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Case Report

Ear, Nose and Throat Section

Hemiagenesis of the Thyroid in a Seven-year-old Child: A Case Report

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ABSTRACT

Thyroid Hemiagenesis (TH) is a rare condition characterised by developmental abnormalities of the thyroid, often observed on the left-side, with a higher prevalence in females. It is commonly associated with a lack of the isthmus. A seven-year-old boy presented to the Ear Nose Throat (ENT) Department with chief complaints of bilateral ear block sensation and dull aching earache persisting for two months, for which syringing was performed. No family history of TH or any past surgeries related to the neck, thyroid, or parathyroid were reported. In recent laboratory tests, his Thyroid Stimulating Hormone (TSH) level was 7.23 µIU/mL, free tetraiodothyronine was 8.91 mcg/dL, free triiodothyronine was 75.06 ng/dL, and Antithyroid Peroxidase (TPO) was 2.0 IU/mL. The neck ultrasound revealed the absence of the left thyroid lobe and isthmus. He was diagnosed with congenital TH and hypothyroidism and was treated with a dosage of 25 mcg of levothyroxine. TH can be incidentally discovered and may manifest in later stages of life. Patients with TH are more prone to developing hypothyroidism compared to individuals with a complete thyroid, as they have lower reserves of thyroid hormones.

Keywords: Hypothyroidism, Scintigraphy, Thyroid development, Thyroid hemiagenesis

CASE REPORT

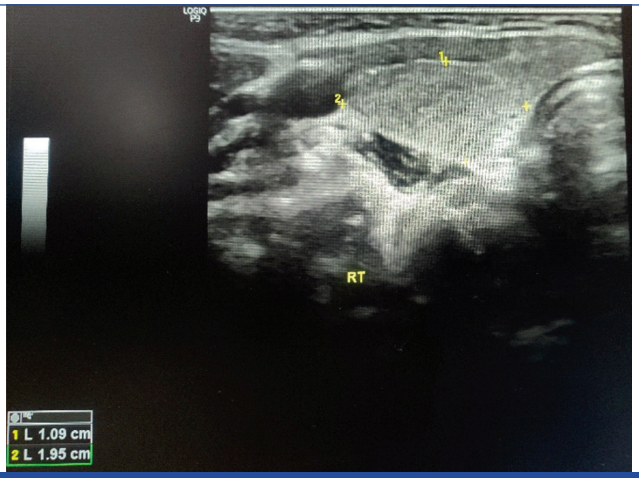
A seven-year-old boy presented to the ENT department with chief complaints of bilateral ear block sensation and dull aching earache for two months. He had a positive history of mouth breathing and snoring. There were no complaints of ear discharge, vertigo, tinnitus, swelling, nasal discharge, epistaxis, dysphagia, or change in voice. He had no other known health problems or relevant personal and family history.

The general physical examination was normal. On local examination, the patient had bilateral earwax, for which syringing was performed, and the bilateral tympanic membrane appeared Grade-1 dull. He had an externally pinched nose. The oral cavity showed a high-arched palate with overcrowding of teeth. On neck examination, there was asymmetry of the neck, with diffuse hypertrophy on the right-side, while the left-side appeared flat. Palpation of the thyroid gland revealed diffuse enlargement of the right thyroid lobe without any nodules, significant pulsations, or tenderness. There was no significant cervical lymphadenopathy. Laryngeal crepitus was present, and tele indirect laryngoscopy did not reveal any other anomalies of the thyroglossal tract.

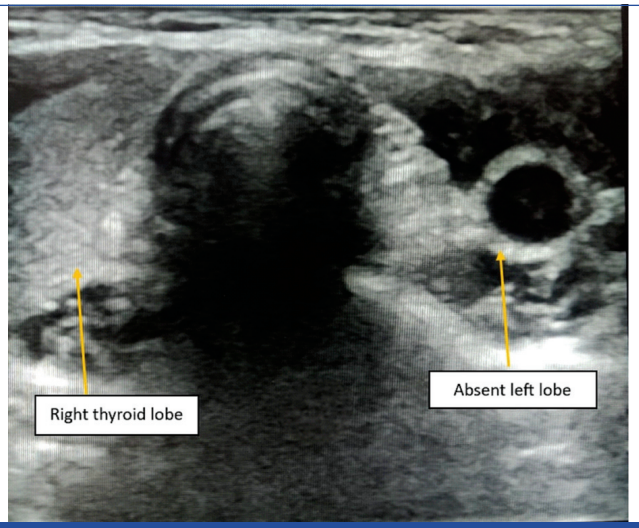
Based on the history, clinical examination, and further investigation, the patient was diagnosed with congenital TH with hypothyroidism, although no enlargement of the thyroid gland was found. The laboratory test results are displayed in [Table/Fig-1]. The right thyroid lobe was measured via ultrasound and had dimensions of 1.1×1.9×0.51 cm, as shown in [Table/Fig-2]. There were no visible aberrant lymph nodes, suspicious lesions, or signs of compression. The left thyroid lobe or isthmus showed no abnormalities, as seen in [Table/Fig-3]. The patient had never undergone thyroid surgery before, and neither the child nor his parents were aware of the condition.

Test type	Result	Biological reference range
Thyroid Stimulating Hormone (TSH)	7.23 µIU/mL	0.3-5.0 µIU/mL
Tetraiodothyronine (T4)	8.91 mcg/dL	5.6-13.3 mcg/dL
Triiodothyronine (T3)	75.06 ng/dL	82-241 ng/dL
Anti Thyroid Peroxidase Antibody (anti-TPO antibody)	2.0 IU/mL	Below 35 IU/mL

[Table/Fig-1]: Laboratory investigations of thyroid.



