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Case Report

Thyroid isthmus agenesis: An uncommon yet clinically relevant entity

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ABSTRACT

The thyroid, a crucial endocrine gland composed of two lobes connected by isthmus tissue, rarely exhibits isthmus agenesis or hypoplasia, conditions scarcely documented in medical literature. This discussion centers on a 54-year-old female patient whose incidental discovery of isthmus agenesis occurred during surgery for a multinodular goiter. Identification of isthmus abnormalities prompts thorough consideration of potential associated anomalies, including hypoplasia, absence of a thyroid lobe and the presence of ectopic thyroid tissue. Increased awareness of potential isthmus agenesis or hypoplasia and associated thyroid anomalies among patients scheduled for thyroid surgery is essential for improving procedural safety and reducing the risk of complications related to surgery.

1. Introduction

Thyroid gland congenital anomalies, like hypoplasia, ectopic thyroid, hemiagenesis, or complete agenesis, are frequently documented in medical literature. However, agenesis of the thyroid isthmus is exceptionally rare, with only a handful of cases reported, mostly identified during cadaveric dissections.[1,2] Presenting a unique case of a thyroid follicular nodular disease, planned for total-thyroidectomy, where isthmus agenesis was unexpectedly found intra-operatively during the surgical procedure.

2. Case Details

A 54 year old female reported to the OPD of a tertiary care centre with the chief complaint of hoarseness of voice. The patient was evaluated for the same and advised a contrast enhanced computed tomography scan (CECT) which revealed multiple thyroid nodules (Figure 1). The patient underwent fine needle aspiration cytology (FNAC) from the thyroid nodule at some other centre, which was reported as features suggestive of Follicular neoplasm (Bethesda IV). The patient was taken up for total thyroidectomy after pre-anesthetic clearance. Laboratory parameters were suggestive of Hypothyroid status. Intra-operatively, it was found that the isthmus was absent/hypoplastic (Figure 2A). Total thyroidectomy was performed uneventfully and the resected specimen was submitted for histopathological examination. The right and left lobes were submitted separately (Figure 2B). The microscopic features were consistent with Adenomatoid nodules of thyroid Follicular Nodular Disease with focal Oncocytic change and areas of hyalinization and calcification and extensive lymphocytic thyroiditis (Figure 3). Three unremarkable parathyroid glands were also observed. The patient recovered well and is under follow up with medication for hypothyroidism.

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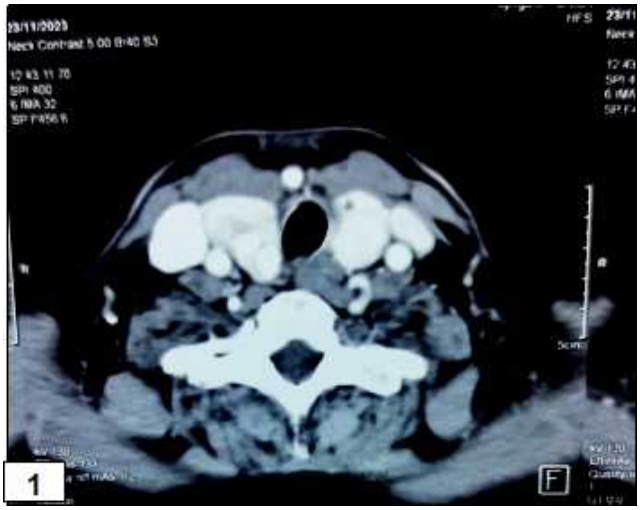

Figure 1: CECT showing multiple thyroid nodules in both lobes

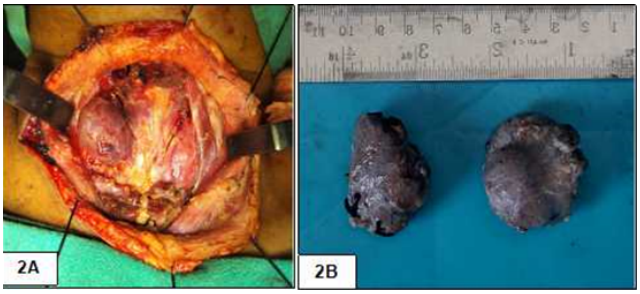

Figure 2: A: Intra-operative image showing missing thyroid isthmus; **B:** Gross picture showing separate lobes and missing isthmus

