



THYROID NODULE

Utility of genetic testing in thyroid nodule biopsies

BACKGROUND

Thyroid nodules are very common and are seen in up to 50% of adults. Biopsy of a thyroid nodule is commonly done to rule out thyroid cancer. The decision to treat with thyroid surgery is straightforward if the biopsy results are positive for thyroid cancer. Similarly, surgery is usually avoided if the biopsy results are benign. However, in 10-20% of cases, the cytology is indeterminate making the decision to refer to surgery more difficult. The risk of thyroid cancer in nodules with indeterminate biopsy results varies from 10-75%.

The Afirma gene expression classifier (GEC) test uses thyroid cells obtained at the time of biopsy to screen for molecular markers (genes) that are associated with thyroid cancer. Its manufacturers claim that the test can predict which of the indeterminate nodules are likely to be benign (with a 95 % certainty), and therefore do not require surgery, from those that are likely cancerous (with a 50% certainty) and need to be referred to surgery. The hope is that this test will prevent unnecessary thyroid surgeries. This study details the results of the use of the Afirma GEC in five medical centers.

THE FULL ARTICLE TITLE

Alexander EK et al Multicenter clinical experience with the afirma gene expression classifier. *J Clin Endocrinol Metab* 2014;99:119-25. Epub December 20, 2013; doi: 10.1210/jc.2013-2482.

SUMMARY OF THE STUDY

The authors collected data on 339 nodules with indeterminate biopsy results and were tested by Afirma GEC from five medical centers. A total of 174 patients had a benign GEC as compared with 141 patients with suspicious GEC. Only 2% of patients with benign GEC were recommended to have surgery compared to 44% of patients in the GEC suspicious group.

WHAT ARE THE IMPLICATIONS OF THIS STUDY?

The authors concluded that the clinical experience of the five medical centers support the usefulness of the Afirma GEC in preventing unnecessary thyroid surgeries in the subset of patients with indeterminate nodules who have a benign GEC result. The major criticism to this study is that patients with indeterminate nodules and benign GEC who did not have surgery were not followed long enough to truly rule out thyroid cancer. Long term studies are needed to validate the results of this study.

— Mona Sabra, MD

ATA THYROID BROCHURE LINKS

Thyroid cancer: <http://www.thyroid.org/cancer-of-the-thyroid-gland>

Thyroid Nodules: <http://www.thyroid.org/what-are-thyroid-nodules>

ABBREVIATIONS & DEFINITIONS

Thyroid nodule: an abnormal growth of thyroid cells that forms a lump within the thyroid. While most thyroid nodules are non-cancerous (Benign), ~5% are cancerous.

Thyroid fine needle aspiration biopsy (FNAB): a simple procedure that is done in the doctor's office to determine if a thyroid nodule is benign (non-cancerous) or cancer. The doctor uses a very thin needle to withdraw cells from the thyroid nodule. Patients usually return home or to work after the biopsy without any ill effects.

Genes: a molecular unit of heredity of a living organism. Living beings depend on genes, as they code for all proteins and RNA chains that have functions in a cell. Genes hold the information to build and maintain an organism's cells and pass genetic traits to offspring.

Molecular markers: genes and microRNAs that are expressed in benign or cancerous cells. Molecular markers can be used in thyroid biopsy specimens to either to diagnose cancer or to determine that the nodule is benign.