

AGENESIS OF ISTHMUS OF THYROID GLAND– A CASE REPORT

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ABSTRACT

The thyroid gland is a highly vascular endocrine gland placed anteriorly in lower neck in level with the fifth cervical to the first thoracic vertebrae ensheathed by pretracheal layer of deep cervical fascia and composed of two lateral lobes connected by a narrow median isthmus giving an H shaped appearance to the gland. A wide range of morphological variations and developmental anomalies of the thyroid gland have been reported in literature. We report a case of agenesis of isthmus of thyroid gland seen during routine dissection of cadavers in the Department of Anatomy at Sri Devaraj Urs Medical College, Kolar. The agenesis of isthmus of thyroid gland may be due to high separation of thyroglossal duct which can engender two independent thyroid lobes with absence of isthmus.

Key words - Thyroid gland, Isthmus, Agenesis.

INTRODUCTION

Agenesis of the thyroid isthmus is the complete and congenital absence of the thyroid isthmus. Thyroid gland is well known for its congenital anomalies.¹ Phylogenetically, in some species in which the thyroid follicles are organized in a gland, this gland can acquire a bilobate shape, in which the lobes join together in front of upper part of trachea by an isthmus or bridge of thyroid tissue.² The thyroid gland lies deep to sternothyroid and sternohyoid muscles. Accessory nodules/ ectopic tissue of the thyroid are usually found at the embryonic origin of the thyroid gland, at the foramen caecum or on the lingual duct and thyroglossal tract within the tongue.³ Thyroid isthmus agenesis does not cause clinical symptoms by itself and most of the time the diagnosis is incidental due to existence of any other thyroid pathology.⁴ Agenesis of isthmus can be diagnosed via scintigraphy, Ultrasound, Computer Tomography and Magnetic Resonance Imaging.

CASE REPORT

During routine dissection of an embalmed male cadaver

in the Department of Anatomy of Sri Devaraj Urs Medical College, Kolar, we observed a case where the thyroid gland showed absence of isthmus. The thyroid gland was composed of two enlarged lateral lobes each measuring 6 X 3.5 cms with no type of nexus between them. There were no scars in the cervical region in the cadaver suggesting that he had not undergone any surgery. There was no ectopic thyroid tissue between the root of the tongue to the glands position. The individual lobes were supplied by branches of superior and inferior thyroid arteries. There were no anastomoses between the superior thyroid arteries of the two sides. On the posterior border of the gland, anastomoses were identified between the branches of the superior and the inferior thyroid arteries.

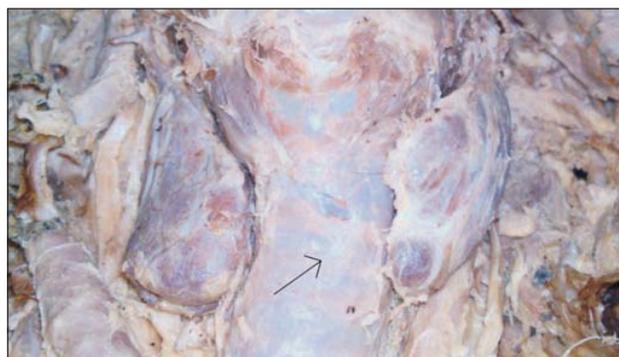


Figure 1 : Arrow indicates agenesis of isthmus of thyroid gland

DISCUSSION

Agenesis of the isthmus can be explained as an anomaly of the embryological development. Most of the variations of the thyroid gland are due to partial persistence of the median or thyroglossal duct. Failure of the development of the entire gland or part of the gland results in agenesis or hemi-agenesis which may be unilateral or isthmic³. Isthmus may be missing in the amphibians, birds and among mammals – monotremes, certain marsupials, cetaceans, carnivores and rodents. It seems that these differences do not have an evolutionary

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origin because the morphology does not result in any changes in the thyroid function, the follicular structure is similar in species that are distant from one other in taxonomic classification.¹ The morphological difference in evolutionary origin does not result in any changes in the thyroid function.

Pastor et al in their study had reported agenesis of isthmus of thyroid gland with enlarged lobes in a caucasian cadaver.² Won and Chung have reported that in 3% of the cases studied the isthmus was absent and the lateral lobes of the thyroid gland were separated.⁵ Marshall documented the variation in the gross structure of the thyroid gland in 60 children varying in age from a few weeks to 10 years and the absence of isthmus was reported to be 10% in this group.⁶ Ranade et al reported absence of isthmus in 35 out of 105 cases (33%) of which 8 were female cadavers.³ According to a study by Braun et al the isthmus was missing in 4 cases of the 58 cadavers they studied.⁷ We have come across 1 case out of 100 cadavers studied so far, hence our incidence is 1%. In our case study the agenesis of thyroid gland isthmus was not associated with any other anomalies of the gland. The glandular branches of superior and inferior thyroid artery did not anastomose in the median plane.

Embryological Hypothesis and Genetic Basis

Thyroglossal duct arises from the endodermal epithelium of the primordial pharynx at the level of second and third pharyngeal arch, when it descends downwards; its caudal end bifurcates and gives origin to thyroid lobes and the isthmus and at the same time cephalic end of the duct degenerates. This isolates it from the pharyngeal endoderm with the cessation of proliferation of the endodermal cells from which follicular cells of the gland are derived. Rarely, a high separation of the thyroglossal duct can engender two independent thyroid lobes and pyramidal lobes with absence of isthmus. Agenesis of isthmus can be associated with dysorganogenesis related to developmental anomalies of the thyroid gland such as absence of either lobe or presence of ectopic thyroid tissue in the nearest cervical region.⁸

The molecular mechanisms leading to thyroid dysgenesis are largely unknown. Studies in murine models and in a few patients with dysgenesis revealed that mutations in regulatory genes expressed in the developing thyroid are

responsible for this condition, thus showing that thyroid dysgenesis can be a genetic and inheritable disease.⁹ Genes associated with thyroid include several thyroid transcription factors expressed in the early phases of thyroid organogenesis (NKX 2.1/T/TF1, FOXE1/TITF2, PAX8, NKX2.5) as well as genes like the thyrotropin receptor gene (TSHR) expressed later during gland morphogenesis.¹⁰

CONCLUSION

An isolated isthmus agenesis is very uncommon. The true incidence is very difficult to be determined since the patients are found to be in non goitrous state. Thyroid isthmus agenesis does not cause clinical symptoms by itself and often the diagnosis is purely incidental due to the existence of other thyroid pathology. Mutations in regulatory genes expressed in developing thyroid has opened way to a novel working hypothesis on the molecular and genetic basis of this frequent human condition and render the thyroid an important model in understanding of molecular mechanisms regulating size, shape and position of the organ.

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