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Case Report
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MEDULLARY CARCINOMA OF THE THYROID: DIAGNOSTIC CHALLENGES IN A RARE THYROID MALIGNANCY

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ABSTRACT
Medullary thyroid carcinoma (MTC) is a rare neuroendocrine tumor arising from parafollicular C-cells. Early diagnosis significantly influences prognosis, especially in sporadic cases. We report a case of a 47-year-old male presenting with a firm thyroid swelling. Fine-needle aspiration cytology (FNAC) confirmed MTC (Bethesda category V), supported by characteristic cytological features and serum calcitonin testing. This case emphasizes the importance of cytological recognition, biochemical correlation, and molecular strategies in the management of MTC.

INTRODUCTION
Medullary thyroid carcinoma (MTC) represents 3–4% of all thyroid malignancies and arises from calcitonin-producing parafollicular C-cells. MTC may be sporadic or inherited (as part of MEN 2A/2B syndromes), with sporadic cases accounting for 75–80%.[1] FNAC remains the frontline diagnostic modality for evaluating thyroid nodules and may suggest MTC when characteristic cytomorphologic features are present. Serum calcitonin and carcinoembryonic antigen (CEA) are essential biochemical markers, and RET proto-oncogene mutation analysis informs familial screening and targeted therapy.[2,3]

Despite its rarity, MTC carries a risk of early lymphatic and distant spread, often to the liver, lungs, or bones, with rare reports of adrenal or bone marrow metastasis.

CASE PRESENTATION
A 47-year-old male presented to the outpatient department with a swelling in the right side of the neck. Examination revealed a firm, diffuse, non-tender thyroid swelling measuring approximately 6×4 cm, moving with deglutition.

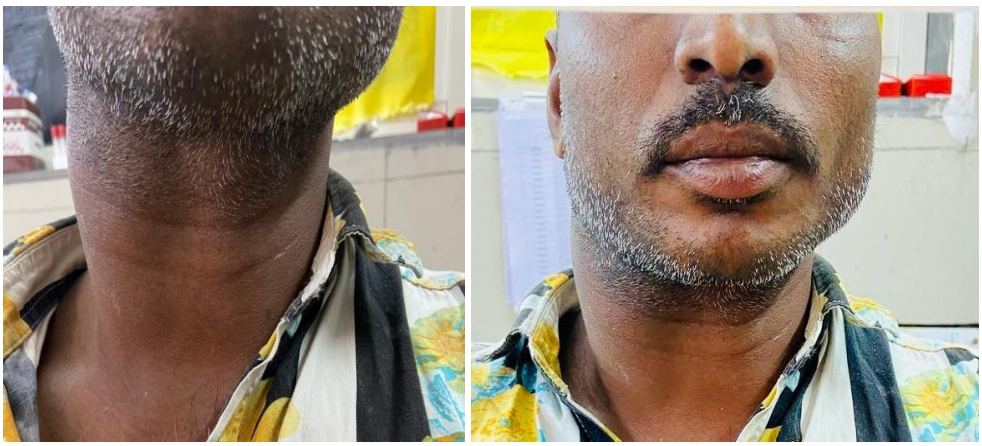

Figure 1A: Clinical photograph of the patient showing diffuse thyroid swelling predominantly on the right side 1B: Clinical Photograph of the patient showing thyroid swelling on the right side.

Investigation and Management Plan
Serum Calcitonin: Elevated at 272.9 pg/mL (reference range: 1.8–18.2 pg/mL), strongly suggestive of MTC. **Urinary VMA (Vanillylmandelic Acid):** 3.94 mg/24 hrs (reference: 1.4–6.5 mg/24 hrs), within normal range, reducing the suspicion of associated pheochromocytoma. **Calcium:** Normal at 10.02 mg/dL (reference: 8.6–10 mg/dL).

USG and CT Neck

- **USG Neck**

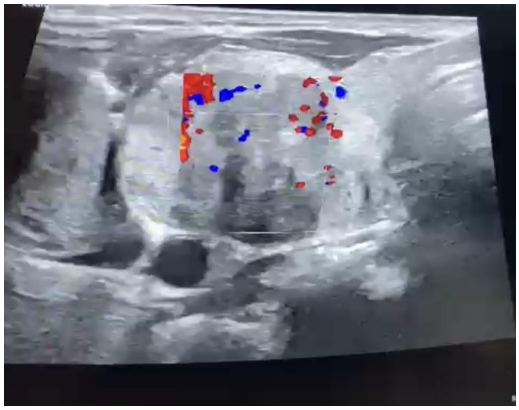

Figure 3: Ultrasound neck (color Doppler) showing a vascular, heteroechoic thyroid nodule with internal vascularity, consistent with malignant features.

