witterick, who is also otolaryngologist-in-chief at Mount Sinai Hospital, says the research is important because identifying differences in genomic variants between patients can enhance precision in cancer detection, especially diagnosing malignancies before surgery and distinguishing low-risk cancers from more aggressive ones.

More information: Guodong Fu et al, Discriminating Interpatient Variabilities of RAS Gene Variants for Precision Detection of Thyroid Cancer, *JAMA Network Open* (2024). DOI: 10.1001/jamanetworkopen.2024.11919

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