

## Case Report

# An unusual case report of thyroid hemiagenesis with multinodular colloid goitre

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## ABSTRACT

Thyroid hemiagenesis is one of the rarest congenital anomaly of thyroid gland and found a total of 256 cases reported with female predominance. The left lobe is more commonly seen with thyroid hemi agenesis than the right lobe, with a ratio of 3.6: 1. We hereby reported a case of 50 years old female with the agenesis of the right lobe and isthmus of thyroid gland. Thyroid hemiagenesis can be associated with hyper or hypothyroidism. In our case patient had normal thyroid function tests. On ultrasonography of neck, a large heterogeneous lesion measuring 8.0×4.0×8.0 cm noted in the anterior aspect of neck, predominantly on the right side and seen crossing midline to the left. In our case we detected thyroid hemiagenesis incidentally with the help of ultrasonography and confirmed it intra-operatively. Ultrasonography is the most cost-effective means of diagnosing thyroid hemiagenesis.

**Keywords:** Thyroid hemiagenesis, Thyroid gland, Thyroid nodule, Congenital anomaly

## INTRODUCTION

Thyroid hemiagenesis is one of the rarest congenital anomaly of thyroid gland and found a total of 256 cases reported.<sup>1</sup> There is female predominance, with a female-to-male ratio between 1.3: 1.00, 1.4: 1.00, and 1.67: 1.00 in various journals.<sup>1-3</sup> The left lobe is more commonly seen with thyroid hemi agenesis than the right lobe, with a ratio of 3.6: 1.<sup>1,4</sup> The isthmus involved in 44% cases.<sup>1</sup> Thyroid hemiagenesis can present with all kinds of thyroid pathologies like nodules, inflammation, hyperthyroidism or hypothyroidism in the remaining gland.<sup>2,5</sup> This leads to a clinical investigation.<sup>2</sup> Most of the patients with thyroid hemiagenesis are asymptomatic, hence found only by coincidence.

## CASE REPORT

A 50 years old female patient came to our ENT OPD in Sapthagiri Institute of Medical Science and Research

Science with swelling in front of the neck since 20 years. It was insidious onset gradually progressive to the present size. It was not associated with pain, hoarseness, difficulty in swallowing and breathing. Past history of hysterectomy in 2008 and nephrectomy in 2016. No history of any chronic disorders and any medications. On examination solitary swelling about 8×10 cm noted in front of the neck in midline, with smooth surface margin diffuse, firm consistency. It moved with deglutition and skin over the swelling was normal. Lymph nodes are not palpable (Figure 1).

The biochemical thyroid profile was as follows: FT3 4.00 (range 2.8-5.3 pg/ml); FT4 0.96 (range 0.45-1.6 ng/ml); TSH 1.740 (range 0.465-4.68 mIU/l). On X-ray neck AP and lateral, soft tissue swelling in the anterior aspect of neck compressing over the trachea and narrowing the lumen-thyroid swelling. On ultrasonography of neck, a large heterogeneous lesion measuring 8.0×4.0×8.0 cm noted in the anterior aspect of neck, predominantly on the

right side and seen crossing midline to the left. Lesion shows few hypoechoic areas within and vascularity noted within the lesion. It is likely arising from thyroid. Isthmus and right lobe of thyroid could not be visualised separately. Few enlarged lymph nodes are noted in left level II and III, largest measuring 12.5×6.5 mm. Tc-99m scintigraphy, CT and MRI was not done. Fine needle aspiration was performed. Cytological features are of benign colloid goitre.

A left thyroidectomy was performed and the agenesis of the right lobe and isthmus was confirmed intraoperatively (Figure 2 and 3). Histopathological features suggestive of multinodular colloid goitre with adenomatoid changes with secondary hyperplasia. No complication was observed and the patient was discharged on the seventh postoperative day after suture removal.



