Neck lump and Cough: The Uncommon Encounter

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A 73-year-old lady with a large midline neck mass [Figure 1] present for the last 40 years presented to the OPD with complaints of a sudden increase in its size and weight loss of about 10 kilograms over the last four months, dry cough, and progressive dysphagia for the past ten days, and shortness of breath for the past three days. There was hoarseness of voice but no stridor. There was no history of fever or sore throat in the recent past. The neck lump was found to be a hard, nodular mass extending from the lower border of the thyroid cartilage to the sternal notch with encasement of unilateral carotid vessels and skin invasion, suggested by dermal oedema. The trachea and the bilateral cervical lymph nodes could not be appreciated because of the large size of the goitre and induration. There was no discharge, signs of inflammation or visible pulsation over the mass.

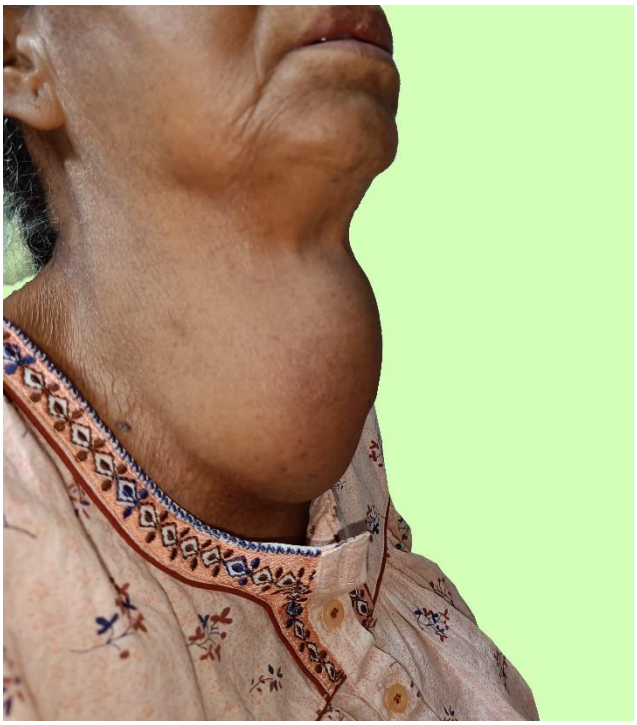

Figure 1: Showing the patients neck with mass.

Ultrasonography of the neck suggests a large, heterogeneous, predominantly hypoechoic lesion replacing the entire thyroid gland with a few small cystic areas within and a few foci of calcifications (TIRADS-5) [Figure 2]. Chest X-ray showed multiple patchy opacities throughout both lung fields, giving a cannonball appearance, right-sided tracheal deviation, and bilateral mild pleural effusion [Figure 3]. The neck CT scan suggested a large hypo-enhancing space occupying lesion (SOL) from the thyroid gland infiltrating bilateral sternocleidomastoid muscles associated with enlarged level VI lymph nodes. CECT thorax suggested multiple randomly distributed nodules in both lung fields and multiple enlarged mediastinal lymph nodes with mild bilateral pleural effusion.

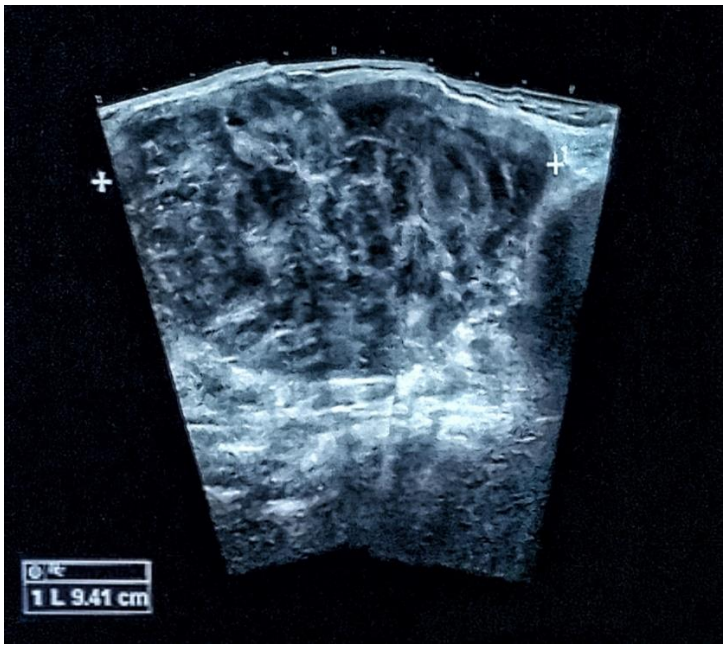

Figure 2: Ultrasound showing large, heterogeneous, predominantly hypoechoic lesion replacing the entire thyroid gland with a few small cystic areas.

