

Hemi agenesis of Thyroid Gland in a Euthyroid Child having Positive Family History of Thyroid Disorder

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ABSTRACT

Thyroid Hemiagenesis (THA) is a rare form of thyroid developmental anomaly. This developmental defect may or may not coincide with hormonal dysfunction. The detection of anomaly is quite incidental and infrequently found during investigations assessing thyroid functions. A three years old boy presented with gradual swelling in front of neck during last 2 months. He had no history of delayed milestones. Ultrasound imaging and thyroid scan by 99m -Technetium- pertechnetate revealed hemi agenetic development of left lobe of thyroid gland. His biochemical assessment showed euthyroid state. He had positive family history with hypothyroid maternal grandmother and congenital hypothyroid maternal cousin. Ultrasonography and thyroid scan are useful tools for demonstrating this form of dysgenesis of thyroid gland. Follow up monitoring of thyroid function at regular interval in children with congenital anomaly of thyroid gland should be done to take timely measures, if required.

Key words: THA-Hemi agenesis of thyroid gland, Thyroid scan, Ultrasonography

INTRODUCTION

Developmental anomaly of thyroid gland is quite rare. Common anomalies include persistence of pyramidal lobe and thyroglossal duct cysts. Rare anomalies include agenesis or hemi-agenesis of thyroid gland, agenesis of isthmus alone or aberrant thyroid gland (1). The detection of anomaly is quite incidental and infrequently found during investigations of thyroid functions. Ultrasonography along with thyroid scan using Technetium-99m Pertechnetate is useful tool for determining rare congenital anomaly of thyroid gland. Hemi agenesis of thyroid gland (THA) means non-development of single lobe with or without development of thyroid isthmus during embryological period. The incidence rate of hemi agenesis varies in different studies; researchers

have estimated an occurrence rate of less than 0.2 % with female population more sufferer than male. Hemiagenesis an asymptomatic developmental defect is usually detected incidentally and may or may not present with hormonal dysfunction. All forms of functional status including euthyroid, hypothyroid and hyperthyroid state can be observed in THA (2). Here, a case of THA in a three year old boy with euthyroid state and positive family history is presented.

CASE REPORT

A three-year-old male was presented with gradual onset of swelling in front of the neck for last 02 months. The painless swelling was not associated with dysphasia or dyspnoea. There was no history of tremors, palpitations, or change in weight or bowel habits. According to his mother his birth was via vaginal delivery and was of without any complications. There was no history of delayed mile stone development. According to his parents, his maternal grandmother and one of his cousins on his mother side are both suffering from thyroid disorder. However exact type of thyroid condition cannot be mentioned except for his baby cousin who is only 06 months old had been taking thyroxine medications from birth, which may suggest a congenital hypothyroid state. On clinical examination of the neck of this presenting baby revealed a soft swelling slight right lateral to midline of neck, which moved freely with deglutition but did not regress during protrusion of tongue. There was no cervical lymphadenopathy and no eye changes. Thyroid hormone assay revealed euthyroid state [Total T4-114.5 n mol/ml (normal 54-173 n mol/ml), TSH -1.45 m IU/L (normal 0.3-5.0 m IU/L)]. Ultrasonography (USG) study showed right thyroid lobe measured about 10.5 mm in AP diameter with isthmus was about 4.4 mm and the left lobe was absent. Ultrasonographic findings were consistent with left thyroid lobe hemiagenesis. Thyroid scan with Technetium-99m (^{99m}Tc) pertechnetate confirmed non visualization of left lobe suggesting the absence of the left thyroid lobe, with homogeneous uptake in the slightly enlarged right lobe and isthmus the characteristic hockey stick sign pattern on scan. No ectopic thyroid tissue was found along its developmental descending path or in any other area. The case was confirmed as left lobe hemi agenesis of thyroid without isthmic involvement. Regarding the possibility of developing hypothyroidism or hyperthyroidism in the future, follow-up with thyroid function tests was advised after 06 months along with fine needle cytological study from the right lobe.

DISCUSSION

Thyroid gland develops from the median endodermal thickening in the floor of the pharynx later form the median diverticulum, which grows caudally as bifurcating tubular ducts to form the

lateral lobes and isthmus. Abnormalities in the development may result in defective organogenesis or descent, complete or partial absence of the gland with or without ectopic thyroid tissue. Hemiagenesis, first described in 1866 by Handsfield-Jones is a form of thyroid dysgenesis in which, one thyroid lobe fails to develop with or without agenesis of the isthmus (3). Accurate incidence of THA is unknown as the patients are clinically asymptomatic. Several studies showed that the prevalence rate of the THA in the normal population is 0.05–0.2% (3-5). Gursoy et al. have indicated the prevalence of TH as 0.025% in the normal population and 0.25% in patients with thyroid disorders (6). In hospital-based studies done in adults, the disorder is seen more common in females with a male: female ratio of 1:3 (5). However, in a prevalence study conducted in healthy children, the male to female ratio was 1.4:1 (4,5). The absence of the left lobe was detected in 80% of cases, and agenesis of the isthmus was seen in 50% of cases (5). In accordance with the literature, left lobe was absent in our case.

In our study, the boy's maternal grandmother had been suffering from hypothyroidism and one of his cousins who had been suffering from congenital hypothyroidism suggesting inherited genetic defect. However ultra sonographic study of his father and clinical history of both his parents suggest clinical euthyroid state.

