Journal of Population Therapeutics & Clinical Pharmacology

CASE REPORT
DOI: 10.53555/jptcp.v31i7.7224

CASE REPORT: METASTATIC MEDULLARY THYROID CANCER PRESENTING IN A YOUNG MALE

Hassam Ali[1*], Tayyaba Munir[2], Muhammad Akbar Sultan[3], Ahmad Rafique[4], Luqman Ahmad[5], Humna Shah Nawaz[6], Shawwal Amjad[7], Fatima Iqbal[8]

[1*]General Surgery Department, Shalamar Hospital, Lahore, Pakistan.
Email: hassamali136@gmail.com
[2]General Surgery Department, Lahore General Hospital, Lahore, Pakistan.
Email: doctabmunir@gmail.com
[3]General Surgery Department, Lahore General Hospital, Lahore, Pakistan.
Email: akbarkhan786997@gmail.com
[4]General Surgery Department, Lahore General Hospital, Lahore, Pakistan.
Email: ahmadrafique2000@gmail.com
[5]General Surgery Department, Lahore General Hospital, Lahore, Pakistan.
Email: lascpmc2017@gmail.com
[6]General Surgery Department, Shalamar Hospital, Lahore, Pakistan.
Email: shah.nawaz.humna1998@gmail.com
[7]Medicine and Allied department, Khawajgan Trust Hospital, Gujrat, Pakistan.
Email: amjadshawal@yahoo.com
[8]General Surgery Department, Allied Hospital, Faisalabad, Pakistan.
Email: fatimaiqbal4173@gmail.com

***Corresponding Author:** Hassam Ali
*General Surgery Department, Shalamar Hospital, Lahore, Pakistan.
Email: hassamali136@gmail.com

INTRODUCTION:

Thyroid cancers stand out as the most prevalent among endocrine system malignancies, contributing to about 3.4% of all diagnosed cancers worldwide[1]. A distinct type within this category is medullary thyroid cancer, originating from parafollicular C cells traced back to neural crest origins. Upon microscopic examination of these cells, a unique amyloid backdrop becomes evident. These tumors have the intriguing ability to generate a range of substances, including calcitonin, carcinoembryonic antigen, serotonin, and chromogranin[2]. While around 75% of cases arise sporadically, the remaining 25% are tied to familial factors. Sporadic instances can emerge at any age, whereas the hereditary form tends to affect children and young adults, giving the disease a multifaceted demographic[3]. Typically, these cancers introduce themselves as solitary nodules within the thyroid. Notably, they often spread to cervical lymph nodes, leading to recognizable neck lymphadenopathy. Clinical presentation involves a blend of traditional thyroid nodule symptoms, along with flushing and diarrhea – indicative of metastatic progression. Intriguingly, about a quarter of MTC cases appear in individuals with an inherited multiple endocrine neoplasia syndrome. In the realm of thyroid cancers, medullary thyroid cancer (MTC) constitutes a relatively rare subset, accounting for only 1-2% of all thyroid cancer instances[4]. Herein lies a case study that delves into one such occurrence of medullary thyroid cancer in a young patient.

CASE REPORT

A 26 year old Pakistani male presented in the outpatient department with painless swelling of 5*4 cm in the anterior aspect of the neck along with restricted neck movements on the left side for the last 6 years. He also complained of pain in right shoulder joint for 1 month and weight loss of 3kg. Past medical and surgical history was insignificant. There was no family history of such presentation. On examination, the swelling originated from left anterior triangle of the neck, extending to the posterior triangle. It moved on deglutition. Trachea was shifted to the left side. There was no visible scar, pulsation or overlying skin discoloration. On palpation of the swelling it had smooth surface, hard consistency and was adherent to Sternocleidomastoid muscle. Lower limit was reachable. Systemic examination was insignificant.

Serum calcitonin levels were 3277pg/mL

Ultrasound suggested highly suspicious thyroid malignancy with ipsilateral cervical adenopathy. Thyroid scan revealed a cold nodule in the right lobe. CT scan head and neck showed a 4.7*3cm mass arising from right lobe of thyroid compressing the tracheal rings. Extensive right cervical and superior mediastinal lymphadenopathy with matted large necrotic lymph nodes along the right cervical vessels measuring 6.2*2.3cm. A 9*7mm lesion was also seen in left lobe of thyroid. Isthmus was thickened. Multiple soft tissue nodules were seen in bilateral lung fields.