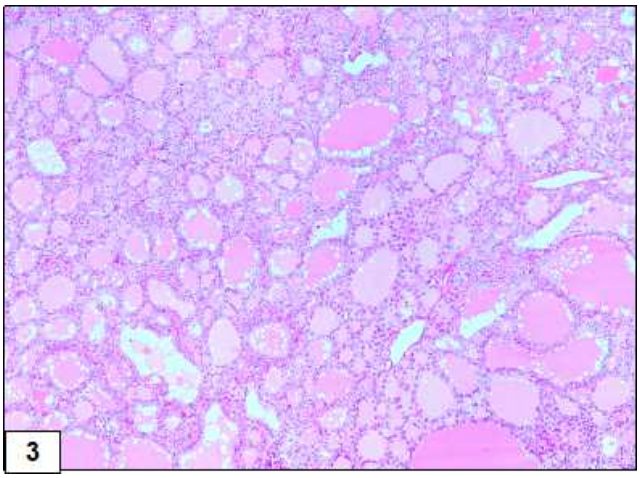

Figure 3: Photomicrograph showing features of Thyroid Follicular nodular disease (HE-100x)

3. Discussion

The thyroid gland is composed of two lateral lobes connected by a narrow median isthmus. The normal size of each lobe of the thyroid gland has been described to be 5 cm long, its greatest transverse and anteroposterior extent being 3 cm and 2 cm respectively. The isthmus measures about 1.25 cm transversely as well as vertically and is usually placed anterior to the second and third tracheal cartilages. The gland is divided into lobules of 20 - 40 round to oval follicles, each 50 - 500 microns, with a single layer of cuboidal to low columnar epithelium.[2,3] Congenital anomalies affecting the thyroid gland are extremely rare with incidence reported between 1-3%, however published literature supporting the same is less.[3,4]

The etiology of isthmus agenesis remains a subject of ongoing investigation, drawing insights from various sources including genetic predisposition, developmental irregularities, and mutations involving chromosome 22 and thyroid transcription factor (TITF) 1–2 genes. While studies and research contribute to our understanding, comprehensive knowledge is still evolving.[5,6] Hemiagenesis rarely causes hypothyroidism and is usually detected incidentally as most individuals are euthyroid. Though patients with thyroid hemiagenesis have increased risk of concomitant thyroid disease, e.g. nodular and autoimmune.[7,8] Furthermore, differential diagnoses should encompass autoimmune thyroid nodules, thyroiditis, primary thyroid carcinoma, metastasis and amyloidosis.[1]

4. Conclusion

In conclusion, the presented case underscores the importance of thorough evaluation and management of thyroid anomalies, such as isthmus agenesis and hemiagenesis. These congenital anomalies, although rare, can have significant clinical implications, including potential risks of hypothyroidism and associated thyroid diseases. While our understanding of the etiology of these anomalies is still evolving, ongoing research efforts continue to shed light on genetic predispositions and developmental irregularities. Comprehensive diagnostic approaches, including imaging studies and thyroid function tests, are essential for accurate diagnosis and guiding appropriate treatment strategies. Additionally, consideration of potential concomitant thyroid diseases and differential diagnoses is crucial for comprehensive patient care.

5. Source of Funding

None.

6. Conflict of Interest

None.

References

1. Ruchala M, Szczepanek E, Sujka-Kordowska P, Zabel M, Biczysko M, Sowinski J, et al. The immunohistochemical demonstration of parafollicular cells and evaluation of calcium-phosphate balance in patients with thyroid hemiagenesis. *Folia Histochem Cytobiol*. 2011;49(2):299–305.
2. Dixit D, Shilpa MB, Harsh MP, Ravishankar MV. Agenesis of isthmus of thyroid gland in adult human cadavers: a case series. *Cases J*. 2009;2:6640. doi:10.1186/1757-1626-0002-0000006640.
3. Kesici U, Kesici S. Agenesis of the isthmus of the thyroid gland. *Turk J Surg*. 2015;34(1):60–1.
4. Ozkan OF, Asık M, Toman H, Ozkul F, Cıkman O, Karaayvaz M, et al. Agenesis of isthmus of the thyroid gland in a patient with graves-basedow disease and a solitary nodule. *Case Rep Surg*. 2013;p. 608481. doi:10.1155/2013/608481.
5. Gupta M. Thyroid isthmus agenesis: A rare congenital anomaly. *J Otorhinolaryngol Allied Sci*. 2018;1(1):1–2.
6. Taty-Anna K, Farihah HS, Norzana AG, Farida H, Das S. Absence of the isthmus of the thyroid gland: anatomical and clinical considerations. *Clin Ter*. 2012;163(6):503–4.
7. Ruchala M, Szczepanek E, Szaflarski W, Moczko J, Czarnywojtek A, Pietz L, et al. Increased risk of thyroid pathology in patients with thyroid hemiagenesis: results of a large cohort case-control study. *Eur J Endocrinol*. 2009;162(1):153–60.
8. Sencha AN, Tukhbatullin MG. Thyroid Ultrasound. In: Sencha A, Patrunov Y, editors. Congenital Thyroid Anomalies and Thyroid Diseases in Children. Cham: Springer; 2019. doi:10.1007/978-3-030-14451-7_3.

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