

6th International Conference on

DIABETES AND ENDOCRINOLOGY

December 05-07, 2016 Dallas, USA

Diagnostic value of sonography, FNAC and genetic alteration in diagnosis of malignant thyroid nodules

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Background: Thyroid nodule is common, malignancies found in \approx 9-15% of the nodules evaluated with FNAB (1). High resolution US and FNAB are the most important steps in diagnostic workup of thyroid nodule. 20–40% of FNABs yield undetermined cytology (2). Surgery is required in indeterminate FNAB, but only quarter of them prove to be malignant after surgery (3), so 77% underwent unneeded surgery. So, finding a mean to diagnose malignant nodule preoperative is important. Advances in molecular genetics can be applied to develop a new diagnostic markers for FNA samples.

Objective: Evaluating the diagnostic value of high resolution U/S, FNAC, and BRAFV600 in diagnosing malignant thyroid nodules.

Patients & Methodology: 50 patients from Kasr alainy endocrine outpatient clinic with solitary or multiple thyroid nodules. All subjected to full history and examination, thyroid profile, high resolution neck U/S, U/S guided FNAC and BRAFV600E analysis on FNAB using DNA sequencing then HRMA for confirmation.

Results: The incidence of BRAFV600E mutation among PTC patients was 55.6%, P value $<$ 0.001. The sensitivity of BRAFV600E mutation was 42.9%, specificity was 100%. Ultrasound sensitivity in detecting malignancy was 88.2%, specificity 72.7%. Addition of ultrasonography to BRAFV600E analysis increased ultrasound sensitivity in detecting PTC preoperative to 92%. There was a positive correlation between most suspicious ultrasonography findings and presence of BRAFV600E mutation (increased AP/T diameter, Increase intra-nodular blood flow, cervical lymphadenopathy, absent or incomplete halo (all p value $<$ 0.001), irregular border p value 0.004, micro calcifications p value 0.007).

Conclusion: 1. High resolution US and FNAB are the most important steps in diagnosis of thyroid nodule. 2-BRAFV600E mutation detected mainly in PTC. 3. Adding BRAFV600E analysis to U/S and FNA will increase the sensitivity of preoperative diagnosis of PTC especially in indefinite nodules. 4. BRAFV600E positivity was associated with most known suspicious U/S finding and significantly present in cases with cervical lymphadenopathy that may give it a prognostic value. 5. HRMA is simple, accurate, and a low cost tool for BRAFV600E analysis.

Biography

Marwa Sayed Abdel Latief Eassa graduated from Cairo University in 2005 with an excellent degree. She finished her BCS degree in Internal Medicine in Cairo University in 2010, and her thesis was insulin resistance as predictor for response to interferone based therapy in HCV infected patients. Currently, she is a MD candidate, and finished her MD thesis. She is working as Teaching Assistant in Internal Medicine Department, and in Endocrine Out Patient Clinic and Department in Kasralainy Hospital, Cairo University. She is a member of primary aldosteronism group undersupervision of Pro. Dr. Mona Mansour.

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