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Sublingual Ectopic Thyroid Gland Misdiagnosed as Atrophic Hashimoto's Thyroiditis: A Case Report

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Abstract

BACKGROUND: Ectopic thyroid gland is a rare developmental anomaly caused by aberrant migration of thyroid tissue during embryogenesis. Misdiagnosis as autoimmune thyroiditis, particularly the atrophic form, may occur due to overlapping clinical features, especially in the hypothyroid state.

CASE PRESENTATION: We report the case of a patient who was treated for autoimmune thyroiditis for more than 20 years. Comprehensive imaging with ultrasound and SPECT/CT ultimately revealed an ectopic sublingual thyroid gland, with no orthotopic thyroid tissue present.

CONCLUSION: This case underscores the importance of thorough imaging evaluation in patients with hypothyroidism and absent thyroid tissue in the normal anatomical location, to avoid prolonged misdiagnosis and inappropriate management.

Introduction

Ectopic thyroid tissue (ETT) is a rare congenital anomaly resulting from aberrant migration or defective embryological development of the thyroid gland. Under normal conditions, the thyroid originates at the foramen cecum and descends along the thyroglossal duct to reach its definitive pretracheal position in the neck by the seventh week of gestation. Disruption of this process leads to the presence of thyroid tissue outside its typical anatomical location—a condition termed ectopic thyroid tissue [1], [2], [3]. The prevalence of ectopic thyroid is estimated to be between 1 in 100,000 and 300,000 individuals, with a marked female predominance. It is more frequently observed in Asian populations [2], [3]. The most common site of ectopic thyroid is the lingual region, accounting for approximately 90% of cases, followed by sublingual, subhyoid, and infrahyoid locations. Less frequently, ectopic thyroid tissue may be found in the mediastinum, lateral cervical regions, or in distant sites such as the adrenal glands and ovaries (struma ovarii) [3], [4].

Clinically, ETT can be asymptomatic and incidentally detected or may manifest with symptoms of hypothyroidism or local compressive effects, such as dysphagia, dysphonia, dyspnea, or airway obstruction, depending on its size and anatomical location [4]. Due to overlapping clinical and biochemical features, ectopic thyroid tissue is frequently misdiagnosed as autoimmune thyroiditis, particularly the atrophic variant of Hashimoto's thyroiditis. The latter is characterized by chronic autoimmune destruction of thyroid parenchyma, with the atrophic form representing an advanced stage marked by fibrosis, glandular shrinkage, and often positive thyroid autoantibodies [5]. Both conditions typically present with elevated thyroid-stimulating hormone (TSH) and require lifelong levothyroxine therapy, making clinical differentiation challenging. Accurate diagnosis requires high-resolution cervical ultrasound and functional imaging techniques such as radionuclide scintigraphy or

SPECT/CT for definitive localization of thyroid tissue [6], [7].

Case Presentation

A 45-year-old woman presented for a routine endocrinology follow-up with a longstanding diagnosis of hypothyroidism, managed with levothyroxine 100 µg daily for over 20 years. The initial diagnosis, presumed to be autoimmune thyroiditis (atrophic variant), had been made based solely on elevated TSH and low free T4 levels. No thyroid autoantibodies were detected at the time, and no thyroid imaging—specifically ultrasound—had been performed to confirm the diagnosis.