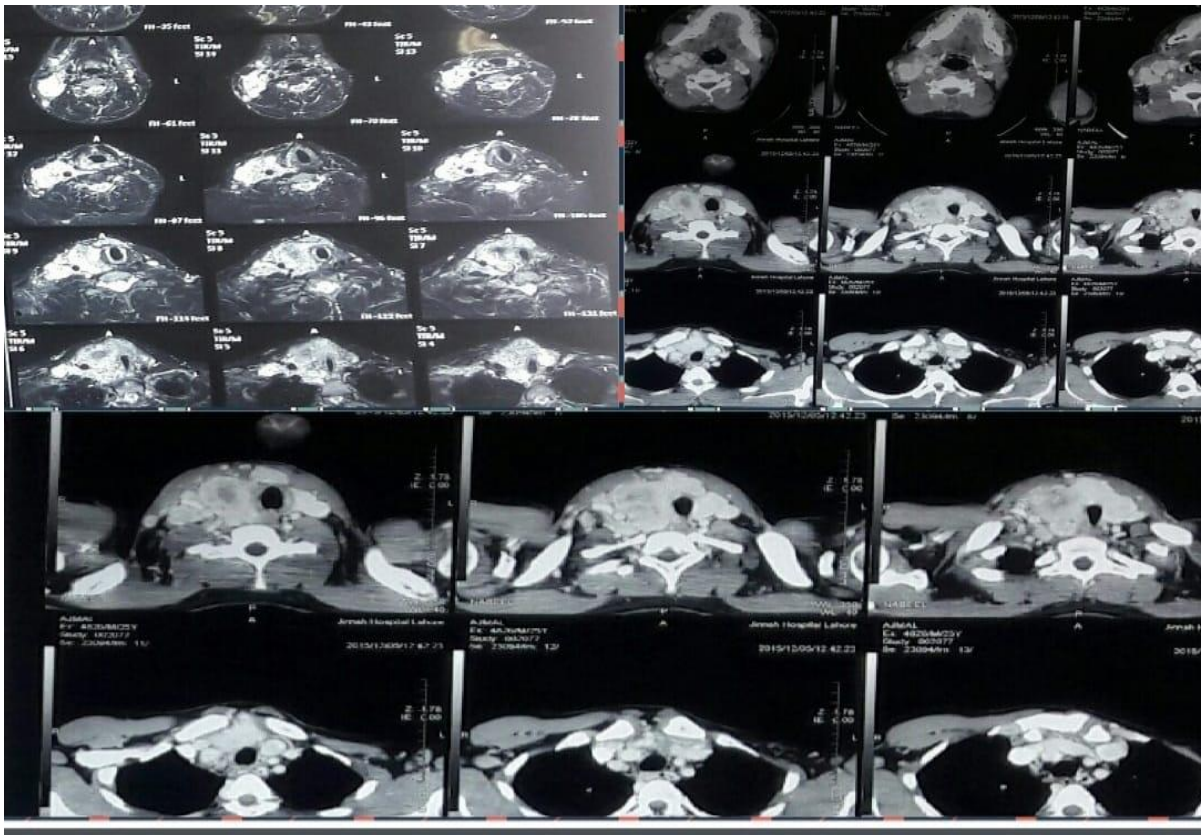

Figure 1: CT SCAN HEAD AND NECK

Fine needle aspiration cytology of right level IV cervical lymph node revealed atypical cells with a high N/C ratio, some showing nuclear molding, salt and pepper chromatin pattern with dendritic cytoplasmic processes. A diagnosis of metastatic medullary carcinoma Bethesda category-VI was made.

Right lobectomy + isthemectomy along with right sided classical block neck dissection with R2 resection was performed.

