Thyroid

Papillary Thyroid Cancer Discovered After Benign Afirma Gene Testing, A Case Report

Mobeen Ahmad, MD, Franklin Thelmo, DO, and Peter Ucciferro, DO
Abington Jefferson Health, Abington, PA, USA

Introduction: Grave’s disease is an autoimmune form of hyperthyroidism characterized by antibody stimulation of the thyrotropin receptor. Although there are conflicting study results, patients suffering from Grave’s disease appear to have an increased incidence of thyroid cancer. Fine needle aspiration helps detect potential carcinoma, however, it is indeterminate in 15-30% of cases. In cases where fine needle aspiration of a thyroid nodule returns Atypia of Undetermined Significance (AUS) or Follicular Lesion of Undetermined Significance (FLUS), next-generation sequencing, including Afirma gene testing (created by Veracyte inc) provides guidance for therapy. However, like any screening test, the chance for false negatives still exists. We describe the case of a 41-year-old man suffering from Grave’s disease with a solitary thyroid nodule which was found to be papillary thyroid carcinoma despite benign Afirma gene testing. Case description: A 41-year-old male with no past medical history presented to his primary care provider with no active complaints but was found to have a blood pressure of 142/92 mmHg. He notably had a diffusely enlarged thyroid gland on examination. The patient was sent for thyroid function testing which demonstrated TSH: < 0.01 uIU/ml and a free T4: 1.1 ng/dL. The patient was sent for a thyroid ultrasound which demonstrated a right-sided nodule measuring: 3.5 cm x 2.4 cm x 3.2 cm (TI-RADS 4). Radioactive iodine uptake scan revealed this was a cold nodule and the patient underwent fine-needle aspiration biopsy and Afirma gene testing. The patient’s biopsy demonstrated AUS while Afirma gene testing was benign. After discussing treatment options for Grave’s disease with his endocrinologist, the patient underwent total thyroidectomy. Post-surgical biopsy of the patient’s thyroid demonstrated follicular variant papillary thyroid carcinoma with capsular invasion. The patient is currently being followed closely by endocrinology with regular TFT and thyroglobulin testing. Discussion: Grave’s disease is postulated to increase patients’ risk of developing papillary thyroid cancer. Many of the proposed hypotheses explaining this higher risk center around thyroid-stimulating antibodies promoting tumor invasion and angiogenesis. When these patients demonstrate suspicious thyroid nodules on imaging, they undergo fine-needle aspiration, however, these biopsies are indeterminate in 15-30% of cases. Afirma gene testing has been proposed as a sensitive test to rule out carcinoma in these indeterminate cases. We describe a case where, despite benign Afirma gene testing, the patient was found to have papillary thyroid cancer after thyroidectomy. We propose that Grave’s disease patients with suspicious nodules with indeterminate biopsy and benign Afirma testing still be considered for total thyroidectomy given their increased risk of thyroid cancer.

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