ThyroSeq V2 Application Study for Indeterminate Thyroid FNAs with Surgical Follow-up; Experience at a University Hospital

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Objectives: Fine needle aspiration (FNA) followed by cytological examination is a standard approach to detect thyroid cancer or identify a benign nodule. Results are reported as Bethesda Thyroid Categories (BTC). In about 25% of thyroid nodules, the presence of cancer cannot be ruled out by FNA cytology and is reported as indeterminate (BTC III, IV, and V). Targeted mutation detection by next generation sequencing in thyroid FNA has been applied to detect cancer-related genes and gene fusions. Molecular studies or profiling for the BTC III to V cases can be helpful in the clinical management of these patients. We report our preliminary experience with ThyroSeq V2, a targeted mutation detection system for thyroid specimens.

Methods: From January 2015 to April 2016 we examined 223 thyroid FNAs. 42 of the 223 cases (18.8%) classified as BTC III (n = 34), IV (n = 4), and V (n = 4) were submitted for ThyroSeq V2 tests at University of Pittsburgh, and rate of mutation was calculated. Follow-up surgical resection, ultrasound examination and other relevant clinical outcomes were also collected.

Results: Two category V, 1 category IV, and 4 category III cases were positive for different mutations. No mutations were found in other 35 cases. Subsequent surgical histology confirmed thyroid cancer in four patients with positive mutation. Two patients without mutations who had surgery had benign histology. The other patients who had negative mutation and did have follow-up for 3-6 months after FNA, showed no significant changes in the follow-up ultrasound.

Conclusions: Our preliminary results show that ThyroSeq V2 test could be a valuable complementary tool to enhance diagnostic accuracy in thyroid nodules reported as indeterminate in FNA cytology. Positive ThyroSeq V2 results have the potential to serve as a predictive indicator for thyroid cancer. Negative results in correlation with other findings can be helpful for patient management.