CASE REPORT

Ectopic thyroid tissue in the superior mediastinum: a rare presentation

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ABSTRACT

Background: Ectopic thyroid tissue (ETT) can occur anywhere along its embryological descent, most commonly at the tongue base and neck. While superior mediastinal occurrence is extremely rare, such cases are usually asymptomatic and detected incidentally. Differentiating ETT from other masses in the mediastinum can be challenging using conventional imaging, and thyroid scintigraphy provides valuable functional information for accurate diagnosis and management.

Case Presentations: In our first case, a 24-year-old female presented with a complaint of swelling in the neck region. The thyroid scan with pertechnetate injection was performed, which showed foci of abnormal increased uptake inferior to the normal thyroid gland, corresponding to soft tissue nodules in the superior mediastinum on correlative single-photon emission computed tomography/computed tomography (SPECT/CT). In our second case, a 32-year-old female presented with a complaint of neck swelling. A focus of abnormal increased uptake was noted inferior to the thyroid gland on thyroid scan. Correlative SPECT/CT revealed the abnormal uptake to be a soft tissue nodule in the superior mediastinum with a similar Hounsfield unit to the orthotopic thyroid gland, suggestive of ETT.

Conclusion: ETT in the superior mediastinum, in the presence of a normally located thyroid gland and normal thyroid function tests, is a very rare finding. The combined use of thyroid scintigraphy and SPECT/CT in such cases allows precise anatomical and functional characterization of the tissue, thereby guiding appropriate management and potentially avoiding unnecessary invasive procedures.

Keywords: Ectopic thyroid tissue, superior mediastinum, thyroid scintigraphy, single photon emission tomography/computed tomography, case report.

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Introduction

The thyroid gland originates from an endodermal thickening in the floor of the primitive pharynx and descends anteriorly in the neck during embryological development. Ectopic thyroid tissue (ETT) may be located anywhere along this migratory path, extending from the foramen cecum at the base of the tongue to the mediastinum [1,2]. The lingual region is the most common site of ETT, followed by other cervical locations. In contrast, ETT within the superior mediastinum is exceptionally rare [3]. These cases are frequently asymptomatic and are often identified incidentally during imaging performed for unrelated indications. The occurrence of ETT in the superior mediastinum remains a rare entity, with only a limited number of cases documented in the literature.

Case 1

A 24-year-old female presented with a complaint of swelling in the neck region. Thyroid function tests revealed her to be euthyroid. Ultrasonography of the neck demonstrated a solid-cystic nodule in the upper part of the right thyroid lobe. A thyroid scan with pertechnetate injection was performed, 20 minutes after the injection, to assess the functional status, which showed a photon-deficient area corresponding to the right lobe nodule. Additionally, a focus of abnormal increased uptake was noted inferior to the thyroid gland, corresponding to a soft tissue nodule in the superior mediastinum with a density of 85 Hounsfield unit (HU) on correlative single-photon emission computed tomography/computed tomography (SPECT/CT); similar to that of the orthotopic thyroid gland (Figure 1).

Case 2

A 32-year-old female presented to the outpatient department with a complaint of neck swelling. She was biochemically euthyroid. Ultrasound (USG) neck showed a solid nodule in the mid to lower part of the right thyroid