[Table/Fig-2]: Measurement of the right thyroid lobe.



[Table/Fig-3]: Absent left thyroid lobe.

The patient was prescribed mometasone nasal spray twice daily and started on a dosage of 25 mcg of levothyroxine. However, the patient was lost to further follow-up.

DISCUSSION

The TH is a rare birth defect caused by incomplete development of the thyroid gland during the embryonic stage. The commonly observed patterns of TH include the absence of the left lobe alone or the absence of the right lobe along with the absence of the isthmus [1]. Interestingly, and for reasons that are still unknown, the absence of the left lobe is the more common presentation, found in approximately 87.5% of the investigated subjects [1]. Other researchers have reported that the left thyroid lobe is missing in 80% of cases, with a left-to-right hemiagenesis ratio of 4:1 [1]. In present case, the male patient also exhibited left-sided hemiagenesis.

According to the literature, thyroid hemiagenesis is considered one of the rarest developmental abnormalities of the thyroid gland, affecting only 0.02% of the population [2]. However, there have been reports of a slightly higher prevalence in areas with an increased incidence of goiter and thyroid nodules [3]. It has been observed that children with congenital hypothyroidism may have an increased susceptibility, reaching up to 3.7% [4]. Research conducted on school-aged children in Northern Poland, ranging from 7 to 15 years old, found a prevalence of 0.05% [5]. Similarly, a survey conducted on schoolchildren aged 11 to 14 years in Sicily also reported a prevalence of 0.05% [5]. Another study by Shabana W et al., in Belgium found a slightly higher incidence of 0.2% in asymptomatic schoolchildren [6]. The actual cause of thyroid hemiagenesis remains unknown, but genetic abnormalities have been suggested to play a role in its aetiology, as reported in monozygotic twins [7].

Patients with TH may experience similar pathological alterations in the functioning lobe, similar to what is typically observed in a fully developed thyroid gland. The most commonly observed conditions include hyperthyroidism, hyperparathyroidism, toxic adenoma, diffuse toxic goiter (Graves' disease), toxic multinodular goiter, primary or secondary hypothyroidism, colloid nodule, chronic thyroiditis, and follicular or papillary carcinoma [8]. Patients diagnosed with TH often have a higher incidence of morphological, functional, and autoimmune thyroid disorders compared to those with a bilobate thyroid [9].

Despite typically being clinically euthyroid, patients with TH may have significantly elevated levels of free triiodothyronine (FT3) and TSH [5]. This suggests that the stable thyroid function in the unilobed thyroid has been altered. In some cases, free tetraiodothyronine (FT4) levels did not change, while in others, FT4 was also increased [10]. In this particular case, there were no TPO antibodies, and the ultrasonography revealed no suspicious tumours, aberrant lymph nodes, or signs of compression. Thyroid disorders are more prevalent in individuals with TH. The high frequency of associated illnesses is assumed to be caused by long-term TSH overstimulation. Following a TH diagnosis, thorough monitoring and appropriate levothyroxine dosage should be administered [10].

In clinical practice, there is a question regarding the appropriate follow-up for patients with TH. According to the existing literature, clear answers to this question are still needed. When it comes to managing patients with TH, there are two essential factors to consider. Firstly, the follow-up and management of TH itself, and secondly, the follow-up, diagnosis, and management of thyroid disorders in the remaining thyroid lobe.

Ultrasound is the preferred imaging method for detecting the absence of a lobe and any changes in the remaining lobes [11]. It is readily available, cost-effective, and does not expose patients to radiation. However, the accuracy of the results depends on the operator's skill. Following ultrasound, scintigraphy of the thyroid using technetium or iodine is useful in providing a functional anatomy of the gland. It can also detect increased diffuse uptake and hyperactivity in the glands [12]. Scintigraphy is also capable of identifying ectopic thyroid tissue and aiding in the diagnosis of thyroid conditions in the remaining lobe, such as hyperthyroidism or a suspicious nodule for cancer [13,14]. However, scintigraphy has limitations, including artifacts caused by factors like cancer, a solitary nodule, and infiltrative or inflammatory pathologies of the thyroid gland.

Ayaz UY et al., published a case report on an eight-year-old boy with left TH and minimal right lobe hyperplasia [3]. The presence of TH was identified during ultrasound, and before any unnecessary or invasive attempts, complementary methods like scintigraphy were used to confirm the diagnosis. Computed tomography scans can also provide additional support in establishing a diagnosis. Ultrasound is a fast and effective way to confirm the diagnosis, and in our patient, it revealed that the isthmus and left thyroid lobe were not appreciated. Since there is no history of any surgery in present patient, it indicates that their TH was congenital in nature. Further research is needed to determine if levothyroxine treatment can effectively lower thyrotropin levels in TH patients.

CONCLUSION(S)

The TH is an uncommon developmental defect of unclear origin that is typically identified incidentally through imaging techniques. If only one thyroid lobe is found in a child during an initial ultrasound or scintigraphy, it suggests a diagnosis of TH. To confirm the diagnosis, another complementary method such as scintigraphy or ultrasound should be performed before considering any invasive procedures. For individuals with TH who have normal thyroid function and no other thyroid diseases, there are no immediate clinical implications. However, patients should be informed about these disorders and receive appropriate follow-up if, necessary. The same standard of treatment should be provided to patients with TH and other thyroid conditions as those with anatomically normal thyroid glands. Making informed decisions based on preoperative diagnostics and knowledge would benefit both patients and healthcare professionals.

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