



THYROID CANCER

How often are BRAF genetic mutations found in biopsy samples of thyroid nodules?

BACKGROUND

Papillary cancer is the most common thyroid cancer. While most patients with papillary cancer do well and have an excellent prognosis, some do have a more aggressive form of cancer. The focus of studies on patients with the more aggressive papillary cancers is on mutations in cancer-associated genes, especially a gene known as BRAF. In Korea, more than 90% of patients with thyroid cancer have papillary cancer. Surprisingly, >80% of Korean patients who have papillary thyroid cancer have a mutation in the BRAF gene called BRAF-V600E. The rate of this mutation is higher in Korea than in many western countries. The aim of this study was to determine if it was useful to check for the BRAF-V600E in thyroid fine needle aspiration biopsy (FNAB) samples of thyroid nodules. This study looked at how often the BRAF-V600E mutation was found in FNAB samples of thyroid nodules and how often thyroid cancer was diagnosed after surgery.

THE FULL ARTICLE TITLE:

Kim et al. Surgical results of thyroid nodules according to management guideline based on BRAF-V600E mutation status. *J Clin Endocrinol Metab.* Jan 14, 2011 [Epub ahead of print] doi:10.1210/jc.2010-1082.

SUMMARY OF THE STUDY

A total of 849 FNAB samples were tested for the presence of the BRAF-V600E mutation. The FNAB diagnosis and percentage that had the BRAF-V600E mutation were as follows: 504 samples “benign” – 0% mutation, 141 samples “atypical” – 32% mutation, 54 samples “suspicious” for cancer – 85% mutation,

140 samples “malignant” – 92% mutation, 10 samples “indeterminate” – 10% BRAF-V600E mutation. All patients whose FNAB was read as suspicious for cancer or positive for cancer, whether or not the BRAF-V600E mutation was present, had papillary thyroid cancer at surgery. For patients who had an atypical FNAB and went to surgery, papillary thyroid cancer was diagnosed in 29 of 30 patients with the BRAF-V600E and 3 of 12 patients without the mutation. Only 3 of 8 patients whose FNAB was read as indeterminate and had surgery were diagnosed with cancer and none of them had the BRAF-V600E mutation.

WHAT ARE THE IMPLICATIONS OF THIS STUDY?

The authors of this study concluded that for patients who have an atypical result on their thyroid nodule FNAB sample, a positive BRAF-V600E mutation test may be helpful in guiding the need for thyroid surgery. Because there were patients without the mutation that had thyroid cancer, it is important to realize that it is not a 100% accurate test. However, it is another option that can be used to help identify those patients that will benefit the most with surgery.

— Anna Sawka, MD

ATA THYROID BROCHURE LINKS

Thyroid cancer: http://thyroid.org/patients/patient_brochures/cancer_of_thyroid.html

Thyroid Surgery: http://thyroid.org/patients/patient_brochures/surgery.html

ABBREVIATIONS & DEFINITIONS

BRAF gene — this is gene that codes for a protein that is involved in a signaling pathway and is important for cell growth. Mutations in the BRAF gene in adults appear to cause cancer.

Cancer-associated genes — these are genes that are normally expressed in cells. Cancer cells frequently have mutations in these genes. It is unclear whether mutations in these genes cause the cancer or are just associated with the cancer cells. The cancer-associated genes important in thyroid cancer are BRAF, RET/PTC and RAS.

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THYROID CANCER, continued

Papillary thyroid cancer — the most common type of thyroid cancer.

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.

Indeterminate thyroid biopsy — this happens usually when the diagnosis is a follicular or hurtle cell lesion. Follicular and hurtle cells are normal cells found in the thyroid. Current analysis of thyroid biopsy results cannot differentiate between follicular or hurtle cell cancer from noncancerous adenomas. This occurs in 15-20% of biopsies and often results in the need for surgery to remove the nodule.

Atypical thyroid biopsy — this happens when there are some abnormal/atypical cells in the biopsy sample but not enough to diagnose a cancer. However, because there are abnormal cells in the biopsy sample, the specimen cannot be called benign. Sometimes a repeat biopsy may be helpful but often surgery is recommended to remove the nodule.