

## **GENETIC TESTING REVEALS THE MALIGNANT POTENTIAL OF A CYTOLOGICALLY BENIGN THYROID NODULE: A CASE REPORT.**

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**Background/Purpose:** The potential for malignancy in cytologically benign thyroid nodules is not truly known. This case report demonstrates the utility of genetic analysis in identifying thyroid cancer in a cytologically benign thyroid nodule.

**Methods:** A 50 y/o female being followed for a 2.5 cm thyroid nodule discovered at age 43 and found to be benign by fine needle aspiration biopsy (FNAB) cytology. The nodule was stable in size through age 49 as measured on annual visits. At age 50 the nodule was noted to have increased in size to 4 cm. She had no history of radiation to her head and neck nor history of thyroid cancer.

On examination, the 4 cm right-sided thyroid nodule moved freely with swallowing. Her voice was not hoarse, and no cervical adenopathy. A FNAB was performed which demonstrated benign adenomatous nodular goiter but genetic testing indicated an N-RAS mutation. She underwent partial thyroidectomy with removal of the right lobe. About 25% of the adenomatous goiter was involved with follicular variant of papillary thyroid carcinoma.

**Results:** The risk of malignancy in cytologically benign thyroid nodules is low; In this case, the growth of the thyroid nodule necessitated further testing despite benign cytologic result. The FNAB sample was tested for thyroid cancer fusion genes and proto-oncogenes. The genetic testing was positive for an N-RAS activating mutation found in thyroid cancer, and the final pathology at thyroidectomy revealed papillary thyroid cancer, present throughout the adenomatous goiter suggesting a malignant transformation of the benign nodule.

**Discussion & Conclusion:** We postulate that genetic testing of cytologically benign thyroid nodules may identify nodules at risk of malignant potential.