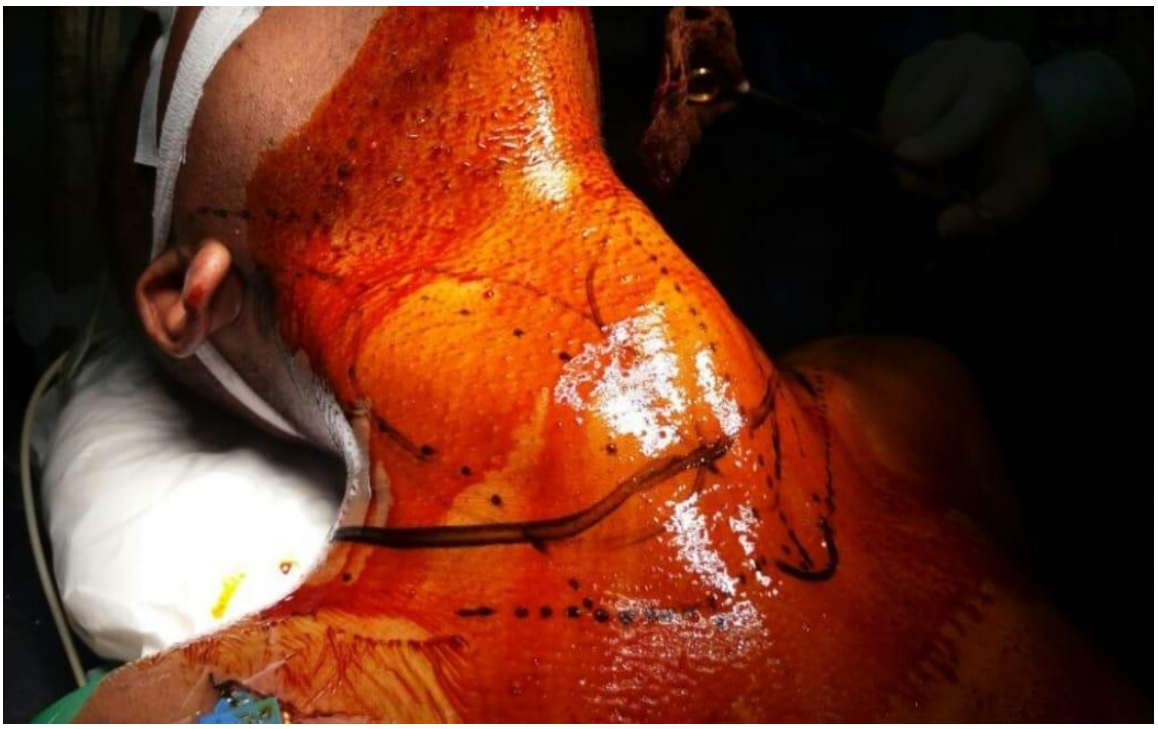

Figure 2: DEMARCATION OF THYROID LOBES ALONG WITH ANTISEPSIS AND DRAPING.

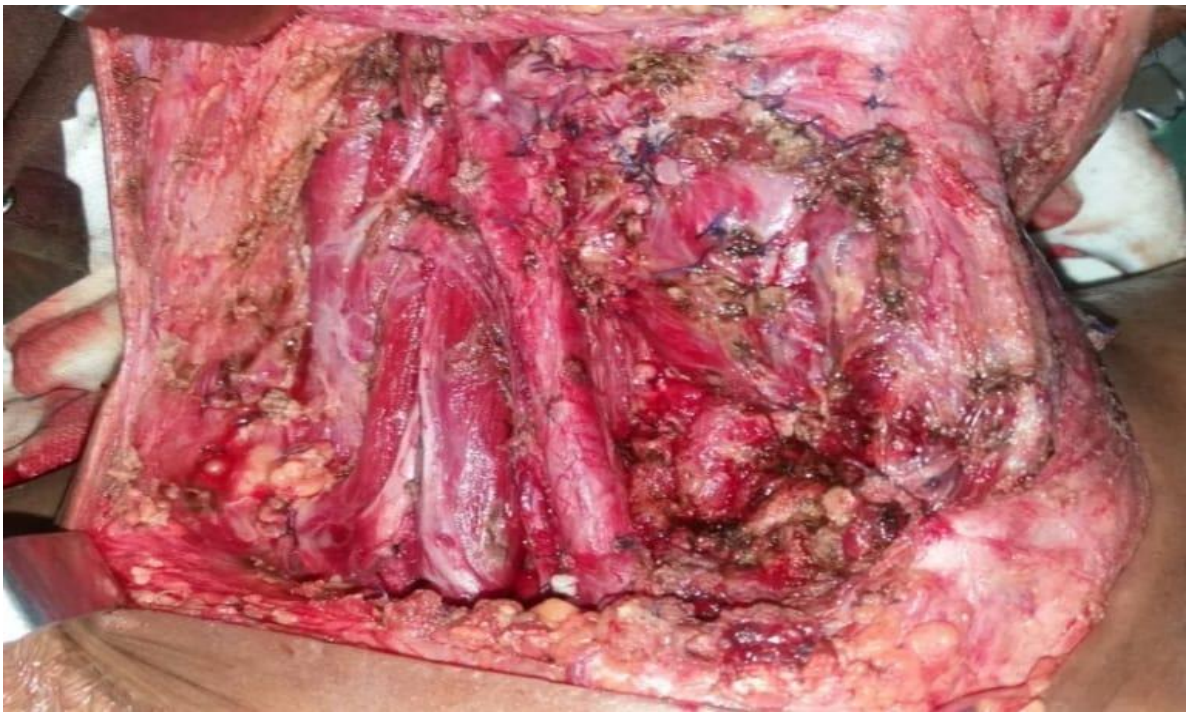

Figure 3: INTRAOPERATIVE NECK DISSECTION.

