Hemiagenesis of thyroid gland - a rare presentation
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Abstract:
Thyroid dysgenesis (TD) due to embryonic defect in the development of thyroid represents a major cause of congenital hypothyroidism. Hemiagenesis of a thyroid lobe is a developmental anomaly in which one thyroid lobe fails to develop. It can be either due to complete absence or abnormal descent of one lobe. Several genetic mechanisms or environmental factors are proposed for defective lobulation or defective descent but conclusive evidence is yet to be postulated. We report a case of a 21 year old woman who presented with a gradually progressive neck swelling for 3 years. An ultrasound of the neck revealed absent left thyroid lobe with nodule right lobe and Technetium 99m Scintigraphy confirmed it. Thyroidectomy was performed with removal of right lobe and histopathology revealed adenomatous goitre with degenerative changes. This case is reported for its rarity and the importance of hemiagenesis in congenital hypothyroidism.

Keyword:
Hemiagenesis thyroid, Thyroid dysgenesis, Congenital Hypothyroidism

Introduction:
Hemiagenesis of a thyroid lobe is a developmental anomaly and a rare anatomic deformity in which one thyroid lobe fails to develop. It can be either due to complete absence or abnormal descent of one lobe. The anomaly is more commonly seen on the left side (ratio 4:1) and with a female preponderance (female: male ratio 3:1) [1]. It is usually diagnosed incidentally with unilateral thyroid enlargement or during work up of thyroid disorder [2]. The true prevalence of thyroid hemiagenesis is hence not known, however it is estimated to be about 0.02–0.05 % [2, 3]. We report a case of a thyroid hemiagenesis in a 21 year old female presenting with a nodule in right lobe with absence of left lobe.

Case report: A 21 year old woman presented with a gradually progressive neck swelling for 3 years (Figure 1). Examination revealed enlarged right lobe with a nodule. The left lobe of thyroid was not palpable. Biochemically she was hypothyroid and on thyroxine replacement. An ultrasound of the neck revealed absent left thyroid lobe (Figure 2) with nodule in the right lobe thyroid. A Technetium 99m pertechnatate Scintigraphy (Figure 3) was done and it was confirmative with the USG findings.

Discussion:
The development of the thyroid gland begins as a midline endodermal thickening in the floor of the pharynx. This thickening grows inferiorly as a diverticulum and is called the thyroglossal duct. This duct elongates in the course of development and its distal end becomes bilobed and this bilobed terminal due to epithelial proliferation expands to form the thyroid gland. The duct becomes a solid cord of cells. The thyroid gland now migrates inferiorly in the neck and passes either anterior to, posterior to, or through the developing body of the hyoid bone. By the seventh week, it reaches its final position in relation to the larynx and trachea. The thyroid gland now consists of two lateral lobes connected by a small isthmus. Agenesis or hemiagenesis of the gland or ectopic thyroid can occur in case of defect in the above process. Thyroid hemiagenesis is a rarer congenital abnormality characterized by the absence of one lobe of the thyroid gland with or without the absence of the isthmus. The exact aetiology of unilateral agenesis of the thyroid gland is not known. Several genetic mechanisms or environmental factors are proposed by various studies for defective lobulation or defective descent but conclusive evidence is yet to be postulated. [4, 5] It is unclear if the disturbance of the lobulation process is due to environmental factors or inherited genetic defect. Innovative experimental approaches using novel model systems are therefore required to enhance our understanding of the cellular and molecular mechanisms of thyroid development. The cellular dynamics of mammalian thyroid organogenesis are poorly understood due to the inaccessibility of embryonic thyroid tissue for live imaging.
One such promising model system in an organism distantly related to mammals from an evolutionary perspective is the zebrafish embryo toolkit according to Robert Opitz, Francesco Antonica et al where they found that the thyroid development is either absent or severely impaired in mutant embryos displaying defects in early endoderm formation [6]. Thyroid development is also compromised in no isthmus mutants carrying a null allele of pax2a [7, 8].

Another study by Henrik Fagman et al on Genetic Deletion of Sonic Hedgehog causes Hemiagenesis and Ectopic Development of the Thyroid in Mouse identified sonic hedgehog (Shh) as a novel regulator of thyroid development. Members of the hedgehog family are soluble ligands for patched receptors and act as key regulators of embryogenesis particularly for gut development. But the Shh-deficient mice display distinct features of thyroid dysgenisis. Whereas the thyroid anlage buds normally during early morphogenesis, the gland fails to separate into two distinct lobes and instead forms a single thyroid mass that is located unilaterally to the presumptive trachea and mostly to the left of the midline. Thyroid-specific transcription factors (TTF-1 and TTF-2) and thyroglobulin are expressed indicating terminal differentiation. Mice deficient in TTF-1 are athyreotic and TTF-2 knockouts either lack a thyroid gland or retain an ectopic, sublingual thyroid rudiment [7, 8].

In Pax2.1-null mutant mice thyroid follicular cells are lacking whereas C cells develop normally [9]. But few cases with TTF-1, TTF-2, or Pax8 mutations in humans with thyroid dysgenesis are identified. Thus the molecular defects underlying congenital hypothyroidism because of impaired thyroid development remain unknown. Almost all patients with thyroid hemiagenesis are diagnosed incidentally with most presenting with agenesis of left lobe (80%) and isthmus (44-50%) [2].The prevalence is higher in women (3:1) like our present case denoting a possible gender association [2]. Hyperthyroidism has been reported to be the most common clinical presentation leading to an incidental diagnosis of the hemiagenesis [10]. Our case presented with a palpable nodule right lobe with only the tracheal rings palpable on the left side and USG diagnosed the hemiagenesis. The confirmation of thyroid hemiagenesis was by a thyroid Scintigraphy which showed accumulation of tracer on one side within the thyroid region. A dominant autonomous functioning thyroid Scintigraphy which showed accumulation of tracer on one side within the thyroid region. A dominant autonomous functioning.

Conclusion:

Hemiagenesis is rare event in thyroid embryogenesis. It remains unsuspected until a disease of the existing lobe brings it under clinical attention. Radionuclide scan is the investigations of choice for accurate diagnosis. This case is reported for its rarity and the importance of hemiagenesis in congenital hypothyroidism which can be familial.

Figure 1: 21 year old female with nodule right lobe thyroid

Figure 2: High frequency Ultrasonography confirmed absent left lobe thyroid

Figure 3: thyroid Scintigraphy showing absent left lobe with nodule right lobe thyroid

Figure 4: Intra operative picture of right lobe thyroid with pyramidal lobe and absent left lobe and isthmus

References


