Rare Report of a Familial Case of Lingual Ectopic Thyroid

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ABSTRACT

Thyroid developmental defects lead to 85% cases of congenital hypothyroidism (CH). Among those, few (about 2%) cases are familial, having dominant genetic predisposition with low penetrance. A case of ectopic positioned lingual thyroid gland are reported in a little girl and her father, where a routine and simple nuclear medicine scintigraphy of thyroid gland using 99mTc-pertechnetate played a vital role. In vitro analysis of thyroid hormones and high resolution ultrasound of neck aided the diagnosis. Screening of the family members and relatives in such cases appear to be essential.

Keywords: Congenital hypothyroidism, ectopic thyroid gland, lingual thyroid, Newborn screening, 99m Technetium pertechnetate thyroid scan

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INTRODUCTION

Congenital hypothyroidism (CH) occurs in approximately 1:2000 to 1:4000 newborns (1). Among those, 85% are caused by thyroid developmental anomalies (dysgenesis), with the remaining 15% caused by defects in thyroid hormone synthesis (dyshormonogenesis). Though the pathogenesis of thyroid dysgenesis remains unclear, 2% of familial cases are identified as having a dominant genetic predisposition with low penetrance (2). CH is one of the most common causes of preventable intellectual disability. Lower neurocognitive outcomes may occur in those who have untreated maternal hypothyroidism and started levothyroxine at a later age (>30 days of age). The clinical manifestations are often very delicate or may be absent at birth due to transplacental passage of maternal thyroid hormones (1). So, it is very crucial for a physician to detect CH before the age of 30 days.

We report a A rare case of familial thyroid developmental defect in a female child and her father, characterized by lingual ectopic thyroid tissue, absence of normal thyroid gland in position, and hypothyroidism.

CASE REPORT

An 11-year-old girl with a low socioeconomic background was referred to the thyroid division of Institute of Nuclear Medicine and Allied Sciences (INMAS), Kushtia, with the complaints of delayed growth and mental retardation and high TSH level (>100 µIU/ml). There was history of constipation and umbilical protrusion since birth associated with delayed developmental milestones. Physical examination showed low height and weight for age (Figiure 1A), subnormal intelligence, dry skin, hoarseness of voice, umbilical hernia, pseudo myotonia, and delayed relaxation of ankle jerk. There was no goiter, short round nose, cleft palate, low-set ear, abnormal movement, or macroglossia associated with pseudohypertrophy of oral muscle. Laboratory tests revealed hypothyroidism, with TSH= > 100 µIU/ml ,T3 = 0.56 ng/ml, T4= 12.68 ng/ml and it was suspected as a case of CH.

Imaging the neck with high-resolution ultrasound (HRUS) showed no thyroid tissue in the thyroid bed. However, a soft tissue mass like area having the tissue echopattern of thyroid gland was located in the midline at the upper neck region, posterior to the base of the tongue and measuring about 8mm X 7mm in size. Which was suspected as a lingual ectopic thyroid (Figure 2).

For confirmation, a 99mTc-pertechnetate scan was performed at the scintigraphy division of INMAS, Kushtia. Scan reported no radio-tracer activity at the thyroid bed region (Figure 1C & D), but focal increase radio-tracer activity at midline upper neck, which corresponds to HRUS findings of lingual ectopic thyroid (Figure 2). Regarding hormone profile, her thyroglobulin level is significantly low; anti-TPO Ab, TRAB, and anti-TG Ab are all in normal limits. Anterior pituitary hormones: LH, FSH, and

prolactin are all in normal limits. Interestingly, her parents did not seek any management since birth, as her father also has some common features (low intelligence, short stature, hoarseness of voice, umbilical hernia, constipation). They assumed that she was a normal child, phenotypically like her father. Other family members were screened to detect their thyroid status: her only brother, father, mother, and grandparents. All were found euthyroid except the father, who showed overt hypothyroidism. There was no history of maternal hypothyroidism during pregnancy as well. HRUS of the neck of the gentleman revealed empty thyroid bed with lingual ectopic thyroid tissue. Thyroid scan confirmed the diagnosis of lingual ectopic thyroid (Figure 1D). So, the reported case of CH turned out to be a rare familial thyroid dysgenesis i.e. lingual ectopic thyroid. Proper doses of Levothyroxine supplementation was prescribed to both the patients with assurance of lifelong follow up at the thyroid division of INMAS, Kushtia.