**Figure 1: Midline solitary swelling.**



**Figure 2: Left lobe of thyroid gland.**



**Figure 3: Thyroidectomy specimen for histopathology.**

## DISCUSSION

The thyroid gland development begins with an invagination of the endoderm in the primitive pharynx. It grows ventrally and remains attached to the pharyngeal floor. The rudiment thyroid migrates to its anatomical position, which lies anterior to the pharynx. Then grows laterally to form bilobed thyroid gland.<sup>6</sup> The most common congenital abnormalities of the thyroid include a lingual thyroid and a thyroglossal cyst.<sup>7</sup>

The congenital thyroid anomalies caused either by abnormal descent of the gland or by incomplete genesis of a lobe. The etiology of the hemiagenesis still unknown.<sup>7-10</sup> A high incidence of the condition in monozygotic twins and also association with other thyroid malformations within the family indicate a possible role of genetic factors. PAX-8, TTF-1 and TTF-2 genes have been implicated in thyroid morphogenesis and descent.<sup>8-16</sup> PAX-8 participates in regulation of embryogenesis of the thyroid gland and müllerian and upper urinary tracts.<sup>11</sup> The reported cases are commonly associated with Grave's disease, nodular goiter and thyroid neoplasm, hyperparathyroidism with least incidence.<sup>3-12</sup> The absence of the left lobe in 80% of cases and agenesis of the isthmus observed in 44-50% of cases.<sup>18,19</sup> The patient in our case report had right lobe agenesis with isthmus agenesis. The prevalence in women and girls is greater than in men and boys (75% versus 25%).<sup>18,19</sup> There is no explanation for female predominance but it may indicate a possible sex-related background.

Thyroid hemiagenesis is commonly diagnosed by thyroid scan via accumulation of tracer on one side within the thyroid region even after thyrotropin application. But several clinical conditions mimicking thyroid hemiagenesis in scintigraphic evaluation like autonomously functioning nodules with suppressed normal thyroid tissue, primary or secondary neoplasms, infiltrative diseases such as amyloidosis, and unilateral inflammations of one lobe can mimic thyroid hemiagenesis.<sup>18-22</sup> In all these above clinical conditions functional hemiagenesis based on thyroid scan alone, can be misdiagnosed as congenital hemiagenesis. Therefore, thyroid scans should be confirmed by other methods revealing the morphology of the thyroid gland. Ultrasonography is cost effective and easy to perform on patient with no radiation exposure. Other techniques are computed tomography (CT) and magnetic resonance imaging (MRI). Both are expensive and time consuming. The diagnostic performance of these investigations is comparable with ultrasonography of the thyroid gland. Thyroid hemiagenesis is often diagnosed during the investigation of thyroid disorder. The thyroid scintigraphy for functional hemiagenesis should be combined with revealing morphologic hemiagenesis like USG, CT or MRI. The most cost effective way of diagnosis is USG. The fine needle aspiration biopsy should be performed in order to rule out primary and secondary malignancies in thyroid nodules.

## CONCLUSION

The patients with thyroid hemiagenesis, with a normal thyroid function and no other thyroid pathologies, there are no relevant clinical consequences in the short term. However, based on the current literature, a susceptibility to other thyroid pathologies especially hypothyroidism, increased rates of thyroid autoimmune diseases, and thyroid enlargement (diffuse or nodular) may be possible consequences in the long term. Thus, regular follow-up visits concerning thyroid function is advisable.

