

Next generation sequencing test improves detection of thyroid cancer, reduces unnecessary surgeries

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A new test for genetic markers that can identify which lumps in the thyroid gland are cancerous and which are harmless – potentially preventing unneeded operations – will make its debut Oct. 1 for patients seeking care at the UPMC/UPCI Multidisciplinary Thyroid Center (MTC).

Growth of a small mass or nodule of the [thyroid gland](#), which is located in the "Adam's apple" area of the neck, is very common, particularly with aging, said Yuri Nikiforov, M.D., Ph.D., director of thyroid molecular diagnostics at the thyroid center and a professor of pathology at the University of Pittsburgh School of Medicine. The nodule will be benign 90 percent of the time, but an ultrasound-guided biopsy of the suspicious tissue is needed to confirm it is harmless.

"The test we had been using can distinguish between cancerous and benign nodules about 70 percent of the time, but that means the result is uncertain in nearly a third of cases," Dr. Nikiforov explained. "When that happens, the patient has to have the nodule surgically removed so that more extensive testing can be done. If it turns out to be cancerous, yet another operation might be needed to remove the entire thyroid gland."

Approximately 100,000 thyroid nodule biopsies done annually yield uncertain results, and most of these patients must undergo follow-up

testing and diagnostic surgery, even though the likelihood of having a cancer is very low, he noted.

The new test, called ThyroSeq, was designed and developed by Dr. Nikiforov and his team and uses a technique called next-generation sequencing. This revolutionary technology allows pathologists to simultaneously test for multiple [genetic markers](#) using just a few cells collected from the nodule and at low cost. The test detects mutations associated with [thyroid cancer](#) in nearly 300 sites of 12 genes in the thyroid cells obtained during the biopsy procedure. UPMC will be the first academic institution in the country where next-generation sequencing technology will be used to improve care for patients with thyroid nodules.

"The ultimate goal of molecular testing is to allow patients to have the initial definitive operation for thyroid cancer and to avoid diagnostic surgery for benign conditions," said MTC co-director Sally E. Carty, M.D. "The next -generation sequencing [test](#) is very exciting, innovative and promising because it's designed to pick up a much higher percentage of cancers." Dr. Carty added that the new panel is the same price as the earlier one, which was shown to be cost-effective in a 2012 study led by MTC expert Linwah Yip, M.D.

According to Dr. Nikiforov's research, ThyroSeq findings suggest that certain gene alteration patterns may be associated with more aggressive cancers, so there is potential to develop tailored treatment approaches for the specific kinds of tumors.

"If we can distinguish aggressive thyroid cancers from cancers that are growing much more slowly and will not metastasize, we might be able to avoid surgery in these low-risk patients in favor of watchful waiting," said MTC co-director Steven Hodak, M.D. "That's really the Holy Grail – finding patients both with and without thyroid cancer for whom

surgery is unnecessary and not putting them through the expense and risk of surgeries they don't need."

Each year, approximately half of the 25,000 patients assessed at the MTC have thyroid conditions and more than 900 thyroid operations are performed by MTC surgeons. The center aims to provide patients with one-stop evaluation from [thyroid](#) experts in a variety of fields, including surgery and endocrinology.

Provided by University of Pittsburgh

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