A publication of the American Thyroid Association

### **THYROID CANCER**

## Gene mutations and papillary thyroid cancer

#### BACKGROUND

The rate of diagnosis of papillary thyroid cancer has increased markedly in the past 20 years. Many cancers have mutations in cancer-associated genes that are thought to have caused the cancer. Analysis of mutations in cancer-associated genes are helpful in making a diagnosis of cancer on a thyroid biopsy sample as well as predicting the risk of recurrence of the cancer after thyroid surgery. One common cancer-associated gene mutation is BRAFV600E. Identification of this mutation in cells of thyroid nodules that are biopsied is said to ensure that the nodule is cancerous. Further, this mutation has been shown to be common in larger papillary thyroid cancers that are more aggressive. However, not all thyroid cancers have this, or any, mutation. The goal of this study were to determine whether the presence of BRAF<sup>V600E</sup> is associated with more aggressive types of papillary thyroid cancer.

#### THE FULL ARTICLE TITLE

Lin JD et al . Clinical manifestations and gene expression in patients with conventional papillary thyroid carcinoma carrying the BRAF<sup>V600E</sup> mutation and BRAF pseudogene. Thyroid. March 30, 2016 [Epub ahead of print].

#### **SUMMARY OF THE STUDY**

This study looked at surgical specimens from 62 women and 16 men and normal appearing tissue from "cancerfree" areas in 48 of the patients. Cancers were graded using a standard system. Patients were treated according to the 2009 ATA cancer management guidelines and were followed for on average for 77 months. The study assessed the presence of  $BRAF^{V600E}$  and numerous other tissue markers.

The authors reported that, in patients with papillary thyroid cancer, there was no significant relationship between the presence of BRAF<sup>V600E</sup> and patient age, sex, cancer size, cancer extending outside of the thyroid, presence of multiple cancers, lymph-node spread, distant spread, clinical stage, "risk category," or the likelihood of remaining disease-free during an average follow-up period of 41 months.

# WHAT ARE THE IMPLICATIONS OF THIS STUDY?

Where the presence of BRAF<sup>V600E</sup> mutations may be helpful in diagnosing papillary thyroid cancer by a needle biopsy, in this study, the presence of BRAF<sup>V600E</sup> did not help predict whether a cancer would be aggressive in its appearance, tumor stage, or the likelihood of disease-free survival. This study is in conflict with some other studies, so further research is needed to determine the role of BRAF<sup>V600E</sup> in the management of papillary thyroid cancer. — Ronald B. Kuppersmith, MD, FACS

#### **ATA THYROID BROCHURE LINKS**

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

Thyroid Surgery: <u>http://www.thyroid.org/thyroid-surgery/</u> Thyroid Cancer: <u>http://www.thyroid.org/thyroid-cancer/</u>

#### DEFINITIONS

Papillary thyroid cancer: the most common type of thyroid cancer. There are 3 variants of papillary thyroid cancer: classic, follicular and tall-cell.

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. 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.

BRAF gene: this is gene that codes for a protein that is involved in a signaling pathway and is important for cell

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## CLINICAL THYROIDOLOGY FOR THE PUBLIC

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### THYROID CANCER, continued

growth. Mutations in the BRAF gene in adults appear to cause 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.

Thyroidectomy: surgery to remove the entire thyroid gland. When the entire thyroid is removed it is termed

a total thyroidectomy. When less is removed, such as in removal of a lobe, it is termed a partial thyroidectomy.

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 (noncancerous) 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.

# **Thyroid Awareness Monthly Campaigns**

The ATA will be highlighting a distinct thyroid disorder each month and a portion of the sales for Bravelets<sup>™</sup> will be donated to the ATA. The month of June is **Differentiated Thyroid Cancer Awareness Month** and a bracelet is available through the <u>ATA Marketplace</u> to support thyroid cancer awareness and education related to thyroid disease.