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## REFERENCES

- Mikosch P, Gallowitsch HJ, Kresnik E, Molnar M, Gomez I, Lind P. Thyroid hemiagenesis in an endemic goiter area diagnosed by ultrasonography: report of sixteen patients. *Thyroid*. 1999;9(11):1075-84.
- Mikosch P, Gallowitsch HJ, Kresnik E, Molnar M, Gomez I, Lind P. Thyroid hemiagenesis in an endemic goiter area diagnosed by ultrasonography: report of sixteen patients. *Thyroid*. 1999;9(11):1075-84.
- Suzuki S, Midorikawa S, Matsuzuka T, Fukushima T, Ito Y, Shimura H, et al. Prevalence and Characterization of Thyroid Hemiagenesis in Japan: The Fukushima Health Management Survey. *Thyroid*. 2017;27(8):1011-6.
- Hamburger JI, Hamburger SW. Thyroidal hemiagenesis. Report of a case and comments on clinical ramifications. *Arch Surg*. 1970;100(3):319-20.
- Schneider C, Dietlein M, Faust M, Drzezga A, Schmidt M. Thyroid hemiagenesis is combined with a variety of thyroid disorders. *Nuklearmedizin*. 2019;58(3):265-71.
- Maiorana R, Carta A, Floriddia G, Leonardi D, Buscema M, Sava L, et al. Thyroid hemiagenesis: prevalence in normal children and effect on thyroid function. *J Clin Endocrinol Metab*. 2003;88(4):1534-6.
- Chang J, Gerscovich EO, Dublin AB, McGahan JP. Thyroid hemiagenesis: a rare finding. *J Ultrasound Med*. 2011;30(9):1309-10.
- Peña S, Robertson H, Walvekar RR. Thyroid hemiagenesis: report of a case and review of literature. *Indian J Otolaryngol Head Neck Surg*. 2011;63(2):198-200.
- Parulska E, Zybek-Kocik A, Woliński K, Czarnocka B, Ruchała M. Does TSH Trigger the Anti-thyroid Autoimmune Processes? Observation on a Large Cohort of Naive Patients with Thyroid Hemiagenesis. *Arch Immunol Ther Exp (Warsz)*. 2016;64(4):331-8.
- Nsame D, Chadli A, Hallab L, El Aziz S, El Ghomari H, Farouqi A. Thyroid Hemiagenesis Associated with Hashimoto's Thyroiditis. *Case Rep Endocrinol*. 2013;2013:414506.
- Bhartiya S, Verma A, Basu S, Shukla V. Congenital thyroid hemiagenesis with multinodular goiter. *Acta Radiol Short Rep*. 2014;3(9):2047981614530286.
- Kirdak T, Gulcu B, Korun N. Thyroid hemiagenesis associated with retrosternal nodular goiter: a case report. *Acta Med Iran*. 2014;52(9):725-7.
- Ayaz ÜY, Ayaz S, Döğen ME, Api A. Ultrasonographic and scintigraphic findings of thyroid hemiagenesis in a child: report of a rare male case. *Case Rep Radiol*. 2015;2015:917504.
- Maiorana R, Carta A, Floriddia G, Leonardi D, Buscema M, Sava L, et al. Thyroid hemiagenesis: prevalence in normal children and effect on thyroid function. *J Clin Endocrinol Metab*. 2003;88(4):1534-6.
- Chang J, Gerscovich EO, Dublin AB, McGahan JP. Thyroid hemiagenesis: a rare finding. *J Ultrasound Med*. 2011;30(9):1309-10.
- Castanet M, Leenhardt L, Léger J, Simon-Carré A, Lyonnet S, Pelet A, et al. Thyroid hemiagenesis is a rare variant of thyroid dysgenesis with a familial component but without Pax8 mutations in a cohort of 22 cases. *Pediatr Res*. 2005;57(6):908-13.
- Szczepanek-Parulska E, Szaflarski W, Piątek K, Budny B, Jaszczyńska-Nowinka K, Biczysko M, et al. Alternative 3' acceptor site in the exon 2 of human PAX8 gene resulting in the expression of unknown mRNA variant found in thyroid hemiagenesis and some types of cancers. *Acta Biochim Pol*. 2013;60(4):573-8.
- Eroglu M, Ozkul F, Barutcu EC, Arik K, Adam G, Bilen Y, et al. Severe hyperparathyroidism in patient with right thyroid hemiagenesis. *J Pak Med Assoc*. 2015;65(9):1022-3.
- Mikosch P, Gallowitsch HJ, Kresnik E, Molnar M, Gomez I, Lind P. Thyroid hemiagenesis in an endemic goiter area diagnosed by ultrasonography: report of sixteen patients. *Thyroid*. 1999;9(11):1075-84.
- Prakash R, Narayanan RV, Chakravarty SK. Differentiation between suppressed thyroid tissue and thyroid hemiagenesis with Tc-99m pertechnetate radionuclide angiography. *Clin Nucl Med*. 1990;15(9):605-9.
- Marwaha RK, Khanna CM, Gupta RK, Sharma R. Hemiagenesis associated with chronic lymphocytic thyroiditis. *J Assoc Physicians India*. 1990;38(7):507-8.
- Matsumura LK, Russo EM, Dib SA, Maciel RM, Chacra AR. Hemiagenesis of the thyroid gland and T3 hyperthyroidism. *Postgrad Med J*. 1982;58(678):244-6.

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