

Ectopic Thyroid Gland

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Abstract

Ectopic thyroid is a rare condition resulting from aberrant embryological development, primarily involving abnormal migration of thyroid tissue. Genetic mechanisms affecting ectopic thyroid development have been identified. The lingual region is the most common site, and diagnostic methods include US, CT, and MRI, with thyroid scintigraphy considered the gold standard.

Keywords: head and neck, thyroid gland, congenital

Case Summary

A neonate presented with congenital hypothyroidism identified via neonatal screening. On laboratory evaluation, the thyroid-stimulating hormone levels were markedly elevated and T4 levels were low.

Imaging Findings

Technetium-99m pertechnetate thyroid scan was performed (Figure 1). There were 2 foci of uptake in the neck. The normal thyroid was not identified.

Diagnosis

Ectopic thyroid gland (ET).

The imaging differential diagnosis is primarily a thyroglossal duct cyst.

Discussion

ET refers to the presence of thyroid gland tissue in a location other than the typical pre-tracheal region of the neck. It is estimated to occur with a prevalence of approximately 1 in 100,000 to

1 in 300,000 people.¹ Ectopic thyroid is more common in patients with associated thyroid disease or dysfunction, occurring with an incidence of 1 in 4000-8000 in these patients.²⁻⁴ Females account for 65-80% of diagnosed patients.²

In normal embryologic development, the thyroid descends from the foramen cecum at the base of the tongue to its final position anterior and lateral to the 2nd through 4th tracheal rings. Certain transcription factors, including *TITF1/NKX2-1*, *PAX8*, *HHEX*, or *FOXE1*, play crucial roles in thyroid development and migration.^{2,4} Mutations in any of these genes can affect the gland's migration. Ectopic thyroid tissue can occur anywhere along the thyroid's pathway of descent, as well as in unexpected positions.¹ While the base of the tongue is the most common location for ectopic thyroid tissue to reside (accounts for 90% of cases), it can occur in other locations in the head and neck such as sublingual, submandibular, and lateral cervical regions.⁵ Remote locations have also been described, including the superior mediastinum, heart, esophagus, or diaphragm.

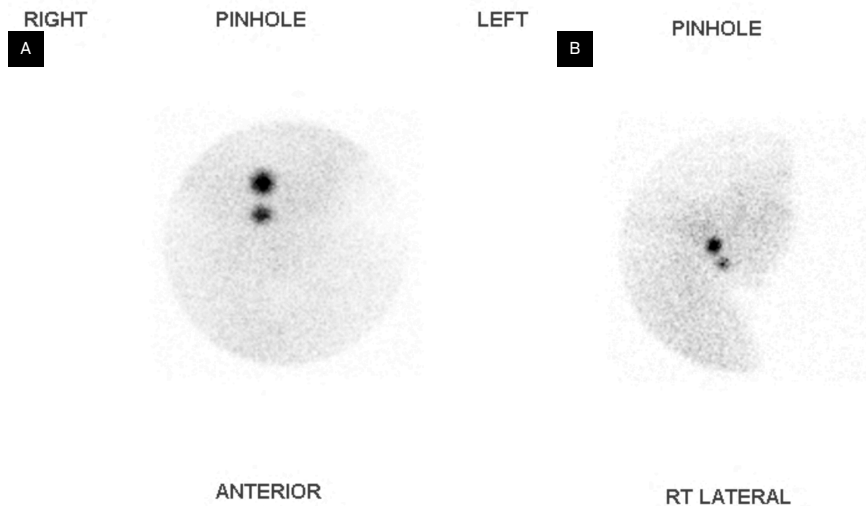
Patients with an ET are typically asymptomatic. The condition may be discovered incidentally through imaging.^{1,2,6} When present, symptoms are commonly attributed to mass effect on adjacent structures or endocrine dysfunction.^{1-3,6} For example, a lingual thyroid can manifest with symptoms such as cough, localized pain, dysphagia, dysphonia, dyspnea, and hemorrhage.² A notable physical exam finding in the case of a lingual thyroid is enlargement of the posterior base of the tongue due to a midline mass.²

Thyroid dysfunction is a common clinical feature, with hyperthyroidism being more prevalent than hypothyroidism.¹ Hyperthyroidism is uncommon in this setting and estimated to occur in <10% of cases. An ectopic thyroid is the most common cause of congenital hypothyroidism in infants. Abnormal migration during embryological development may result in insufficient blood supply to the thyroid tissue, leading to inadequate function.² While resulting hypothyroidism is subclinical in most cases, hormone production from the ectopic thyroid may be insufficient in situations requiring a higher physiological demand, such as during pregnancy, puberty, trauma, and infection.² Hyperthyroidism is rarely associated with ET. Malignant transformation of ectopic thyroid tissue is possible, with follicular and papillary neoplasms being the most common, though the frequency of such transformation is <1%.^{1,6}

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Figure 1. (A) Anterior and (B) lateral projection pinhole images from a technetium-99m pertechnetate thyroid scan showing 2 foci of uptake in the upper and lower neck at midline. There was no evidence of a normal thyroid gland.



US is the preferred initial assessment for a possible ectopic thyroid in the cervical region due to its cost-effectiveness and availability.^{1,2} CT or MRI can be used to differentiate ectopic thyroid tissue from adjacent structures.⁶ An ectopic thyroid appears as a hyperdense soft-tissue mass on noncontrast CT. The high density of thyroid tissue on noncontrast CT is due to the presence of iodine within the gland. Ectopic tissue enhances homogeneously after the administration of contrast. On MRI, ectopic thyroid appears as a well-defined and rounded mass with higher signal intensity in relation to surrounding tissue and musculature on both the T1- and T2-weighted images.^{1,7}

Iodine-123 or technetium-99 thyroid scintigraphy remains the most effective diagnostic tool, confirming the presence of uptake in an ectopic location.^{4,6} Technetium-99 typically yields better quality of imaging, lower radiation exposure, and longer half-life, but can accumulate in the background of the ectopic gland, making it less advantageous in smaller masses. Iodine-123 is also widely used,

but with a comparatively higher cost and shorter half-life. It is important to note that while some thyroid tissue may be ectopic, some may also be in the thyroid's expected location.

Asymptomatic ectopic thyroid usually does not require treatment but is monitored clinically.^{1,2,6} Treatment, when indicated, depends on factors such as location, size, functional status, symptom severity, and risk of malignancy. Suppressing therapy using exogenous thyroid hormone can be beneficial in managing obstructive symptoms related to the mass effect by suppressing Thyroid-Stimulating Hormone release and reducing gland size.^{1,2} Surgical excision is reserved for patients with severe symptoms related to local obstruction or if malignancy is suspected. The risk of an ectopic thyroid carcinoma is <1%. Assessment of the presence of a eutopic thyroid (thyroid tissue in a normal location) is crucial before surgery, as it may either contraindicate the operation or prompt lifelong hormone replacement therapy if the ectopic thyroid tissue being

removed is the patient's only functional thyroid tissue.^{1,2} Radioactive iodine-131 therapy is an alternative for patients with contraindications for surgery or when surgery is not feasible.^{1,2}

Conclusion

Ectopic thyroid is a rare condition resulting from aberrant embryological development, primarily involving abnormal migration of thyroid tissue. Genetic mechanisms affecting ectopic thyroid development have been identified. The lingual region is the most common site, and diagnostic methods include US, CT, and MRI, with thyroid scintigraphy considered the gold standard.

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