

Next-gen sequencing test identifies cancerous thyroid nodules with high degree of accuracy

September 15 2015, by Anita Srikameswaran

A next-generation sequencing test is successfully predicting which thyroid nodules are cancerous and require surgical removal, reducing the need for multiple invasive diagnostic procedures, according to researchers at the University of Pittsburgh Cancer Institute (UPCI) and Pitt School of Medicine. Their findings were recently published online in the journal *Thyroid*.

In up to 80 percent of cases, examination of cells collected with a fine needle from a suspicious lump in the thyroid, a gland in the front of the neck, typically can tell a pathologist whether it is benign or malignant, said lead investigator Yuri Nikiforov, M.D., Ph.D., professor of pathology and director of Pitt's Division of Molecular and Genomic Pathology.

"However, in 20 percent of cases, the result is indeterminate, meaning we can't say for certain whether the nodule is cancerous," he said. "That means the patient might have a repeat needle biopsy, or will go to the operating room to have the affected thyroid lobe removed for further assessment. If it turns out to be cancer, the patient has to have yet another surgery to have the rest of the thyroid taken out."

Three-quarters of such diagnostic surgeries are performed on what turn out to be [benign nodules](#). Such patients could have avoided surgery if physicians were confident without surgical excision that the nodules are very likely to be harmless.

In the new study, the researchers used the latest version of the test they developed, called ThyroSeq v2.1, to look for more than 300 cancer-associated mutations in 56 genes using cells obtained from fine-needle aspiration biopsies in more than 440 patients. Of that group, 96 patients had established diagnoses through surgery, allowing the team to assess ThyroSeq's predictive power. The team found the test was able to correctly classify 20 out of 22 cancers with high precision and accuracy. Most importantly, when the test was negative, the residual risk of cancer in those nodules was so low that surgical excision was not needed.

"We finally have a test that offers high accuracy in predicting whether a nodule is cancerous or if it is benign," Dr. Nikiforov said.

"This molecular testing panel holds great promise for streamlining and eliminating unnecessary surgery, not just here, but nationwide," said co-author Sally E. Carty, M.D., professor and chief of endocrine surgery, Pitt School of Medicine, and co-director of the UPMC/UPCI Multidisciplinary Thyroid Center, which has been offering the [test](#) since 2014. It also is available to and used by [thyroid](#) clinics around the country.

"Thyroid cancer now is the fifth most common cancer diagnosed in women, and it is one of the few cancers that continues to increase in incidence," she noted. "It's important to get to the diagnosis quickly and correctly."

More information: "Impact of the Multi-Gene ThyroSeq Next-Generation Sequencing Assay on Cancer Diagnosis in Thyroid Nodules with Atypia of Undetermined Significance/Follicular Lesion of Undetermined Significance Cytology." *Thyroid*, Online Ahead of Print: September 10, 2015. [DOI: 10.1089/thy.2015.0305](https://doi.org/10.1089/thy.2015.0305)

Provided by University of Pittsburgh

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