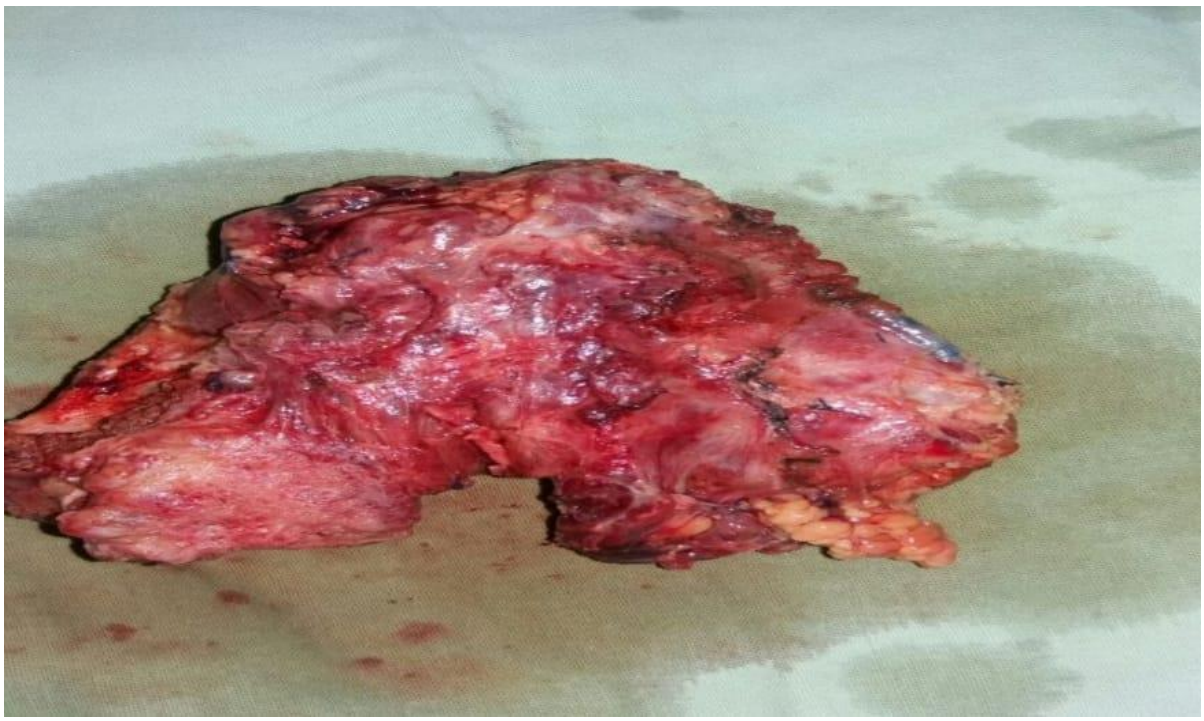

Figure 4: HISTOPATHOLOGY SPECIMEN

Histopathology report revealed medullary carcinoma thyroid T3N1bMx. 3/24 lymph node positive for metastatic carcinoma plus 4 tumor nodules. Follow up was done after 14 days and Thyroxine along with appropriate antibiotics and painkillers were advised.

DISCUSSION

The incidence of medullary thyroid cancer (MTC) in the demographic of young adults aged 20-24 is characterized by a relatively low occurrence, approximating 0.6 cases per million per annum. This malignancy manifests diversely, with familial instances attributed to germ line RET mutations, while somatic RET mutations drive sporadic cases. Predominantly, the somatic landscape is marked by the frequently encountered M918T mutation, linked to a discernibly unfavorable prognostic profile. Patients harboring this mutation are often confronted with a challenging clinical trajectory characterized by suboptimal outcomes[5]. Somatic RET mutations tend to impart a focal impact, commonly precipitating unilateral thyroid lobe involvement. In contrast, germline mutations elicit a distinct bilateral manifestation of MTC[6]. The diagnosis of MTC involves a combination of clinical, biochemical, and imaging findings. Measurement of serum calcitonin levels is a critical step in the diagnostic workup. Fine-needle aspiration cytology (FNAC) biopsy of the thyroid nodule and immunohistochemistry can help confirm the presence of C-cells and distinguish MTC from other thyroid cancers. Genetic testing for RET mutations is also essential to determine hereditary cases and guide treatment decisions[7].