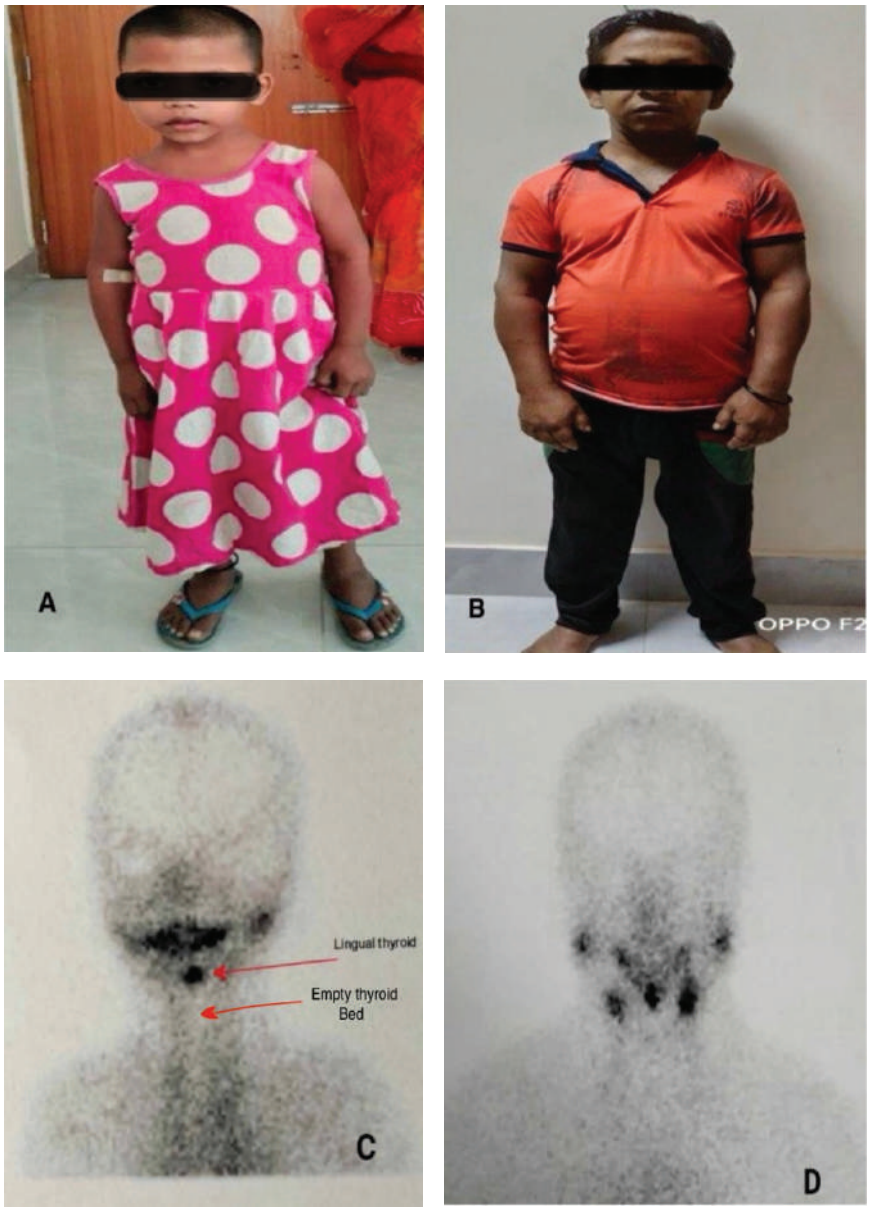

Figure 1: A. 11 years old congenital hypothyroid girl having delayed growth and development. B. Father of the girl having typical hypothyroid features. 99mTc- pertechnetate scintigraphy image of C. The 11 years old girl and D. her father. Both scan images show no radiotracer activity in the thyroid bed but focal areas of increased radiotracer activity in the upper mid neck (above the level of thyroid gland) near the base of the tongue, suspected lingual ectopic thyroid.

