

## Case Report

# A Case Report on Thyroid Isthmus Agenesis

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### ABSTRACT

Agnesis of the thyroid isthmus is a rare entity with a incidence rate of 0.5 and 10%. Thyroid isthmus agnesis could be associated with ectopic thyroid tissue or agnesis of the thyroid lobes. We discuss a case of a multinodular goitre which presented with morphological variations which was not detected on ultrasonography of the neck.

#### Keywords

Thyroid; Head and neck; Isthmus.

### INTRODUCTION

There are a large number of morphological variations of the thyroid gland. Dysgenesis of the thyroid isthmus is however, poorly reported with an incidence rate ranging between 0.5 and 10% in literature.<sup>1,2</sup>

The thyroid glands original position is marked by the foramen caecum at the junction between the anterior two-thirds and posterior one-third of the tongue. At the 4<sup>th</sup> week of intrauterine life, invagination of the endodermal cells of the ventral floor of the primitive pharynx, gives rise to the thyroglossal duct. The duct descends to loop round the hyoid before its descent to the neck to give rise to two lateral buds which develop into the lateral lobes of the thyroid at the level of the second and third tracheal rings.<sup>3</sup>

Variations in thyroid morphology are poorly understood and often associated with ectopic thyroids tissue and morphological anomalies in the gland. Mutation of thyroid transcription factor-2 has been identified as a factor leading to poor development of the gland.<sup>4</sup>

Here, we discuss the case of a 25-year-old female who presented to the Ophthalmic Outpatient Department (OPD) in a euthyroid state with a multinodular goitre, the isthmus agnesis was an incidental finding during total thyroidectomy.

### CASE REPORT

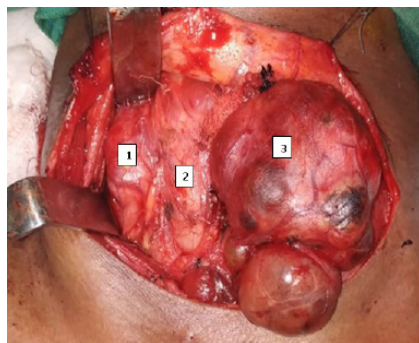
A 25-year-old Indian female patient presented with a single progressively enlarging swelling in the midline of the neck. The patient reported to the OPD due to concern over the size of the neck swelling. There was no associated difficulty in breathing or swallowing due to pressure exerted on the neck by the swelling. Patient gave no history suggestive of change in weight, intolerance to heat or cold, changes in the regular menstrual cycle or fatigue. The general physical examination revealed all parameters were within normal limits. Physical examination of the swelling revealed a 5×4 cm firm swelling with palpable nodules over the left side of the neck within the anterior triangle which moved on deglutition. There was no elevation of the swelling on protrusion of the tongue. No palpable cervical lymph nodes were identified on examination.

The patient was subjected to ultrasonography of neck which revealed an enlarged thyroid gland with multiple oval shaped, well marginated hypoechoic nodules in both lobes of thyroid. The largest nodule measured 4×3 cm on the left lobe. The isthmus could not be visualized separately from the lobes of the thyroid gland. On doppler interrogation, the left lobe showed increased peripheral vascularity. There also evidence of multiple anechoic cystic lesions with no wall calcification or internal septations seen noted in both lobes of the thyroid. There was no mention of a remnant thyroglossal duct.

A fine-needle aspiration cytology following the ultrasonography of the neck was suggestive of a nodular goitre.

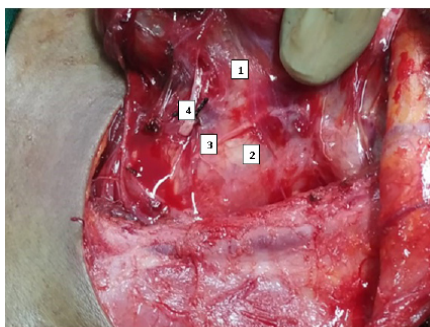
The patient was counselled on the available treatment options, and decided to proceed with an elective total thyroidectomy. Intra-operatively the thyroid gland showed multiple nodules and cysts which appeared to obscure the isthmus. Following the ligation of the superior pole of the left lobe there appeared to be an absence of thyroid tissue connecting both the lobes of the thyroid gland. A thyroid isthmus agenesis was noted (Figure 1). There were no variations in the morphology of the two lobes or in the position of the recurrent laryngeal nerves (Figure 2). Post-operative images include the pre-tracheal fascia removed over the tracheal rings between the lobes (Figure 3). The post-operative period was uneventful.

