

Thyroid hemiagenesis with compensatory hypertrophy of the remaining lobe: A case report

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Abstract

Thyroid hemiagenesis is defined as a failure of one thyroid lobe development. This condition predominantly manifests as an incidental finding during radiological investigation. This paper reports the case of a 53-year-old female, a known case of hypertension, who visited the ENT clinic at AKU, a tertiary care centre in Karachi, Pakistan and was hospitalized from 12th to 15th September 2021. The patient presented with hemiagenesis of the right thyroid lobe with enlargement of the contralateral lobe resulting in airway compression. She was subjected to excision of the thyroid gland without any intra-operative or postoperative complications. There were no complaints of dyspnoea, stridor or hoarseness during the hospital stay. The patient was discharged and was found to be well on subsequent follow-ups.

Keywords: Unilateral Thyroid Agenesis, Thyroidectomy, Case Report.

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Introduction

Unilateral thyroid agenesis is a failure of thyroid lobe development and is a rare anomaly. An estimated prevalence rate of this rare anomaly is 0.05%, however, the true incidence is unknown.¹ Mostly affecting females,¹ thyroid agenesis predominantly manifests as an incidental finding during radiological examination. This paper reports the case of a patient with hemiagenesis of the right thyroid lobe with enlargement of the contralateral lobe resulting in airway compression. The patient was subjected to a thyroidectomy to relieve these compressive symptoms. Postoperatively, the patient was managed on analgesics only. Recognition of this rare entity pre-operatively is very important for reaching a correct diagnosis and further management, including planning surgery. This article also includes a relevant review of literature and discusses the management of this condition diagnosed incidentally.

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Case Report

A 53-year-old female, known case of hypertension, presented to the ENT clinic at AKU, a tertiary care centre in Karachi, Pakistan, with a complaint of left sided neck swelling for the past 2 months. The patient had no history of neck surgery. In addition to this, she reported having difficulty in breathing on lying down for the past 1 month. On general physical examination, the patient was found to be comfortable and at rest, with no obvious signs of distress. She was well-oriented, alert and had no breathing difficulties at rest. Her voice was unremarkable. A neck examination revealed, a left sided neck swelling, approximately 4x4 cm in size. The swelling was non-tender, multinodular and firm in consistency. The overlying skin was mobile with no apparent changes. On deglutition, vertical movement of the swelling was evident. In addition, there were no palpable lymph nodes but laryngeal crepitus was present. Fiber optic laryngoscopy was performed to document the pre-operative vocal cord status which revealed bilaterally mobile vocal cords.

Keeping in view the history of dyspnoea on lying down, an MRI head and neck was performed. The reports showed a large heterogeneously enhancing soft tissue mass lesion in the left lower neck extending to the left superior mediastinum and measuring 3.7x4.0x9.1 cm in APxCC x TS dimensions. The mass also appeared to be extending posterior to the trachea and causing significant mediastinal shift towards the right side (Figure 1 & 2). In addition,

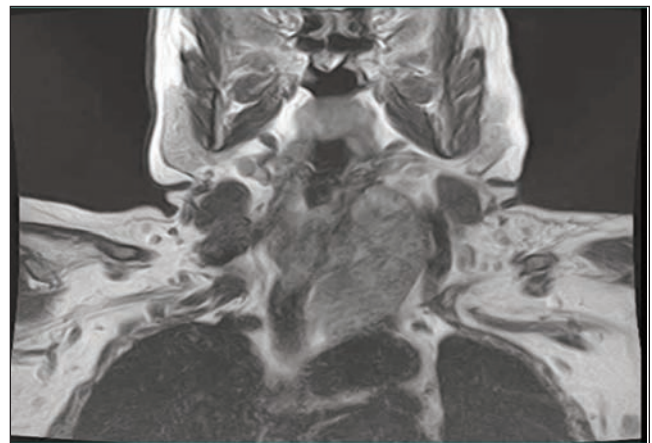


Figure-1: Coronal view of MRI showing unilateral thyroid agenesis and deviated trachea with hyperplasia of the remaining lobe.