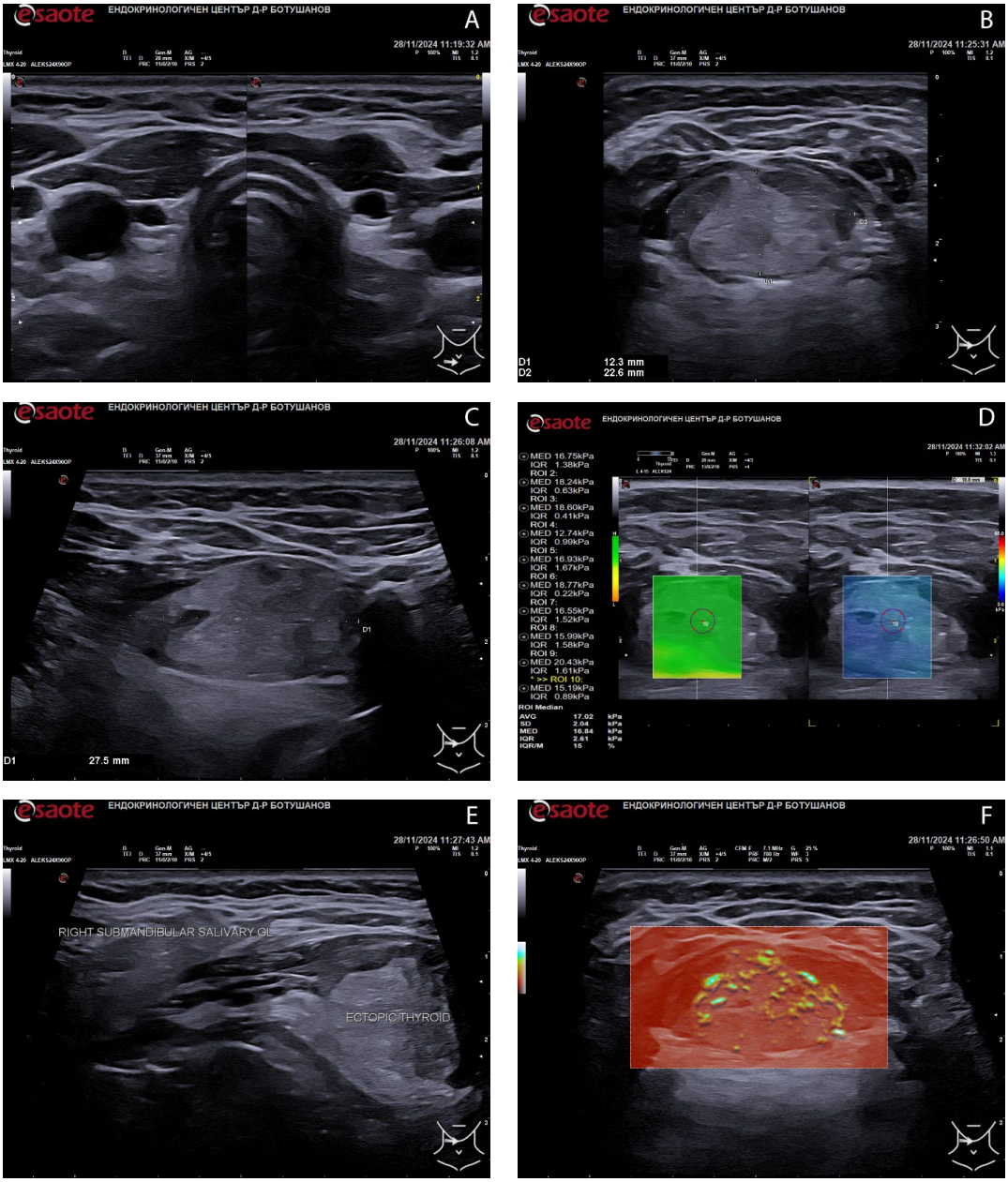

Figure 1: High-resolution ultrasonography of the anterior neck. A. Empty thyroid bed B. & C. Sublingual heteroechogenic mass D. 2D Shear-wave elastography of the sublingual mass E. Relative location of the sublingual mass to the right submandibular salivary gland F. Microvascularization of the sublingual mass

Routine thyroid function and autoimmunity tests were repeated (Table 1), revealing normal TSH and free T4 levels and persistently negative thyroid autoantibodies.

Table 1: Laboratory Results

Parameter	Result	Reference Range
TSH	0.95 µIU/mL	0.27 – 4.20 µIU/mL
Free T4	17.4 pmol/L	12 – 22 pmol/L
Anti-TPO antibody	<10 IU/mL	0 – 34 IU/mL
Anti-Tg antibody	<15 IU/mL	0 – 115 IU/mL

Due to the absence of a palpable thyroid gland and negative autoantibodies, a comprehensive cervical ultrasound was performed for the first time. The scan revealed no thyroid tissue in the pretracheal region, indicating an empty thyroid bed. However, a well-defined, heteroechogenic, vascularized mass measuring 13.4 × 22.5 × 26.3 mm was identified in the sublingual region.

