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Case Report

Metastatic medullary thyroid carcinoma masquerading as breast lesion: lessons from a rare case

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ABSTRACT

Medullary thyroid carcinoma (MTC), derived from parafollicular C-cells, accounts for 5–8% of thyroid malignancies. It has a propensity for hematogenous metastasis, commonly involving liver, lungs, and bones. Breast metastases from MTC are exceedingly rare, with about 40 documented cases reported worldwide. Herein this case describes a 25-year-old female who initially underwent total thyroidectomy and right cervical lymphadenectomy for MTC, staged as pT3bN1bM0 (stage IVA). Following surgery and adjuvant radiotherapy, she developed persistently elevated calcitonin levels. Subsequent Ga-68 DOTANOC PET-CT revealed local recurrence, but the patient declined re-exploration. Three years later, she presented with a breast lump, clinically and radiologically suspected to be fibroadenoma. Wide local excision with intraoperative frozen section revealed metastatic MTC. Immunohistochemistry confirmed the diagnosis (INSM1 positive, GATA3 negative). Breast metastasis from MTC is extremely uncommon and may mimic benign breast lesions, delaying diagnosis. This case highlights the importance of calcitonin surveillance, the role of intraoperative frozen section, and immunohistochemistry in differentiating metastatic MTC from primary breast tumors.

Keywords: Medullary thyroid carcinoma, Breast metastasis, Calcitonin, Rare metastasis, Case report

INTRODUCTION

Medullary thyroid carcinoma (MTC) arises from neural crest–derived parafollicular C-cells of the thyroid gland, accounting for approximately 5-8% of thyroid cancers. In contrast to papillary thyroid carcinoma, which comprises nearly 80% of thyroid malignancies, MTC follows a more aggressive course with a higher likelihood of hematogenous dissemination.[1,2]

Distant metastases most commonly affect liver, lungs, and bones.[3] Breast metastases are exceptionally rare, with about 40 documented cases in the literature, as highlighted in the most recent review by Saadallah et al.[4,5]

We present a case of a young female who developed breast metastasis from MTC four years after primary diagnosis, underscoring the diagnostic challenges posed by such unusual presentations.

CASE REPORT

A 25-year-old female initially presented in September 2014 with a thyroid swelling and right cervical lymphadenopathy. Ultrasound revealed a 2.8×1.5 cm heterogeneous vascular nodule with microcalcifications in the right thyroid lobe and multiple enlarged cervical lymph nodes (levels II–V). Total thyroidectomy with right-sided neck dissection was performed. Histopathology confirmed medullary carcinoma involving the right lobe and isthmus with minimal extrathyroidal extension (pT3bN1bM0, stage IVA). The patient received adjuvant external beam radiotherapy (33 fractions). At the third year of follow-up, serum calcitonin was elevated (932 pg/ml). Ga-68 DOTANOC positron emission tomography-computed tomography (PET-CT) demonstrated a 2.2×1.5 cm recurrence in the thyroid bed. The patient declined operative management after detailed counselling, primarily due to concern about the risk of temporary or

permanent vocal cord palsy. Three years later (2018), she presented with a 3×3 cm firm lump in the upper outer quadrant of the left breast. Serum calcitonin had risen to 2115 pg/ml. Ultrasonography suggested fibroadenoma; PET-CT showed a residual thyroid lesion (2.4×1.3 cm, SUVmax 4.1), and a well-defined breast lesion with a possibility of fibroadenoma.

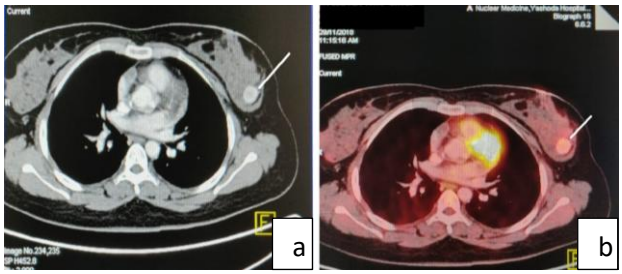

Figure 1 (a and b): PET-CT images.

Wide excision of the breast lump was performed. Intraoperative frozen section suggested metastatic MTC. Resection margins were clear. Immunohistochemistry demonstrated positivity for INSM1 and negativity for GATA3, confirming thyroid origin.

DISCUSSION

MTC metastasizes both via lymphatic and hematogenous routes. Approximately 60% of patients present with nodal disease, and 40% eventually develop distant metastases.[6] The breast is a highly unusual metastatic site, with around 40 cases reported in the literature to date.[4,7] Distinguishing primary breast carcinoma from metastasis is crucial, as management and prognosis differ significantly. Serum calcitonin serves as a highly sensitive biomarker for MTC recurrence and metastatic progression.[8] In our case, rising calcitonin levels prompted advanced imaging and guided suspicion of metastatic disease. PET-CT provided further evaluation but failed to identify the breast lesion as malignant, highlighting the limitations of radiology alone. Histopathology with immunohistochemistry remains the gold standard. INSM1, a neuroendocrine marker, and calcitonin positivity support thyroid origin, whereas GATA3 negativity rules out primary breast carcinoma.[9] Management of metastatic MTC depends on the site and resectability. Surgical excision remains appropriate for isolated or symptomatic lesions. Systemic therapy options include tyrosine kinase inhibitors such as Vandetanib and Cabozantinib, particularly for unresectable or progressive disease.[10] This case emphasizes the need for long-term surveillance with serum biomarkers and imaging in MTC patients, as late and unusual metastatic patterns may occur.

CONCLUSION

Breast metastasis from MTC is exceedingly rare and may clinically mimic benign or primary breast tumors. Calcitonin monitoring, frozen section, and immunohistochemistry are vital in reaching an accurate diagnosis. Awareness of such atypical presentations can aid timely management and improve outcomes.

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