

# The role of detecting BRAF T199A mutation in fine-needle aspiration biopsy in pre-operative diagnosis of nodular goiter.

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## INTRODUCTION & OBJECTIVES

Molecular testing plays increasingly a significant role in the pre-operative diagnosis of nodular goiter, as standard methods may not give conclusive decision in choosing optimal treatment approach. BRAF gene mutations are often discovered in the cytological specimens among the patients with papillary thyroid cancer.

**The aim of the study** was to assess the incidence of BRAF T1799A mutation in cellular specimens derived from fine needle aspiration biopsy (FNAB) of thyroid nodules.

## METHODS

- **85 women** with nodular goiter were enrolled into the study.
- Using **hormonal tests, autoantibodies** and **ultrasound**, both hormonal thyroid dysfunction and autoimmune process were excluded.
- All the patients underwent **FNAB** of revealed nodules.
- **We analyzed genomic DNA** isolated from the thyroid lesions and peripheral blood.
- Standard methods of real-time amplification detection (real-time PCR) were used to **analyze BRAF mutation**, with the use of specific starters surrounding the mutated site.

## RESULTS

- We found BRAF T1799A mutation in thyroid specimens of 6 (7.05%) subjects.
- In 5 of them, benign nature of the thyroid nodules was confirmed by FNAB of the lesions.
- This procedure was non-diagnostic in 1 subject and histopathology post-operative assessment confirmed papillary thyroid cancer.

## CONCLUSIONS

**The presence of BRAF mutation in FNAB cytological specimens of benign thyroid nodules may be useful to evaluate the risk of malignancy, support the diagnosis and choose treatment options of nodular goiter.**

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