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MEDULLARY THYROID CARCINOMA: FROM CLINICAL PRESENTATION TO COMPREHENSIVE MANAGEMENT

General Surgery

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ABSTRACT

We report a case of 58-year-old male who presented with swelling in the anterior neck since 1 year. Ultrasound Neck & Fine Needle aspiration Cytology revealed features suggestive of Medullary Carcinoma Thyroid. The patient underwent Total Thyroidectomy with Central Neck Dissection. The patient had an uneventful postoperative course & discharged in stable condition without any complications.

KEYWORDS

Medullary Carcinoma Thyroid, Papillary Carcinoma Thyroid, MEN Syndrome

INTRODUCTION:

Medullary thyroid carcinoma (MTC) is a rare neuroendocrine tumor that arises from the calcitonin-producing parafollicular C-cells of the thyroid gland, which are of neural crest origin. MTC accounts for approximately 5–10% of all thyroid malignancies and 0.4–1.4% of thyroid nodules. Unlike papillary and follicular carcinomas, which derive from follicular epithelium, MTC shares histological and biological features with other neuroendocrine tumors such as carcinoids and pancreatic islet cell tumors.

The great majority of cases (75–80%) are sporadic, while the remainder occur as part of hereditary syndromes transmitted in an autosomal dominant pattern: multiple endocrine neoplasia type 2A (MEN 2A, or Sipple syndrome), multiple endocrine neoplasia type 2B (MEN 2B, or Wagenmann–Froboese syndrome), and familial MTC (FMTC). These hereditary forms are driven by germline mutations of the RET proto-oncogene on chromosome 10q11.2, which encodes a transmembrane receptor tyrosine kinase expressed in neural crest–derived tissues. Sporadic MTC may also harbor somatic RET mutations in roughly half of cases, with the remainder showing RAS-family mutations (HRAS, KRAS, NRAS). MEN 2A is characterized by MTC together with pheochromocytoma and primary hyperparathyroidism, whereas MEN 2B additionally features mucosal neuromas and a marfanoid habitus and tends to follow a more aggressive, earlier-onset course.

C-cells secrete calcitonin as their principal product, along with carcinoembryonic antigen (CEA), chromogranin A, adrenocorticotrophic hormone, histaminases, and somatostatin. Serum calcitonin is the single most useful biomarker for diagnosis, operative planning, and postoperative surveillance, with reported sensitivity approaching 98–100% and specificity around 95%, although modest elevations can also occur in hypergastrinemia, hypercalcemia, other neuroendocrine neoplasms, and even benign goiter. CEA, while less specific, is a useful complementary marker, particularly in the minority of tumors that lose the capacity to secrete calcitonin. Because MTC does not arise from iodine-avid follicular cells, radioactive iodine has no role in its treatment or in metastasis surveillance, and the tumor generally responds poorly to conventional chemotherapy and radiotherapy — making early diagnosis and complete surgical resection the only realistic route to cure.

Case Presentation:

A 58 year old male patient presented to outpatient department with complaints of swelling in front of neck since 1 year. He is a farmer by occupation. On admission, his vital signs were stable with pulse rate 86 bpm, Blood pressure 110/80 mm of hg, Respiratory rate of 18 cpm, temperature of 98°F and SpO_2 of 98% at room air. On Inspection in sitting position, a swelling in front of neck measuring 6 x 6 cms, Butterfly in shape, Extending between both the sternocleidomastoids, swelling moves with deglutition, Lower border is visible. On palpation, A Swelling of size 6 x 4 cms, present in the front of neck, Surface is smooth, Firm in consistency, All borders are well defined including the lower border. USG of Neck revealed Punctate high echogenic foci resembling calcification. FNAC revealed Cellular specimen with round, ovoid, spindle cells in small cluster; cells have abundant cytoplasm and eccentric nuclei; chromatin has salt and pepper appearance , having pink azurophilic granules and intranuclear pseudoinclusions. Under general anaesthesia, with aseptic precautions a Kocher's incision was made 2 cms above the suprasternal notch in the skin crease. Total Thyroidectomy with Central Compartment Neck Dissection. The procedure lasted 3 hours with an estimated blood loss of 200 ml. Specimen was sent for Hispopathological evaluation. Postoperatively, the patient was managed with nil by mouth, intravenous fluids, antibiotics, analgesics, and antiemetics. The postoperative course was uneventful, and the patient was discharged in stable condition without any complications. Histopathological report revealed Medullary Carcinoma Thyroid.

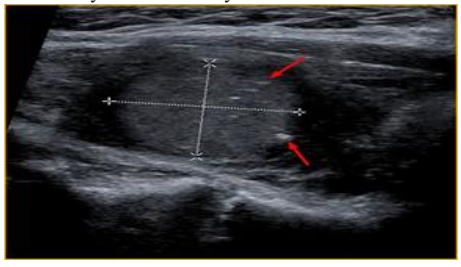

FIGURE 1: Ultrasonography images showing a large swelling arising from thyroid gland of size measuring 6 X 4 cms.

