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CASE REPORT

From Neck to Mid-Chest: Unraveling the Anatomical Extent of Medullary Thyroid Cancer- A Case Report

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ABSTRACT

Medullary thyroid cancer, or MTC, is an uncommon type of thyroid malignancy. It originates from a specific group of cells within the thyroid gland called calcitonin-producing C cells. An even rarer complication of MTC is when the cancer extends beyond the thyroid gland and reaches the area behind the breastbone, known as the retrosternal space. This specific scenario is estimated to occur in only 0.2-0.8% of all thyroid cancers associated with MEN syndromes or a positive family history We report the case of an older man who had difficulty breathing and a large swelling on the right side of his neck. A needle aspiration biopsy revealed MTC, and he was treated accordingly. While large thyroid glands have been documented, our case of MTC with extension from the upper right neck to the mid-chest, not associated with MEN syndrome additionally, is one of the infrequent cases to account as the prognosis for retrosternal MTC is generally worse than for MTC that occurs in the neck only. However, early diagnosis and treatment can improve the chances of survival.

Key Words: Calcitonin, medullary thyroid carcinoma, multiple endocrine neoplasia, Parafollicular cells

INTRODUCTION: Medullary thyroid cancer, a rare form of thyroid carcinoma (around 2-4% of all thyroid cancers), has the ability to produce the hormone calcitonin. In roughly 1 out of 5 MTC cases (20%), an underlying genetic defect is the culprit. This defect is due to change in a gene called the RET proto-oncogene[1]. There are two main presentations of MTC. When it appears on its own, it's called sporadic MTC or erratic MTC. However, MTC can sometimes occur alongside tumors present in the parathyroid gland and the adrenal glands (specifically pheochromocytoma), it's classified as multiple endocrine neoplasia type 2 (MEN 2). The documented case of MEN 2 dates back to 1959[2].

Patients with MTC may need evaluation and management of any co-existing hormonal imbalances by an endocrinologist. This could involve treating conditions like pheochromocytoma, a tumour on the adrenal gland. Once these related issues are addressed, the recommended treatment for MTC itself is a complete removal of the thyroid gland (total thyroidectomy). This surgery often includes removing lymph nodes in the central neck area for further examination. In cases where lymph node metastasis has occurred to other levels, other level of the neck dissection is imperative. Serum calcitonin along with radiological imaging such as chest X-rays and CT-scan is important for detecting metastasis. As serum calcitonin is a specific and sensitive marker for MTC recurrence, annual measurements are highly recommended[3].

An elevated serum calcitonin level, which had previously been low after a complete thyroid removal, suggests the recurrence of medullary thyroid cancer. In ideal scenarios, surgical intervention aims to eliminate all lymph nodes along with entire thyroid gland in the neck that harbour metastatic spread.

Case Report: A 60-year-old male developed a gradually enlarging mass on the right side of his neck over a two-year period. Six months later, he developed progressive shortness of breath, particularly during physical exertion. On examination, a prominent, non-pulsatile neck mass was observed predominantly on the right side, with dilated blood vessels and no visible pulsation. (Fig 1) There was no dysphagia, hoarseness, stridor or signs of thyrotoxicosis. Laryngoscopy using a fibre-optic device demonstrated normal movement of both vocal cords. The patient's medical and surgical records indicated no relevant past conditions. There was no familial predisposition to such disorders.

CT scan showed a outsized, irregularly shaped mass extending from the right side of the neck and into the

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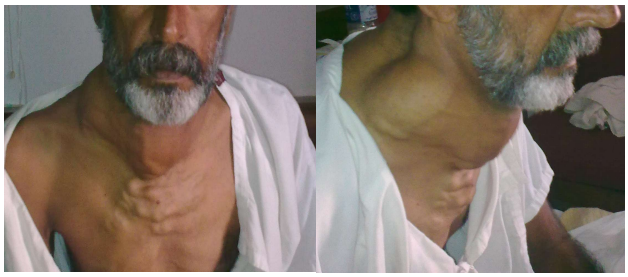