Two-dimensional shear-wave elastography showed intermediate stiffness (average 17.02 kPa, SD 2.04 kPa, median 16.84 kPa, IQR 2.61 kPa, IQR/M 15%), consistent with benign thyroid tissue rather than fibrotic or malignant lesions (Figure 1).

The patient reported no significant symptoms aside from mild fatigue, which was well controlled with ongoing thyroid hormone therapy. She had no history of neck surgery, radiation exposure, or family history of thyroid disease.

On physical examination, she was clinically euthyroid under current treatment. No thyroid tissue was palpable in the cervical region, and there were no visible or palpable neck masses or lymphadenopathy. The clinical condition was stable, with no compressive symptoms.

To confirm the functionality of the ectopic tissue identified on ultrasound, a hybrid single-photon emission computed tomography/computed tomography (SPECT/CT) scan was performed following administration of Technetium-99m pertechnetate. The imaging revealed focal radiotracer uptake corresponding to the mass in the sublingual region, confirming the presence of functional ectopic thyroid tissue. No uptake was observed in the pretracheal thyroid bed or in other ectopic locations, thereby ruling out thyroid agenesis and confirming isolated sublingual ectopy (Figure 2).

Discussion

Differentiating ectopic thyroid tissue (ETT) from autoimmune thyroiditis is of critical importance due to the significant differences in diagnostic approach and long-term management. Both conditions may present with clinical hypothyroidism and absent palpable thyroid tissue, leading to diagnostic ambiguity. However, autoimmune thyroiditis is typically characterized by the presence of circulating thyroid autoantibodies and progressive parenchymal atrophy, whereas ectopic thyroid tissue often presents with persistently negative autoantibodies and a stable or slowly progressive clinical course [5], [6].