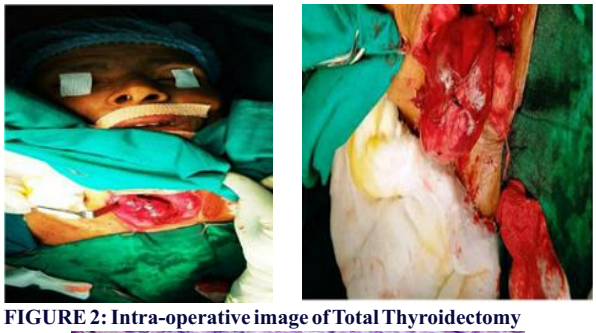

FIGURE 2: Intra-operative image of Total Thyroidectomy

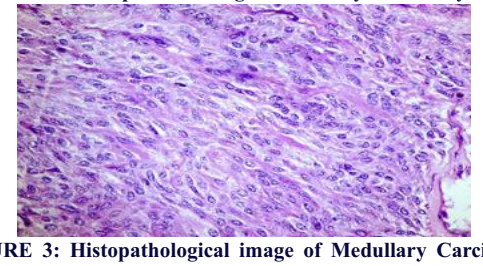

FIGURE 3: Histopathological image of Medullary Carcinoma Thyroid

DISCUSSION:

Diagnostic work-up

The initial evaluation of suspected MTC should combine history and examination with biochemical testing (serum calcitonin and CEA),

neck ultrasonography, and FNAC, followed by RET germline mutation testing and biochemical screening for coexisting pheochromocytoma in all newly diagnosed patients. Rezkallah et al. reported an FNAC sensitivity of only 72.7% for MTC in their series, consistent with the wider literature range of 12.5–88.2%, reflecting the diagnostic difficulty already illustrated by the misclassified Hürthle-cell-pattern oncocytic case. Serum calcitonin, by contrast, achieved a sensitivity of about 98% in the same series and is considered the single most reliable preoperative marker, with levels correlating broadly with tumor burden and nodal involvement; levels greater than 200 pg/mL have been proposed as a threshold favoring contralateral neck dissection, and levels greater than 500 pg/mL generally warrant cross-sectional imaging to exclude distant metastasis.

Surgical management

Total thyroidectomy is the treatment of choice for essentially all patients with MTC, reflecting the tumor's tendency toward multicentricity, its more aggressive biological course relative to differentiated thyroid cancers, and its complete unresponsiveness to radioactive iodine. In the Rezkallah series, all 11 patients underwent total thyroidectomy; ten also had central and lateral neck dissection, while the single patient with a known familial RET mutation underwent prophylactic thyroidectomy alone. Nodal disease was extensive at presentation (five patients had T3 disease and one had T4 disease), and four of eleven patients who tested RET-positive all had either multifocal or bilateral tumors — supporting the guideline-based recommendation that all patients with MTC undergo at least central compartment (level VI) dissection, extending to lateral compartments (levels II–V) when nodal disease is confirmed or strongly suspected. The Cureus case similarly required a radical, multicompartment resection (total thyroidectomy with right radical neck dissection and central compartment clearance) once high-grade cytology and bulky, matted lymphadenopathy were identified.

Postoperative management and follow-up

All patients require lifelong levothyroxine replacement after total thyroidectomy; because C-cells are not TSH-responsive, TSH suppression (as practiced in differentiated thyroid cancer) is not required. Postoperative surveillance relies on serial serum calcitonin and CEA measurements — recommended at three months, then every six months for a year, then annually if undetectable — since biochemical cure (normalized CEA and undetectable calcitonin) carries an excellent prognosis, with a five-year recurrence rate of only around 5%. Rising postoperative calcitonin above 150 pg/mL should prompt cross-sectional imaging (neck ultrasound, chest CT, liver MRI or triphasic CT, and bone imaging) to localize recurrent or metastatic disease. In the Rezkallah cohort, only two of eleven patients achieved complete biochemical remission, two developed biochemically confirmed local recurrence (managed surgically), and two died from advanced metastatic disease — outcomes that correlated closely with higher tumor stage and multiple nodal metastases at presentation, consistent with published AJCC/UICC stage-stratified five-year survival figures of roughly 100% (stage I), 93% (stage II), 71% (stage III), and 21% (stage IV) cited by Sumithra et al.

Diagnostic pitfalls and variant histology

The oncocytic variant of MTC described by Parampalli Srinivas et al. underscores that atypical histological variants can closely mimic other thyroid neoplasms — particularly Hürthle cell tumors — on cytology, risking misdiagnosis and inappropriate management, since MTC carries a distinct treatment pathway and the oncocytic subtype in particular is associated with a poorer prognosis than classical MTC. Recognition of neuroendocrine "salt-and-pepper" chromatin, together with confirmatory immunohistochemistry (positive calcitonin, CEA, synaptophysin, and chromogranin; negative thyroglobulin) and Congo red staining for amyloid, is essential whenever an oncocytic or Hürthle-cell-like lesion is encountered, especially in atypical demographic groups such as young male patients.

CONCLUSION:

Medullary thyroid carcinoma is an uncommon but biologically distinct thyroid malignancy arising from calcitonin-producing C-cells, occurring sporadically in the majority of cases but also as part of well-defined hereditary RET-driven syndromes. Across the case series and case reports reviewed here, the diagnosis hinged on a consistent combination of clinical suspicion, elevated serum calcitonin and CEA, ultrasonography, FNAC, and confirmatory immunohistochemistry, while unusual histological variants such as the oncocytic subtype highlight the need for pathological vigilance. Complete surgical resection — total thyroidectomy with appropriate central and lateral neck dissection guided by nodal status and biomarker levels — remains the only curative treatment, since MTC does not take up radioactive iodine and responds poorly to systemic therapy. Prognosis is closely tied to stage and completeness of resection at diagnosis, reinforcing the importance of early recognition, genetic screening of affected families, and structured lifelong biochemical surveillance to detect recurrence.

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