- **CT Neck-** 16 × 25 mm right thyroid lobe nodule with inhomogeneous enhancement. Multiple enlarged cervical lymph nodes, with the largest measuring 17 mm. Involvement of level 2, 3, 4 lymph nodes; extension from C2 to C6.No abnormality in the left neck; suspicious nodal spread in mediastinum.

FNAC: Confirmed medullary thyroid carcinoma (Bethesda category V) with classic features—hyperchromatic nuclei, anisonucleosis, and amyloid-like material in the background.

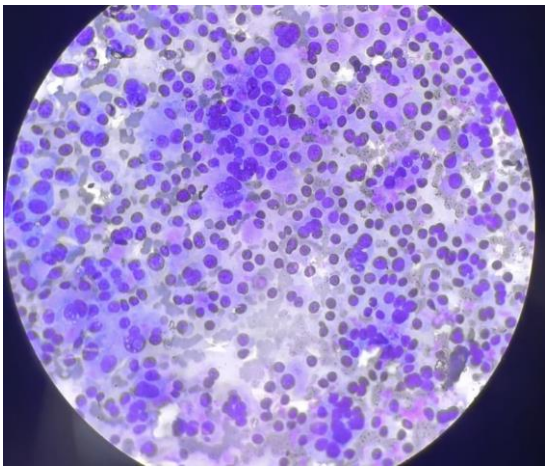

Figure 2A: Fine-needle aspiration cytology smear showing high cellularity with dispersed and clustered tumor cells displaying hyperchromatic nuclei, nuclear pleomorphism, and cytoplasmic granularity (Giemsa stain, 40×).

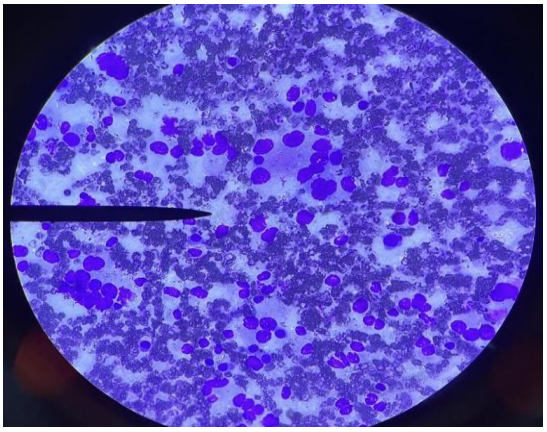

Figure 2B: FNAC smear (Giemsa Stain,40x) Showing hypercellularity, pleomorphic nuclei, cytoplasmic granularity and amyloid-like material- features characteristic of MTC.

Definitive Treatment: Planned total thyroidectomy with central neck dissection.
- Molecular Testing: RET mutation analysis and family screening planned.
- Targeted Therapy: In case of advanced/metastatic disease, consideration of tyrosine kinase inhibitors such as selpercatinib or cabozantinib.

DISCUSSION

MTC arises from neuroendocrine C-cells and has unique clinical and molecular characteristics. FNAC is instrumental in its early diagnosis, with key cytologic features such as plasmacytoid or spindle cells, nuclear pleomorphism, and red cytoplasmic granules.[2]

Serum calcitonin serves both as a diagnostic and prognostic marker. Doubling time is a recognized predictor of recurrence. Although lymph node involvement is common, MTC can rarely metastasize to adrenal glands or bone marrow, mimicking hematologic disorders.[4]

RET proto-oncogene mutations are found in nearly all hereditary and up to 50% of sporadic MTCs. Genetic testing has both therapeutic and familial implications. The advent of targeted therapies like selpercatinib (FDA approved 2024) and cabozantinib has transformed treatment paradigms for advanced MTC.[5]

CONCLUSION

This case underscores the value of FNAC in early diagnosis of MTC, supported by serum biomarkers and molecular evaluation. Timely diagnosis and a multidisciplinary approach—including surgery, genetic testing, and targeted therapy—are critical for optimizing patient outcomes.

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