**Figure 1.** An Intra Operative Image of the Thyroid Gland with Absence of the Isthmus



1. Right lobe of the thyroid; 2. Trachea with pretracheal fascia and isthmus dysgenesis; 3. Left Lobe of the thyroid

**Figure 2.** The Position of the Recurrent Laryngeal Nerve in the Tracheoesophageal Groove

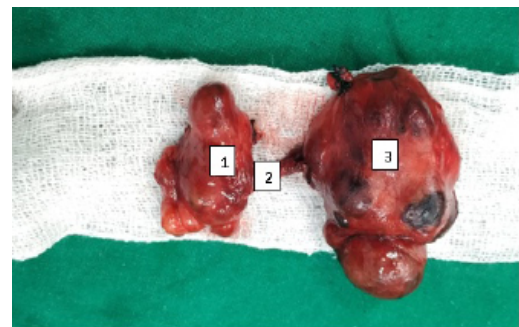


1. Left lobe of the thyroid; 2. Oesophagus below the recurrent laryngeal nerve; 3. Left Recurrent laryngeal nerve; 4. The ligated stump of the Inferior thyroid artery

The block specimen was sent for histopathological evaluation for both the thyroid gland and the connecting pretracheal fascia between the two lobes. This was to ensure there was no remnant thyroid tissue connecting the lobes of the thyroid.

The histopathological report was suggestive of a multinodular goitre. The specimen showed a variability in the size of the nodules present in the thyroid gland with large content of colloid. There was no remnant thyroid tissue present in the fascia.

**Figure 3.** Specimen Following Total Thyroidectomy



1. Right lobe of the thyroid; 2. Pre-tracheal fascia; 3. Left lobe of the thyroid

## DISCUSSION

Normal development of the thyroid gland begins with a median thickening of endoderm on the floor of the pharynx between the first and second pharyngeal pouches. During the 4<sup>th</sup> week of gestation, invagination of this region leads to the formation of the median diverticulum. The thyroid diverticulum which forms into the thyroglossal duct, bifurcates to give rise to the thyroid lobes and the isthmus. The cephalic end of the thyroglossal duct degenerates during this process.<sup>3</sup>

Developmental abnormalities of the thyroid can be divided into three major categories: (1) agenesis of thyroid gland; (2) dysgenesis of the thyroid; (3) abnormalities due to persistence of the thyroglossal duct. Dysgenesis of the thyroid gland most commonly presents with hemigenesis or ectopic thyroid gland tissue.<sup>4</sup> In particular the agenesis of the thyroid isthmus has an incidence rate ranging between 0.5 and 10%.<sup>1,2</sup>

The majority of literature on thyroid dysgenesis is based on cadaveric studies. On dissecting 105 cadavers, Ranade et al<sup>5</sup> reported 34 cadaveric specimens with the agenesis of a thyroid isthmus. Dixit et al<sup>6</sup> studied 41 cadavers with only 6 thyroids showing an absent isthmus with no ectopic or gross change in morphology of the lobes.

Although thyroid hypoplasia has been associated with mutations in the thyrotropin (TSH) receptor, the cause of thyroid agenesis is unknown.<sup>7</sup> Mutations of chromosome 22 or variations in the thyroid *transcription factor 1-2* genes have been reported to play a role in the anatomical variations of the gland.<sup>8-10</sup>

Embryological developmental anomalies due to a high division of the thyroglossal duct can also generate two independent thyroid lobes with failure of fusion in the midline.<sup>11</sup>

The recurrent laryngeal nerve was identified without an anomalous course in the tracheoesophageal groove below inferior thyroid artery on both sides in this case. The authors could not find articles identifying recurrent laryngeal nerve anomalies associated with thyroid dysgenesis at the time of writing.

