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Thyroid

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A Case of Metastatic Medullary Thyroid Cancer Presenting with Pathologic Hip Fracture

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Introduction: Medullary thyroid cancer accounts for less than 5% of thyroid malignancies. Distant metastatic disease typically presents in lymph nodes, lung, liver and bone. We present a patient who initially presented with a pathologic hip fracture with bone biopsy revealing medullary thyroid carcinoma. **Case Description:** A 44-year-old male with no past medical history presented to the emergency department for right hip pain following a mechanical fall. Imaging showed a right femoral neck pathologic fracture as well as lytic lesions within the right femoral neck and right ilium. Family history was significant for pancreatic cancer and breast cancer. The patient underwent bone biopsy of the right femur followed by total hip arthroplasty. Bone biopsy revealed metastatic medullary thyroid carcinoma. Thyroid ultrasound showed a 1.2 centimeter right solid hypoechoic nodule along with extensive cervical lymphadenopathy. Further imaging revealed metastatic disease throughout the thoracic spine, mediastinal lymphadenopathy, and a left 2.2 centimeter adrenal lesion with peripheral enhancement and possible necrosis. Carcinoembryonic antigen (CEA) level was 551 ng/mL (normal <4.7), calcitonin was 2,498 pg/mL (normal <10), and calcium levels were normal. Plasma free metanephrines were normal and plasma normetanephrines were less than two times the upper limit of normal. Genetic testing on the bone biopsy specimen revealed a RET p. C618S variant. Germline RET testing was collected. The patient was discharged with close follow-up with oncology to initiate systemic therapy and follow-up with endocrinology for the adrenal mass. **Discussion:** Majority of cases of medullary thyroid carcinoma are sporadic. Patients typically present with a thyroid nodule with many having cervical lymph node involvement at diagnosis and 5-10% having distant metastatic disease. Our patient's initial presentation was unique in that he presented with a pathologic fracture. Bone metastases can be seen in about 30% of cases of metastatic medullary thyroid carcinoma. Calcitonin levels greater than 500 pg/mL require additional imaging to exclude metastatic disease. If RET mutational status is unknown, patients should be screened for primary hyperparathyroidism and pheochromocytoma. Screening for pheochromocytoma is critical in preoperative management of these patients. Treatment for patients with metastatic disease involves surgery, external beam radiotherapy, and kinase inhibitors.

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