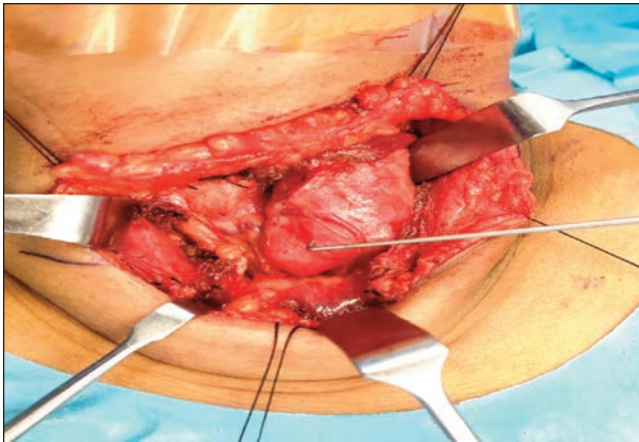


Figure-2: Intraoperative picture showing hemi thyroid agenesis on the right side.

multiple enlarged supraclavicular and occipital lymph nodes were also noted. At this stage, a biopsy was advised for confirmation. A Fine Needle Aspiration Cytology (FNAC) was done which showed benign follicular nodules, with a Bethesda II classification.

The patient was hospitalized from 12th to 15th September 2021 and successfully underwent total thyroidectomy. An enlarged thyroid gland presenting on the left side only was recovered. The right lobe and the isthmus were absent. The size of the left lobe was 5x6 cm extending retrosternally. The recurrent laryngeal nerve was identified and secured on the left side. In addition, the superior and inferior parathyroid glands were identified and secured along with their vasculature. The surgery was uneventful, and no other aberrant features were identified. The wound was closed in layers and the patient was discharged on the 2nd post-operative day. No drain was placed. Post-operative medications included calcium supplements and analgesics initially followed by thyroxin supplements which were dose adjusted according to the patient's weight. The patient's calcium levels were also monitored post-operatively to screen for hypocalcaemia along with TSH levels which were all within normal ranges. The TSH level was also repeated after 1 year on follow-up at which point the thyroxin dose was adjusted accordingly.

On gross examination, the thyroid tissue removed weighed 75 gm and measured 9x3x3 cm. Serial slicing of the organ revealed a multinodular architecture with patchy grey white and haemorrhagic areas. Histopathological evaluation showed multiple nodules of variable sizes comprising of thyroid follicles. These follicles were lined by columnar cells with regular nuclei and filled with colloid. Additionally, the presence of cholesterol clefts, haemorrhage, patchy fibrosis, and hyalinization was observed. However, there was no evidence of nuclear change, increased mitosis, necrosis, or invasion. The final

histopathology report was signed out as benign nodular hyperplasia.

Discussion

Several developmental anomalies of the thyroid gland are reported in the literature, which include partial or total agenesis of the gland, ectopic or accessory thyroid tissues and absence of the thyroid isthmus.² These disorders distort the morphology of the gland and cause clinical and functional disorders.²

The thyroid is the first endocrine gland to develop in the human embryo approximately 24 days after fertilization under the influence of the Fibroblast Growth Factor (FGF) signalling pathways.³ Thyroid progenitors rely on a thyroid-specific signature of transcription factors for their survival and proliferation. Congenital disorders associated with thyroid organogenesis may be caused either by abnormal descent of the gland or by incomplete genesis of a lobe. However, the aetiology of an absent thyroid lobe still remains unclear which may be due to a genetic component. This rare condition has been documented in existing literature in monozygotic twins.⁴ According to a study conducted by Maiorana et al in 2003, the prevalence of thyroid haemiagenesis studied via neck ultrasounds in 24,032 unselected 11 to 14 years old school-going children, reported an estimated prevalence of 0.05%.⁵ Additionally, it was observed that compensatory hypertrophy of the residual thyroid lobe, due to overstimulation of TSH, occurs in most cases but not all. They concluded that the high risk of goitre and hypothyroidism in such patients suggests requiring a systemic follow-up of all identified cases of thyroid hemiagenesis.⁵ In addition, according to existing literature, hemiagenesis of the thyroid involves the left lobe in 80% of cases with the absence of isthmus in about half of these cases.⁵ However, this reported patient had hemiagenesis of the right lobe with compensatory hypertrophy of the left lobe enough to compress the trachea.