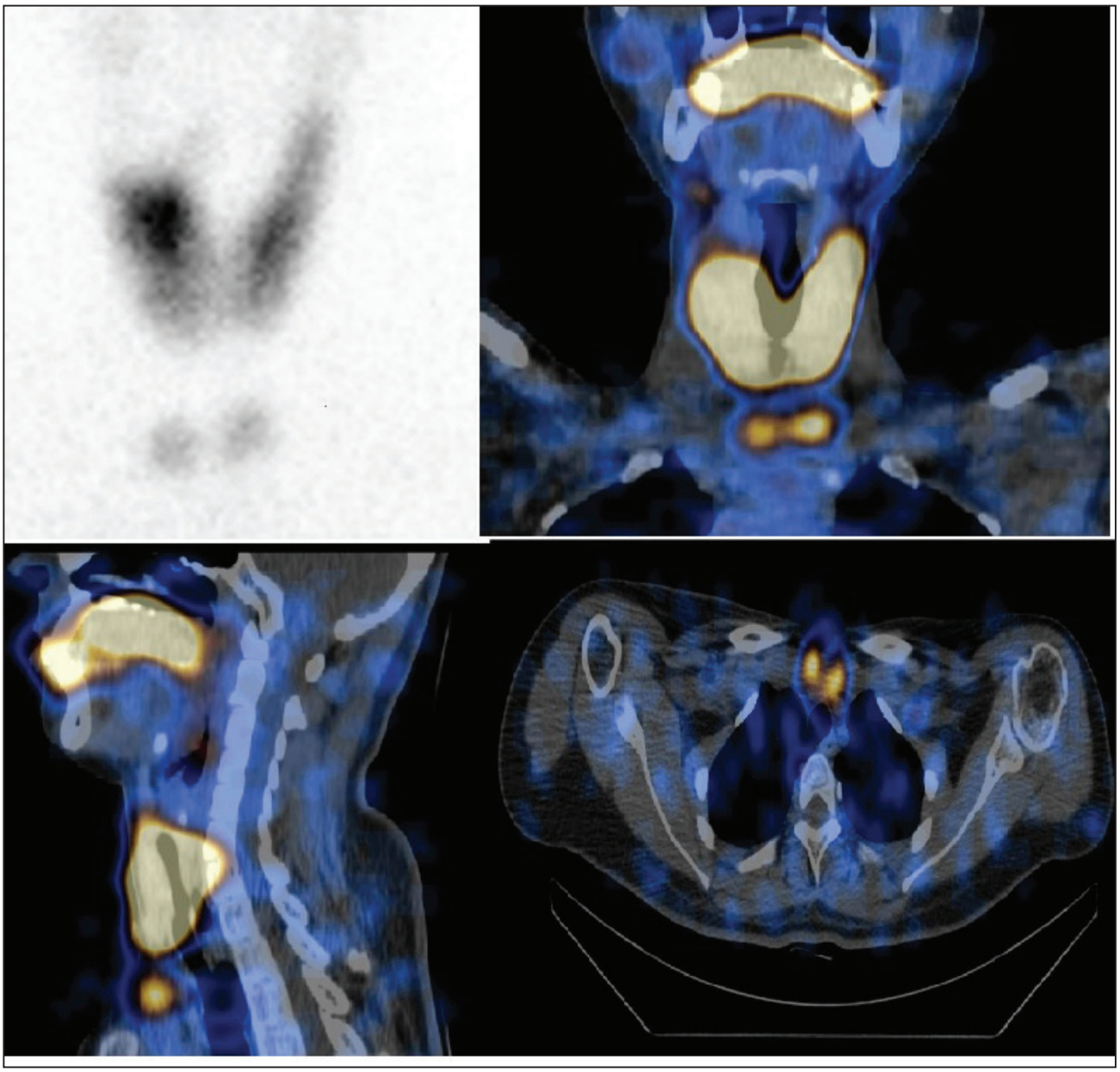

***Figure 1**. Detection of ectopic thyroid tissue in the superior mediastinum via thyroid scan and SPECT/CT imaging.*

lobe, which appeared as a cold nodule on the thyroid scan. Additionally, a focus of abnormal increased uptake was noted inferior to the thyroid gland. Correlative SPECT/CT for anatomical localization and characterization revealed that this abnormal focal uptake corresponded to a soft tissue nodule in the superior mediastinum with an HU value of 79; similar to that of the orthotopic thyroid gland, suggestive of ETT (Figure 2).

Discussion

The presence of thyroid tissue at a location other than its normal anatomical position, anterolateral to the second to fourth tracheal rings, is referred to as ETT. This rare developmental anomaly results from the failure of the thyroid gland to descend properly from its embryologic origin at the pharyngeal floor to its final pre-tracheal position in the anterior neck. The estimated prevalence of ETT in the general population is approximately 1 in 100,000 to 300,000, though this rate increases significantly among individuals with pre-existing thyroid disorders, where it is reported to be as high as 1 in 4,000 to 10,000 cases [4].

The most common site of ETT is the lingual region, accounting for up to 90% of cases. In contrast, ETT within the mediastinum is extremely rare. ETT may be present either in isolation or coexisting with a normally located thyroid gland [5]. Embryologically, the thyroid gland reaches its final location in the anterior neck by the seventh week of gestation, developing into a bilobed structure as it matures [6]. ETT results from aberrations during this migratory process and may be found anywhere along the path of the thyroglossal duct. In rare cases, it may even appear in more distant locations [7]. Recent studies