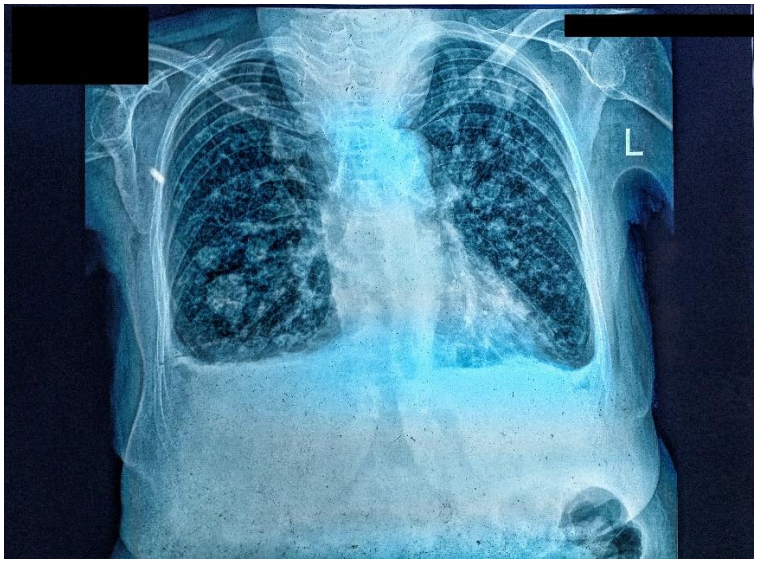

Figure 3: Chest X-ray showed multiple patchy opacities throughout both lung fields

FNAC from the neck mass showed high cellularity with focal clusters of remarkable atypical cells and multiple giant cells. Mitotic figures were prominent. The patient did not give consent for the core biopsy procedure.

Questions

1. What is your diagnosis?

2. What is the stage of this thyroid cancer?

3. What are the immediate palliative measures you would like to adopt?

4. What are the targeted therapies of benefit?

Answers

1. Anaplastic carcinoma of the thyroid with metastatic lung disease.

2. Stage IV C

3. Isthmusectomy with tracheostomy.

4. The targeted therapies of benefit are as follows:

    - Dabrafenib/trametinib (For BRAF V600E mutation positive).

    - Larotrectinib or Entrectinib (For NTRK gene fusion-positive).

    - Pralsetinib or Selpercatinib (For RET gene fusion-positive).

    - Other recommended chemotherapy regimens are Paclitaxel and Doxorubicin-based

Discussion

Anaplastic thyroid carcinoma (ATC), a highly aggressive tumour with nearly 100% mortality, is mostly diagnosed in females at age 70. Its incidence has declined due to improved thyroid cancer management and increased iodine intake. Emerging from dedifferentiation of other thyroid tumours, often linked to p53 protein loss, ATC's clinical signs include rapid neck mass growth, dyspnea, dysphagia, and distant metastases, mainly to the lungs and pleura.

ATCs have diverse morphologies, including features like biphasic spindle and giant cell tumors. Diagnosis is done by core or surgical biopsy. Additionally, larynx examination and imaging (ultrasound, CT scans) are done to assess tumor extension and invasion. PET/CT or MRI aids in staging. Testing for actionable mutations (BRAF, NTRK) is advised for targeted therapy.[1]

The prognosis of ATC is bleak, with a median survival of 5 months, often due to airway obstruction or complications. Age, metastases, and symptoms affect prognosis. Recent studies show improved survival with specific treatments like targeted therapy and surgery after neoadjuvant BRAF-targeted therapy.

ATCs usually respond poorly to conventional therapies, including Radioactive Iodine therapy and hence, palliative and supportive care should commence early for better survivability.[2]

Treatment modalities vary by disease stage:

STAGE IA & IB

- Resectable:
    - Neoadjuvant dabrafenib/trametinib for BRAF V600E-mutated disease.
    - Total Thyroidectomy with lymph node dissection, followed by adjuvant EBRT (External Beam Radiation Therapy) with radiosensitising chemotherapy for R0/R1 resection and EBRT with chemotherapy for R2 resection.
- Unresectable or borderline resectable:
    - Molecularly targeted neoadjuvant therapy or EBRT with chemotherapy.
    - Surgery considered post-neoadjuvant chemotherapy response.

Stage IV C

a. Aggressive Therapy includes:

- Total Thyroidectomy with lymph node dissection if resectable (R0/R1).
- Tracheostomy and steroids if indicated.
- Local radiation therapy and chemotherapy.

b. Palliative Care:

- Palliative local radiotherapy.
- Local excision or radiotherapy.
- Tracheostomy and supportive management.

After initial therapy, contrast CT/MRI scans of the chest, neck, abdomen, pelvis, and brain are crucial for disease monitoring. FDG-PET scan may follow at 3-6 months post-initial therapy. If there is no further disease evidence, continue monitoring and consider palliative local radiotherapy, disease control measures, or second-line chemotherapy/systemic therapy.[1]

Preferred options include dabrafenib, entrectinib, pralsetinib or selpercatinib, with alternative paclitaxel, doxorubicin, and carboplatin regimens.

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