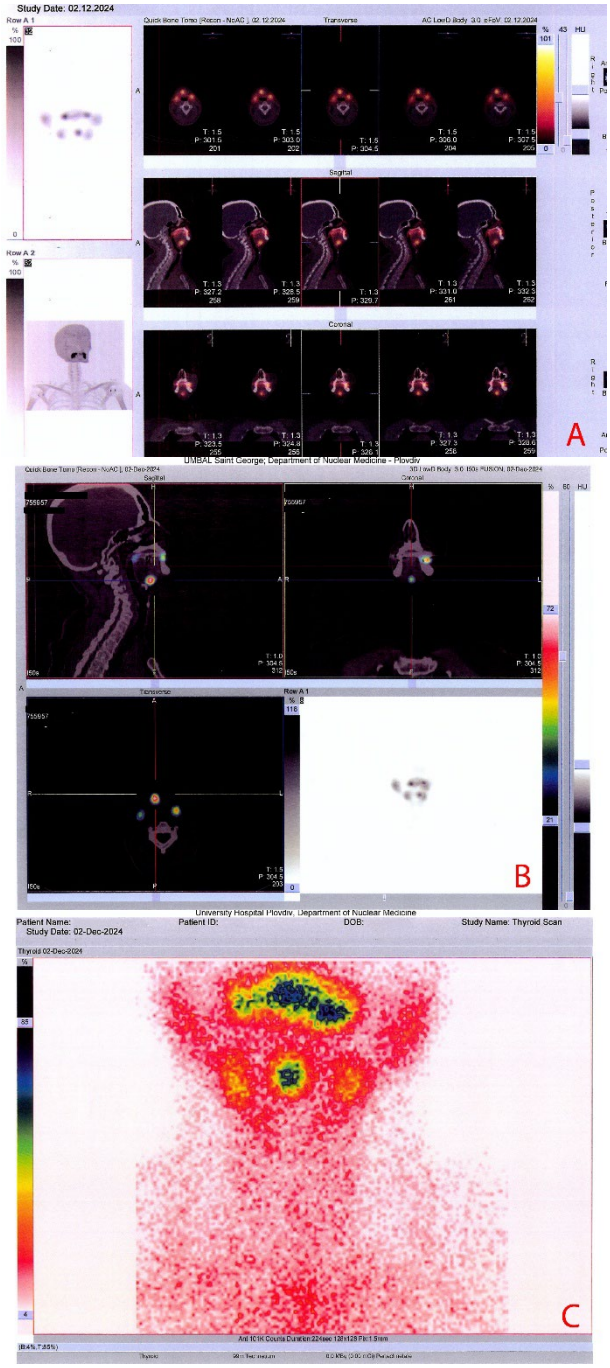

Figure 2: A and B. Hybrid SPECT/CT images demonstrate focal uptake of 99mTc-pertechnetate in the sublingual region, localized between the submandibular salivary glands. C. Planar scintigraphic image showing the same focal tracer accumulation. No radiotracer uptake is observed in the pretracheal thyroid bed or elsewhere in the neck, consistent with ectopic sublingual thyroid tissue and absence of orthotopic thyroid gland

High-resolution ultrasound is the first-line imaging modality for evaluating thyroid anatomy. However, it may not always detect ectopic tissue, particularly in anatomically challenging regions such as the sublingual space, or in cases where operator

experience is limited. In such scenarios, complementary imaging using radionuclide scintigraphy becomes essential. Functional imaging with Technetium-99m pertechnetate, particularly when combined with hybrid SPECT/CT, enables both anatomical localization and assessment of functional thyroid tissue. This dual-modality approach significantly enhances diagnostic accuracy by distinguishing ectopic thyroid from non-thyroidal masses and confirming the absence of orthotopic thyroid tissue [5], [7].

The delayed diagnosis in our case,spanning over two decades, highlights the consequences of relying solely on biochemical parameters without confirmatory imaging. A high index of clinical suspicion is warranted in patients presenting with hypothyroidism, absent cervical thyroid tissue on palpation, and persistently negative thyroid autoantibodies. In such cases, early implementation of comprehensive imaging can prevent misdiagnosis and guide appropriate management [6], [7], [8].

Management of ectopic thyroid tissue is primarily conservative. Lifelong levothyroxine therapy is usually sufficient to maintain euthyroidism, mirroring the treatment strategy for autoimmune thyroiditis. Surgical excision is reserved for patients presenting with obstructive symptoms, cosmetic concerns, or suspicion of malignancy. For asymptomatic patients with confirmed ectopic tissue and no complications, regular monitoring with clinical and imaging follow-up is recommended [4].

Conclusion

This case underscores the diagnostic challenges and clinical implications associated with ectopic thyroid tissue, particularly when it mimics autoimmune thyroiditis. In hypothyroid patients with absent palpable thyroid tissue and negative thyroid autoantibodies, the possibility of ectopic thyroid gland should be strongly considered. Failure to identify ectopic thyroid tissue may result in prolonged misdiagnosis, as seen in our patient who was treated for presumed autoimmune atrophic thyroiditis for over two decades without confirmatory imaging.

The use of high-resolution ultrasound remains a valuable initial tool, but it may be insufficient when thyroid tissue is located in atypical positions. In such cases, functional imaging, particularly hybrid SPECT/CT with Technetium-99m pertechnetate, offers superior diagnostic accuracy by enabling precise anatomical localization and assessment of tissue functionality. Clinicians should maintain a high index of suspicion in patients with atypical presentations, such as persistent negative thyroid antibodies and lack of visualized thyroid tissue in the cervical region. Early and comprehensive imaging evaluation can significantly alter the diagnostic trajectory and avoid unnecessary long-term assumptions of autoimmune thyroid disease. Ultimately, this case highlights the need for increased clinician awareness and a more systematic, imaging-guided diagnostic approach to hypothyroidism, especially in patients with unexplained findings. Accurate identification of ectopic thyroid tissue enables appropriate long-term management, which in most cases consists of thyroid hormone replacement and monitoring, avoiding unnecessary interventions.

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