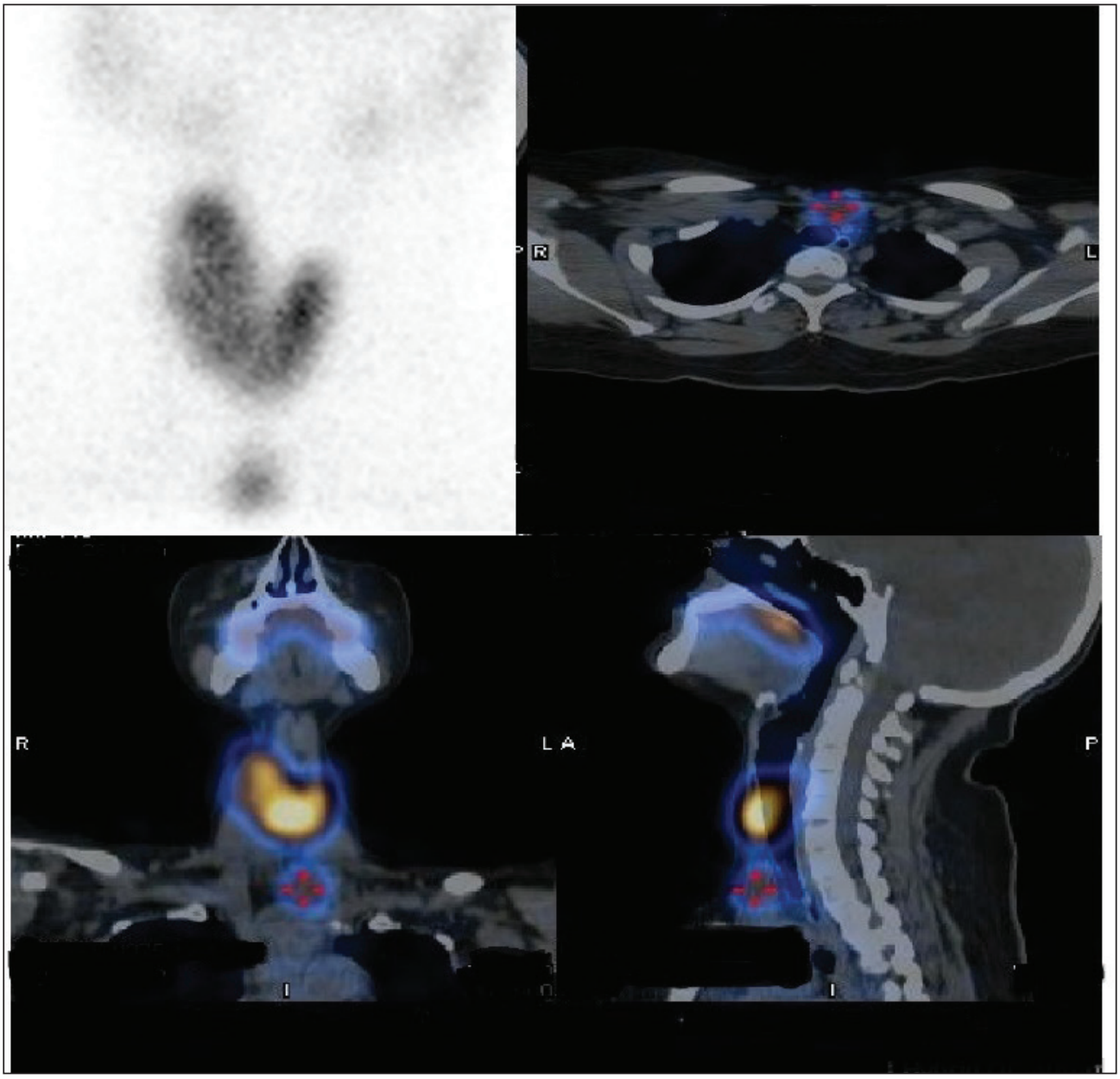

***Figure 2**. Thyroid scan and SPECT/CT reveal ETT in the superior mediastinum.*

suggest that mutations in specific genes involved in thyroid differentiation and migration may also contribute to the development of ETT [8].

In the case of mediastinal ETT, over-descent of thyroglossal duct remnants has been proposed as the primary mechanism [9]. Diagnosis of ETT can be made using various imaging modalities, including USG, computed tomography, and magnetic resonance imaging. However, the gold-standard diagnostic tool remains radionuclide thyroid scintigraphy using technetium-99m, which can confirm functioning thyroid tissue in ectopic sites. The differential diagnosis for mediastinal tissue includes lymph node, mediastinal ectopic thyroid, and substernal goitre. Substernal goitre typically maintains continuity with the normally located thyroid gland, while ectopic tissue is isolated from the native gland [5].

In our case presentations, hybrid imaging with SPECT/CT confirmed the presence of ETT in the superior mediastinum without any anatomical connection to the thyroid gland located in its normal position. This not only confirmed the diagnosis of mediastinal ETT but also effectively differentiated it from substernal goitre, lymphadenopathy, and other mediastinal masses. These cases underscore the diagnostic value of SPECT/CT in identifying ETT in uncommon locations, aiding in precise localization and tissue characterization, which are critical for appropriate clinical management.

Conclusion

ETT in the superior mediastinum, in the presence of a normally located thyroid gland and normal thyroid function tests, is a very rare finding. The combined use of thyroid

scintigraphy and SPECT/CT in such cases allows precise anatomical and functional characterization of the tissue, thereby guiding appropriate management and potentially avoiding unnecessary invasive procedures.

Conflict of interests

The authors declare that there is no conflict of interest regarding the publication of this article.

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Consent for publication

Permission was obtained from the patients to publish the case and the accompanying images.

Ethical approval

Ethical approval is not required at our institution to publish an anonymous case report.

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