INTRODUCTION

Thyroid nodules are common in adults, but only a small fraction of them are malignant.

Fine-needle aspiration (FNA) cytology provides a diagnosis of benign versus malignant disease in majority of cases.

However 25% of nodules are cytologically indeterminate (figure 1), thereby needing repeat FNA or diagnostic lobectomy for definitive diagnosis.

Aim of this study was to evaluate diagnostic and clinical utility of molecular testing using the newly launched EnGenent thyroid cancer mutation panel

MATERIALS AND METHODS

21 cytologically indeterminate samples were used.

16 samples were collected in Cytolyt™ preservative, 2 in PreservCyt™ and 3 samples tested had both Cytolyt™ and FFPE cell block.

In addition, 2 FFPE blocks of positive controls (RET/PTC1 and PAx8/PPARy) along with 12 commercially available FFPE standards were also tested.

16 most common mutations in BRAF, KRAS, NRAS and HRAS and 3 rearrangements which include RET/PTC1, RET/PTC3 and PAx8/PPARy were detected by this assay (figure 2)

Cytolyt™ and PreservCyt™ samples were spun to cytoklyt capsular invasion AUS diagnosis

AUS diagnosis implemented by CPA lab

Three samples were positive for BRAF V600E mutation, 2 of which were suspicious for papillary carcinoma on cytology. (figure 3)

One sample that was diagnosed as atypia of undetermined significance was positive for NRAS codon 61 mutation whereas one suspicious for follicular neoplasm was positive for HRAS mutation.

The samples that were negative for mutations and translocations were cytologically either atypia of undetermined significance or follicular lesion of undetermined significance.

The assay sensitivity was 1% for all mutations except V600E mutation showed papillary thyroid carcinoma (PTC) (n=1), follicular variant of PTC (n=1) and follicular carcinoma (n=1).

RESULTS

Mutation Analysis Significantly Improves the Diagnostic Utility and Patient Management of Cytologically Indeterminate Thyroid Nodules

Figure 1: Risk of malignancy based on cytology diagnosis

Figure 2: Alterations detected by Thyroid mutation panel

Figure 3: Summary of results using Thyroid mutation panel

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CONCLUSIONS

 Thyroid cancer mutation panel is highly sensitive for detection of common genetic abnormalities seen in up to 50% of thyroid carcinomas.

 This test can be performed on routine cytology as well as FFPE tissue and does not require special collection medium or preservative.

 It improves the diagnostic yield of cytology and can therefore help in effective clinical management.

 Better preoperative stratification of patients with thyroid nodule malignancy risk

 Improved ability to guide initial definitive (partial/total) thyroidectomy