

'Gene signature' test diagnoses benign thyroid growths

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A new genetic test accurately and consistently diagnoses benign growths, or nodules, on the thyroid gland, according to a study from Chile. The results will be presented Saturday at The Endocrine Society's 95th Annual Meeting in San Francisco.

"We have developed a 'gene signature' that effectively identifies [benign thyroid nodules](#)," said Hernan Gonzalez, MD, PhD, associate professor at the Pontifical Catholic University of Chile in Santiago. "This test is potentially useful to identify patients who do not require surgery."

The [thyroid gland](#), located in the front of the neck, secretes thyroid hormone. This hormone is involved in many bodily processes, including turning food into energy. For unknown reasons, the thyroid gland sometimes develops small nodules. Although these nodules are extremely common and usually benign, a small number are cancerous, which makes [screening tests](#) essential.

In the United States this year, slightly more than 60,000 cases of thyroid cancer will be diagnosed and about 1,850 related deaths will occur, according to estimates from the [American Cancer Society](#). Of these diagnosed cases, more than 45,000 will occur in women, who develop the disease more often than men.

Fortunately, thyroid cancer usually is curable. Screening for thyroid cancer involves using a thin needle to take a biopsy of the growth, called a fine needle aspirate biopsy, which is then analyzed for [cancer cells](#).

The problem with the currently available screening test is that it often yields inconclusive results. In one-fifth to one-fourth of these tests, the results are unclear. In addition to causing anxiety among patients, inconclusive tests lead to repeated tests to rule out cancer, or even to surgery, given that cancer is present in about 25 percent of inconclusive cases. As a result, three out of four patients have unnecessary surgery, leading to rising treatment costs and the risk of possible complications related to unnecessary medical procedures.

The genetic test developed by the current study accurately identified benign nodules in nearly all samples analyzed. Specifically, the test differentiated between cancerous and non-cancerous tissue in 96 percent of thyroid samples.

"For the general public, this is important since it will offer a diagnostic tool that will avoid thousands of surgeries, with a major impact in health costs, eliminating potential surgical complications and the need for permanent [thyroid hormone](#) supplementation," Gonzalez said. "In addition, it should be widely available to local labs and hospitals and at a reasonable cost for the health system."

To develop the "gene signature" test, investigators conducted a literature search to identify 18 genes associated with [thyroid cancer](#). They then selected 10 of these genes to develop a computer program that detects the presence of cancer in thyroid nodules. Using a sensitive DNA-amplifying test called polymerase chain reaction, they tested samples of thyroid nodules from 300 patients.

A biomedical research consortium in Santiago funded the study.

Provided by The Endocrine Society

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