Clinical and biochemical presentation of THA are highly variable. Although patients with THA may have a normal thyroid lobe with euthyroidism, both hypothyroidism and hyperthyroidism are known to occur (7). The thyroid function may be altered in 38% to 47% of all THA cases (8). Disorders that have been reported in THA include hypothyroidism, hyperthyroidism, multinodular goitre, chronic thyroiditis, adenocarcinoma, and papillary carcinoma. Majority of these patients will however be euthyroid although hyperthyroidism is the most common abnormality in THA (8). Researchers have shown THA to be associated with an increased risk for developing thyroid disorders (6, 9). The presented boy was unsuspecting of any thyroid disease during his development period despite very positive family history. His milestone development and intelligence was normal and during examination he seemed to be very cooperative and understood each command suggesting his adequate neurological development. The family got only concerned when they observed a small swelling in front of his neck and due to their prior knowledge of our successful congenital screening program contacted at our institute for which we duly performed several screening examination for evaluation of his thyroid gland. The soft swelling in front of his neck was palpated just right lateral to midline and it did not regress during tongue movement differentiating it from thyroglossal cyst. It freely moved with deglutition and started just two months ago also separating it from any branchial cyst clinically.

Thereafter several routine examinations were done. On ultrasound examination, only right thyroid lobe could be visualized. Its antero posterior diameter of 11.5 mm which seemed slightly enlarged and might be as a result of prolonged TSH stimulation. Decreased heterogeneous echogenicity is less unusual as in our case normal homogeneous parenchymal echotexture was observed. On the agenesis side, the indentation of the sternothyroid and sternohyoid muscles into the thyroid bed were observed. (Figure 1)



Figure 1: Ultrasound imaging of thyroid gland showing agenesis of left lobe in a three-year-old boy.

The diagnosis of THA was established on the basis of sonographic assessment combined with scintiscan, which were performed to exclude the presence of functional thyroid tissue on the contralateral side to the lobe demonstrated in ultrasonography, and to visualize potential accessory or probable ectopic thyroid tissue located ectopically (10). Scintigraphic scan using Technetium 99m pertechnetate showed a uniform radiotracer concentration just right lateral to midline with tail expanded along its upper pole like a characteristic reverse J pattern creating the hocky stick sign (Figure 2).

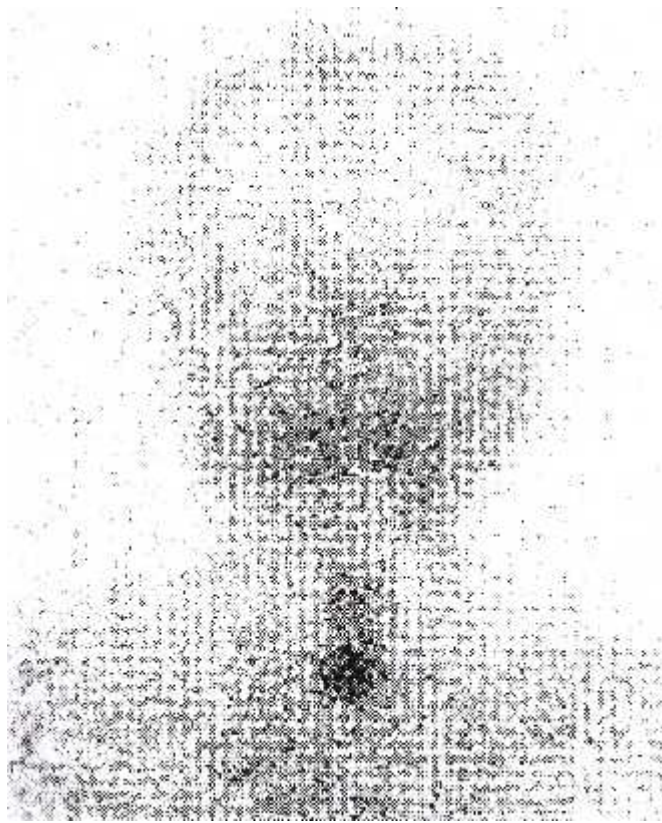


Figure 2: Scintigraphic image of hemi agenesis of thyroid gland in a boy showing hockey stick type of radio tracer concentration at right side of lobe, No radio tracer seen at left lobe.

However, Several clinical conditions like contralateral autonomous solitary thyroid nodule suppressing normal extranodular tissue; focal or unilateral subacute thyroiditis; primary or metastatic carcinoma; post inflammatory atrophy of thyroid tissue as a result of Hashimoto's disease; infiltrative diseases may also show unilateral radiotracer concentration. So, functional hemiagenesis based on thyroid scan alone may be misdiagnosed as congenital THA. Biochemical assay was suggestive of euthyroid state including normal TSH level. Therefore, thyroid scans should be confirmed by other methods revealing the morphology of the thyroid. Some authors had suggested CT or MRI scan to adequately see the morphologic pattern of the gland. However, they are expensive & ultrasonography seems to be suitable means to evaluate thyroid morphology adequately which is cost-effective, non ionizing and readily available.

As the absence of one lobe usually does not cause clinical symptoms unless there is a thyroid hormone imbalance and possible thyroid disease of the other developmentally normal thyroid lobe no further investigation was recommended during diagnosis. However, because of the high risk of goiter or hyperthyroidism, systemic follow up of all identified cases of THA was recommended. In our case we have also recommended regular follow up of six months interval.

CONCLUSION

It may be concluded that THA in a child may present with normal functioning thyroid gland. Therefore, there may not be any presence of any developmental problems. Careful approach must be taken for sudden swelling, as this was the history of present case. This swelling may be due to compensatory hyperplasia. In diagnosed THA cases follow up at certain intervals by biochemical assay together with ultrasound imaging are recommended to avoid subsequent thyroid dysfunction

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