Figure 2: CT Scan Shows Thyroid Mass With Retrosternal Extension in the Anterior Mediastinum Over Arch of Aorta

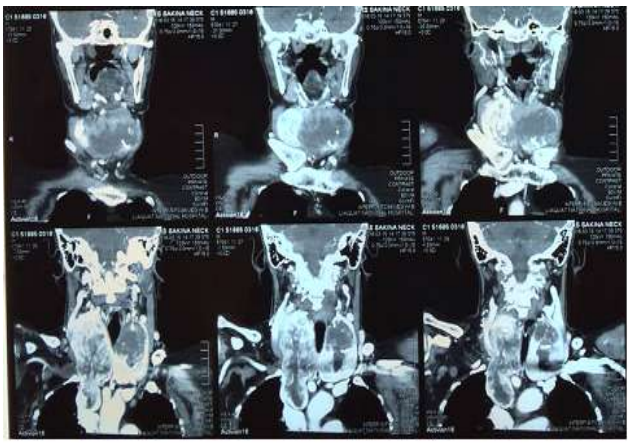

Figure 1: Patient With Neck Lumps Showing Huge Thyroid With Retrosternal Extension and Engorged Vessels

chest. (Fig 2) and up to the right main pulmonary artery compressing the larynx trachea on the contralateral side. The mass was located primarily in the anterior mediastinum, impinging upon and compacting the left brachiocephalic vein, a major blood vessel situated superior to the aortic arch. (Fig 2) This mass was 9x11.3cm transversely and 19 cm craniocaudally. FNA was indicative of medullary thyroid carcinoma. His MEN syndrome workup was negative.

The patient uneventfully underwent a total thyroidectomy, central neck dissection, and removal of the mediastinal mass via a median sternotomy. His postoperative recovery proceeded smoothly, and he was closely monitored for eight months, with no evidence of disease recurrence. Subsequently, he was lost to follow-up.

DISCUSSION: In about 75% of cases, MTC occurs sporadically, while the remaining cases are familial. Familial MTC is associated with multiple endocrine neoplasia (MEN) type 2A and 2B syndromes, but it can also occur independently of MEN. In the United States, MTC accounts for about 2-4% of all thyroid cancers[4]. However, in Asia, the incidence of MTC is significantly lower, with an estimated rate of 1.34 per 100,000 people. A faulty gene (RET) can cause some families to have a higher risk of MTC. Testing for this gene mutation is recommended for MTC patients with a family history or diagnosed young. If the test shows a high-risk mutation, preventive thyroid removal (thyroidectomy) might be an option[5].

While erratic medullary thyroid carcinoma (MTC) classically shows in the fifth or sixth decade, and those associated with multiple endocrine neoplasia (MEN) 2A or 2B typically presents earlier in life. Clinical presentation of MTC might include a lump in the neck, swallowing difficulties (dysphagia), voice changes (hoarseness), or trouble breathing (respiratory distress). Exceptionally, some may show symptoms of carcinoid syndrome. High levels of calcitonin can trigger excessive fluid secretion in the intestines, leading to diarrhoea. Patients with distant metastases may experience symptoms like weight loss, fatigue, and bone pain.

Surgical intervention is the main stay therapy for the MTC. Concomitant pheochromocytoma must be addressed before proceeding for surgery. The standard treatment involves removing the entire thyroid gland (thyroidectomy) and nearby lymph nodes in the central neck. If cancer has spread to the lymph nodes in the neck, a more extensive lymph node removal (modified radical neck dissection) might be needed. Postoperative external beam radiation therapy and chemotherapy may be beneficial for selected patients, though their effectiveness is still debated[5,6]. Monitoring serum calcitonin levels postoperatively helps detect persistent MTC, with elevated levels indicating its presence.

Our case is exceptional in that we were unable to locate any published accounts of such an extensive medullary thyroid gland spanning the neck to the mediastinum, especially one not associated with MEN syndrome

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