# Thyroid isthmus agenesis: A rare congenital anomaly

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## Abstract
Congenital anomalies of thyroid gland like hypoplasia, ectopic thyroid, hemiagenesis or agenesis of gland are commonly reported in literature. Agenesis of thyroid isthmus is very rare, with only few cases reported and most of them on cadaveric dissection. We hereby report a case with right thyroid colloid goiter, planned for hemithyroidectomy and incidental intraoperative finding of isthmus agenesis.

**Keywords:** Thyroid gland, Agenesis of isthmus, Malformations.

## Introduction
Thyroid gland is a most notable and labile gland that varies greatly in its size and structure.[1] Many diseases of thyroid need surgical intervention and thus require good knowledge of its normal and variant anatomy. Persistence of pyramidal lobe and thyroglossal duct cyst are the commonest anomalies, while the rare one are agenesis of partial or total, aberrant thyroid and agenesis of isthmus.[2]

The isthmus joins the two thyroid lobes in there lower part and lies between infrahyoid strap muscles anteriorly and 2nd to 4th tracheal cartilages posteriorly. When there is complete and congenital absence of thyroid isthmus it is termed agenesis.

It is challenging to assess the incidence of agenesis of thyroid isthmus as most previously reported cases were of patients presenting with other thyroid illness. Isolated thyroid isthmus agenesis is asymptomatic and subjects have normal levels of thyroid hormones.[3]

## Case Report
A 20 year female presented to our out-patient department with right lower neck swelling for last 9 months. On examination it was 5cm x 4cm, cystic and moving on deglutition. There was no other swelling or regional lymphadenopathy. Patient was biochemically euthyroid without any symptoms of hypo or hyperfunction of thyroid. There was no previous history of any surgery on neck. The fine needle aspiration cytology suggested colloid goiter, which was further confirmed by neck ultrasonography. All other routine blood investigations were within normal limits. The patient was taken up for right hemithyroidectomy for cosmetic reason. Intra-operatively, surgical exploration revealed complete absence of thyroid isthmus. Both the lobes were completely separate without any intervening thyroid tissue or even fibrous band (Fig. 1). No ectopic thyroid tissue in the migration track of the thyroid from the base of the tongue was seen. Subsequently right lobectomy was performed. Histologic examination gave the diagnosis of colloid goiter.

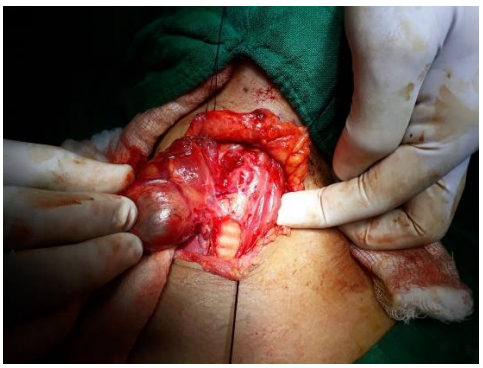

**Fig. 1: Intraoperative clinical picture showing cystic right thyroid lobe and normal left lobe with agenesis of thyroid isthmus**

## Discussion
Agenesis of thyroid isthmus is a developmental anomaly in humans, though it is absent in amphibians, birds, marsupials and rodents[4]. In rhesus monkey also the thyroid glands are normal in position with no isthmus[4]. All more advanced primates as well as humans, have the thyroid formed by two lateral lobes joined by an isthmus in front of trachea.

Embryologically, the thyroid gland develops from the thyroglossal duct, endodermal derivative of primordial pharynx, at the level of 2nd and 3rd pharyngeal arch. It descends downwards and its caudal end bifurcates, to give rise to thyroid lobes with isthmus. Simultaneously the cephalic end of the thyroglossal duct degenerates.[5] High separation of thyroglossal duct can provoke two independent thyroid lobes with or without pyramidal lobes with the absence of isthmus.[6] This may be due to mutation of one of three genes associated with thyroid gland development (TITF1, PAX8, FOXE1/TITF2).[7]

Most of the literature about congenital thyroid abnormalities is related with hemiagenesis which include one lobe and sometimes the isthmus. The important investigation to diagnose isthmus agenesis is ultrasonography, but the companionship of other pathology deflects the attention and lead astray. Same may have happened with our radiologist, as they failed to pick up absent isthmus pre-operatively.

Agenesis of isthmus is usually asymptomatic and diagnosis is often incidental due to some other thyroid pathology.[8] This variation should be kept in mind during tracheostomy procedure.[2] It can be diagnosed via ultrasonography, scintigraphy, Computed tomogram or Magnetic resonance imaging. When diagnosed it is necessary to take detailed history of any previous surgical procedure in the neck region (isthmectomies for neoplasms, decompressive procedure for thyroiditis or transthyroid tracheotomies).[9]

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