Abstract

Background: Morphological and developmental anomalies of thyroid gland have been reported in literature such as hypoplasia, ectopic thyroid, hemiagenesis or agenesis of gland. Agenesis of thyroid isthmus is a rare congenital anomaly and few cases were reported earlier.

Case presentation: A female cadaver in dissection hall was found with agenesis of isthmus of thyroid gland. From the right lobe, the pyramidal lobe arises with levator glandulae thyroideae.

Discussion: Agenesis of isthmus does not cause clinical symptoms by itself and diagnosis is secondary due to the existence of other thyroid pathology. This may be due to mutation of genes associated with thyroid gland.

Key words: Thyroid agenesis, agenesis of isthmus, pyramidal lobe, thyroglossal duct anomalies

Introduction

Thyroid gland (TG) is a most notable labile gland that varies greatly in its size and structure [1]. Being the first endocrine gland to start developing in the embryo [2], it is well known for its developmental anomalies, which distort the morphology of the gland causing clinical and functional disorders and various thyroid illnesses [3, 4]. Many diseases of TG require clinical and surgical intervention with good appreciation and knowledge of its normal and variant anatomy. Persistence of pyramidal lobe and thyroglossal duct cyst are the commonest anomalies of thyroid gland while the rare anomalies are agenesis of thyroid gland either partial or total, aberrant thyroid glands and agenesis of isthmus [5].

The isthmus of TG connects the lower parts of the two lobes and is usually anterior to the 2nd to 4th tracheal cartilages and is well protected anteriorly by the infrahyoid muscles.
**Discussion**

Agenesis of thyroid isthmus is a developmental anomaly reported earlier by anatomists and embryologists [3,4,5]. Phylogenetically the isthmus was absent in amphibians, birds and some mammals like monotremes, certain marsupials, cetaceans, carnivores and rodents. In rhesus monkey (Macacus rhesus), the thyroid glands are normal in position with no isthmus [7]. Usually isthmus agenesis is difficult to determine unless the patients refer for other thyroid diseases.

Embryologically the TG develops from the thyroglossal duct (TGD), endodermal derivative of primordial pharynx, at the level of 2nd and 3rd pharyngeal arch. It descends downwards and its caudal end bifurcates and gives rise to thyroid lobes with isthmus (bifurcation of TGD). Simultaneously the cephalic end of the TGD degenerates [8]; this separates the pharyngeal endoderm from the endodermic cells of the gland from which follicular cells develops (Fig. 2 A-E). It has also been noted that the C-cells of TG are derived from the caudo-pharyngeal endoderm/complex of 4th and 5th pharyngeal pouches [9]. High separation of TGD can provoke two independent thyroid lobes with or without pyramidal lobes with the absence of isthmus [10] (Fig. 2F). The etiology of agenesis of thyroid gland or isthmus is not clearly known and most of the isthmus agenesis is sporadic, familial or genetic predisposition [11]. Genetically the developmental agenesis is due to the result from mutations in one of three thyroid developmental genes (TITF1, PAX8, FOXE1/TITF2), especially TITF2, because these genes are more essential for the normal development of palate and TG [12, 13].

Most of the anomalies of TG are due to a 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 hemiagenesis, which may be unilateral or isthmic. Previous reports of agenesis of isthmus assert its rarity of its incidence [14]. According to Pastor Vazquez [7], the agenesis of isthmus can be associated with dysorganogenesis related to anomalies of TG. In this case, the agenesis of isthmus of TG was not associated with other anomalies of gland and clearly indicates that it was congenital. Because of the absence of isthmus, there were no anastomosis of STA and ITA of right and left sides. This type of variations should be kept in mind during transthyroid tracheotomy procedures [5]. This type of variations can be diagnosed via scintigraphy, ultrasonography, CT and MRI. When suspected, the individual may be directed for a differential pathological diagnosis such as autonomous thyroid nodule, thyroiditis, primary carcinoma, neoplastic metastases and infiltrative diseases such as amyloidosis through thyroid scan.

**References**


Figure 1. Agenesis of isthmus of thyroid gland. PY, pyramidal lobe; RL, right lobe; LL, left lobe; LGT, levator glandulae thyroideae; Arrow, arterial anastamosis of right STA & ITA

Figure 2. A – Schematic diagram showing the descent of TG; B-F showing the development of TG from the floor of primitive pharynx. Dashed lines represent the laryngo-tracheal junction. (TON, tongue; FC, foramen caecum; TGD, thyroglossal duct; TG, thyroid gland; IS, isthmus; HB, hyoid bone; TyC, thyroid cartilage; PP, pharyngeal pouches of 4th & 5