Associated variations in the morphology of the thyroid lobes, presence of ectopic thyroid tissue and parathyroid hyperplasia have been reported with isthmus agenesis.<sup>2,6,10</sup> The poor demarcation of the isthmus on radiological evaluation in this case should have raised pre-operative suspicion. The presence of large multiple large nodules over the medial aspect of the left lobe, might have obscured a clear demarcation of the isthmus. A magnetic resonance imaging (MRI) would have provided a clearer clinical image of the thyroid.<sup>12</sup> This however, could not be performed due to the poor financial status of the patient.

A high-level of suspicion for ectopic thyroid tissue is required in cases of thyroid dysgenesis, due to the higher incidence. No ectopic thyroid tissue was identified in this particular case.

## CONCLUSION

Thyroid isthmus dysgenesis is a rare clinical presentation which could be associated with ectopic thyroid tissue or agenesis of the thyroid the lobes. A clear determination of thyroid anomalies during pre-operative evaluation would contribute significantly to safer surgical outcomes and adequate surgical management.

## CONFLICTS OF INTEREST

The authors declare that they have no conflicts of interest.

## REFERENCES

- Ozkan OF, Asik M, Toman H, Ozkul F, Cikman O, Karaayvaz M. Agenesis of isthmus of the thyroid gland in a patient with graves based own disease and a solitary nodule. *Case Rep Surg*. 2013; 2013: 608481. doi: [10.1155/2013/608481](https://doi.org/10.1155/2013/608481)
- Vayisoglu Y, Ozcan C, Gen R, Eti CM, Sut H, Gorur K. Thyroid isthmus agenesis associated with thyroid papillary carcinoma. *J Craniofac Surg*. 2013; 24: e428-429. doi: [10.1097/SCS.0b013e3182942d5c](https://doi.org/10.1097/SCS.0b013e3182942d5c)
- McDougall IR. Thyroid structure, development and developmental abnormalities. In: McDougall IR, ed. *Thyroid Diseases in Clinical Practice*. 1<sup>st</sup> ed. London: Chapman and Hall; 1992: 4-5.
- Van VG. Development of the thyroid gland: Lessons from congenitally hypothyroid mice and men. *Clin Genet*. 2003; 63: 445-455. doi: [10.1034/j.1399-0004.2003.00107.x](https://doi.org/10.1034/j.1399-0004.2003.00107.x)
- Ranade AV, Rai R, Pai MM, Nayak SR, Prakash, Krisnamurthy A, et al. Anatomical variations of the thyroid gland: Possible surgical implications. *Singapore Med J*. 2008; 49: 831-834.
- 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](https://doi.org/10.1186/1757-1626-0002-0000006640)
- Clifton-Bligh RJ, Wentworth JM, Heinz P, Crisp MS, John R, Lazarus JH, et al. Mutation of the gene encoding human TTF-2 associated with thyroid agenesis, cleft palate and choanal atresia. *Nat Genet*. 1998; 19(4): 399-401. doi: [10.1038/1294](https://doi.org/10.1038/1294)
- Sankar KD, Bhanu PS, Bapuji P. Agenesis of isthmus of thyroid gland with embryological and clinical basis. *Narayana Medical Journal*. 2012; 1: 32-34.
- Gangbo F, Lacombe D, Alberti EM, Taine L, Saura R, Carles DN. Trisomy 22 with thyroid isthmus agenesis and absent gall bladder. *Genet Couns*. 2004; 15: 311-315.
- Pastor VJF, Gil VJA, De Paz Fernández FJ, Cachorro MB. Agenesis of the thyroid isthmus. *Eur J Anat*. 2006; 10: 83-84.
- Duh QY, Ciulla TA, Clark OH. Primary parathyroid hyperplasia associated with thyroid hemiagenesis and agenesis of the isthmus. *Surgery*. 1994; 115(2): 257-263.
- Yaman H, Durmaz A, Arslan HH, Ozcan A, Karahatay S, Gerek M. Thyroglossal duct cysts: evaluation and treatment of 49 cases. *B-ENT*. 2011; 7: 267-271.