Congenital thyroid lobe agenesis is often diagnosed incidentally due to some pathology in the existing lobe. These include hyperthyroidism, hypothyroidism, multinodular goitre, chronic thyroiditis, adenocarcinoma, papillary thyroid carcinoma and lingual thyroid.² Hyperthyroidism is the most common condition associated with thyroid hemiagenesis.² In the presented case, this anomaly was found as an incidental finding on computerized tomography scan and MRI of the head and neck region. Thyroid morphogenesis can be found by multiple different diagnostic methods including ultrasonography, scintigraphy, CT scans and MRIs. Ultrasonography can display an absent lobe of thyroid

gland.⁶ Scintigraphy can be additionally used but is not always required to analyze absent thyroid lobe; however, it is useful in identifying the presence of an ectopic thyroid tissue.⁷ Cardiac pathologies are also often reported among patients with congenital thyroid dysgenesis.⁸ The strong spatial connection between the heart and the thyroid primordial components, suggests an early morphogenesis, contributing to patterning characteristics with transcriptional activities of Nkx2.5 and GATA4. In response to which cardiac progenitor cells likely to mediate FGF8's effect on heart development. Nkx2.5 is expressed in the pharyngeal endoderm. This implies that FGF8 regulates Nkx2.5 in the thyroid progenitors as well.³ In humans, NKX2-1 haploinsufficiency results in a syndrome which is characterised by a triad of thyroid hypoplasia, choreoathetosis and pulmonary disease. According to a study, carried out on human and mice; the neurological manifestations cannot be rescued by thyroxin replacement. Illustrating the importance of NK2-1 transcriptional activity in the forebrain development.⁹

Conclusion

There are several points that merit the discussion regarding post-operative care, hormone replacement and follow-up in such patients including replacement of calcium and vitamin D. There are still no well settled guidelines for the management of such patients, due to which more studies are needed. Clinicians need to keep this differential in mind when seeing similar patients. As, low prevalence of it may often be overlooked or placed lower down on the differential list. In addition, the possibility of a threatened airway due to compensatory hypertrophy of the contralateral lobe is also an important point to keep in mind while dealing with such cases.

Compliance with Ethical Standards: This paper complies with ethical standards.

Consent: Consent for publishing the case was obtained from the patient before writing the manuscript.

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References

1. Mikosch P, Gallowitsch HJ, Kresnik E, Molnar M, Gomez I, Lind P. Thyroid hemigenesis in an endemic goiter area diagnosed by ultrasonography: report of sixteen patients. *Thyroid* 1999;9:1075-84. doi: 10.1089/thy.1999.9.1075.
2. Veerahanumaiah S, Dakshayani KR, Menasinkai SB. Morphological variations of the thyroid gland. *Int J Res Med Sci* 2015;3:53-7. DOI: 10.5455/2320-6012.ijrms20150110.
3. Shanmugalingam S, Houart C, Picker A, Reifers F, Macdonald R, Barth A, et al. *Ace/Fgf8* is required for forebrain commissure formation and patterning of the telencephalon. *Development* 2000;127:2549-61. doi: 10.1242/dev.127.12.2549.
4. McLean R, Howard N, Murray IP. Thyroid dysgenesis in monozygotic twins: variants identified by scintigraphy. *Eur J Nucl Med* 1985;10:346-8. doi: 10.1007/BF00251309.
5. Maiorana R, Carta A, Floriddia G, Leonardi D, Buscema M, Sava L, et al. Thyroid hemigenesis: prevalence in normal children and effect on thyroid function. *J Clin Endocrinol Metab* 2003;88:1534-6. doi: 10.1210/jc.2002-021574.
6. Shabana W, Delange F, Freson M, Osteaux M, De Schepper J. Prevalence of thyroid hemigenesis: ultrasound screening in normal children. *Eur J Pediatr* 2000;159:456-8. doi: 10.1007/s004310051307.
7. Schoen EJ, Clapp W, To TT, Fireman BH. The key role of newborn thyroid scintigraphy with isotopic iodide (¹²³I) in defining and managing congenital hypothyroidism. *Pediatrics* 2004;114:e683-8. doi: 10.1542/peds.2004-0803.
8. Konno N, Kanaya A. Thyroid hemigenesis associated with the right aortic arch. *J Endocrinol Invest* 1988;11:685-7. doi: 10.1007/BF03350216.
9. Pohlenz J, Dumitrescu A, Zundel D, Martiné U, Schönberger W, Koo E, et al. Partial deficiency of thyroid transcription factor 1 produces predominantly neurological defects in humans and mice. *J Clin Invest* 2002;109:469-73. doi: 10.1172/JCI14192.

Author Contribution:

AAU: Conception, design, literature review, patient data review.

SA: Final approval, critical revisions.

SKG: Patient data review and organization, critical revisions.

MOA: Drafting, critical revisions.

SA: Design, analysis and interpretation, literature review.