Surgery is the primary treatment for MTC. The extent of surgery depends on factors such as tumor size, stage, and the presence of metastases. In young patients, total thyroidectomy with central lymph node dissection is typically performed for localized disease. In hereditary cases with known RET mutations, prophylactic thyroidectomy may be considered for individuals at high risk to prevent the development of MTC. After surgery, additional treatments, such as radioactive iodine therapy or targeted therapies like tyrosine kinase inhibitors, may be used in advanced or metastatic cases[8]. After treatment, patients with MTC require regular follow-up and monitoring to detect any signs of disease recurrence or metastasis. Periodic measurements of calcitonin and carcinoembryonic antigen (CEA) levels are commonly used for surveillance. Imaging studies, such as ultrasound, CT scans, or MRI, may be performed to assess the status of the thyroid bed and to detect any evidence of residual or recurrent disease. The prognosis of MTC varies depending on the stage at diagnosis, the presence of

metastases, and whether it is sporadic or hereditary. Generally, hereditary cases tend to have a worse prognosis compared to sporadic MTC. Early detection, appropriate surgical intervention, and long-term follow-up are crucial for improving outcomes[9]. Previous case reports showed that hereditary MTC patients were significantly younger than sporadic MTC patients. Cytology and calcitonin (CT) assay were used for diagnosis, with CT assay showing higher sensitivity. Patients diagnosed through genetic screening had better outcomes and were diagnosed at an early stage. The cure of medullary thyroid cancer involves total thyroidectomy as the curative form of therapy. For patients with advanced or metastatic disease, systemic therapies such as tyrosine kinase inhibitors may be considered. Additionally, biomarkers like procalcitonin (PCT) play a role in the diagnosis and surveillance of medullary thyroid cancer, helping with early detection and monitoring of the disease's progression.

CONCLUSION

In conclusion, medullary thyroid cancer is a rare and distinct form of thyroid cancer that arises from the C-cells of the thyroid gland. Early diagnosis, appropriate surgical management, and long-term monitoring are essential for improving outcomes in patients with MTC. For individuals with a family history or known genetic mutations associated with hereditary MTC, genetic testing and counseling are crucial for identifying at-risk individuals and implementing appropriate preventive measures.

REFERENCES

1. Chmielik E, Rusinek D, Oczko-Wojciechowska M, et al. Heterogeneity of Thyroid Cancer. Pathobiology. 2018;85(1-2):117-129. doi:10.1159/000486422
2. Maxwell JE, Sherman SK, O'Dorisio TM, Howe JR. Medical management of metastatic medullary thyroid cancer. Cancer. 2014;120(21):3287-3301. doi:10.1002/cncr.28858
3. Elisei R, Schlumberger MJ, Müller SP, et al. Cabozantinib in progressive medullary thyroid cancer [published correction appears in J Clin Oncol. 2014 Jun 10;32(17):1864]. J Clin Oncol. 2013;31(29):3639-3646. doi:10.1200/JCO.2012.48.4659
4. Cabanillas ME, McFadden DG, Durante C. Thyroid cancer. Lancet. 2016;388(10061):2783-2795. doi:10.1016/S0140-6736(16)30172-6
5. Ying AK, Huh W, Bottomley S, Evans DB, Waguespack SG. Thyroid cancer in young adults. Semin Oncol. 2009;36(3):258-274. doi:10.1053/j.seminoncol.2009.03.009
6. Machens A, Niccoli-Sire P, Hoegel J, et al. Early malignant progression of hereditary medullary thyroid cancer. N Engl J Med. 2003;349(16):1517-1525. doi:10.1056/NEJMoa012915
7. Leimbach RD, Hoang TD, Shakir MKM. Diagnostic Challenges of Medullary Thyroid Carcinoma. Oncology. 2021;99(7):422-432. doi:10.1159/000515373
8. Grubbs EG, Rich TA, Li G, et al. Recent advances in thyroid cancer. Curr Probl Surg. 2008;45(3):156-250. doi:10.1067/j.cpsurg.2007.12.010
9. Hadoux J, Pacini F, Tuttle RM, Schlumberger M. Management of advanced medullary thyroid cancer. Lancet Diabetes Endocrinol. 2016;4(1):64-71. doi:10.1016/S2213-8587(15)00337-X