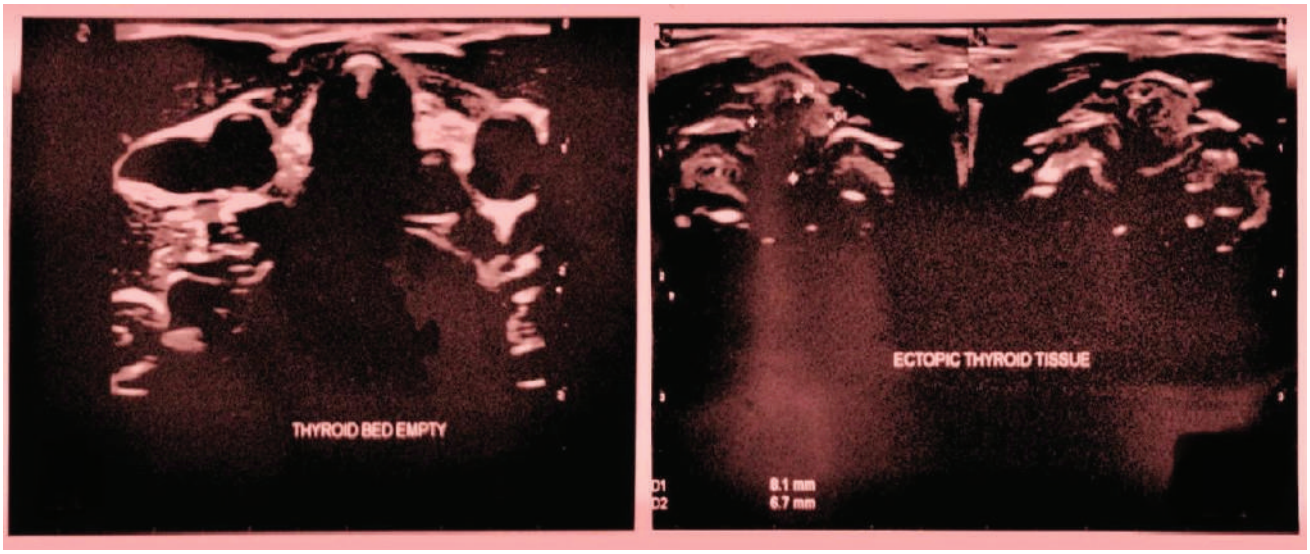

Figure 2: High resolution ultrasound images of neck of an 11 years old girl with congenital hypothyroidism, showing no thyroid tissue in the thyroid bed but a 8X7mm sized area resembling thyroid tissue posterior to the base of the tongue.

DISCUSSION

CH (CH) is classified into permanent and transient forms. Approximately 85% of permanent cases are attributed to thyroid dysgenesis, while 10–15% are due to dyshormonogenesis (1). Transient CH is commonly associated with maternal antithyroid medications, transplacental passage of thyroid receptor antibodies (TRAb), and maternal or infant iodine deficiency or excess. Rarely, CH can result from hypothalamic or pituitary dysfunction (3,4). Additionally, infants with trisomy 21 exhibit a higher incidence of CH (4).

While the exact etiology of permanent CH is not fully understood, familial cases of thyroid dysgenesis have been reported, accounting for approximately 2% of cases. A French study conducted between 1980 and 1998, involving over 2,500 patients with thyroid dysgenesis, revealed a significantly higher prevalence of thyroid dysgenesis among first-degree relatives, with the risk being 15 times greater than in the general population. Paternal-to-son transmission was observed, consistent with an autosomal mode of inheritance with incomplete penetrance. The study also highlighted a significant difference in the female-to-male ratio between familial and isolated cases of athyreosis, suggesting a potential role of sex-dependent etiological factors. The genetics of thyroid dysgenesis appears complex, possibly polygenic, and does not follow a simple Mendelian pattern of inheritance (2).

In our case, both the father and his only daughter were diagnosed with lingual ectopic thyroid, a form of thyroid dysgenesis. Importantly, no maternal factors associated with transient hypothyroidism were identified, and hypothalamic and pituitary hormones were normal. Additionally, the patient exhibited no features of trisomy. Although we were unable to screen the thyroid status of her paternal uncles, aunts, and cousins, their clinical history revealed no symptoms of hypothyroidism. Based on these findings, this case can be classified as a familial form of lingual ectopic thyroid.

Newborn screening for CH should be conducted in relatives of patients with thyroid dysgenesis, regardless of whether they exhibit hypothyroid features (2) . Screening is also recommended for offspring of individuals with thyroid dysgenesis, newborns with transient hypothyroidism associated with maternal thyroid dysfunction, and infants born in endemic goiter zones. The optimal sample for screening is cord blood or a postnatal sample taken after 72 hours of age to avoid false positive result . As the normal postnatal surge in TSH (60–80 mIU/L) occurs within hours after birth and resolves over the next five days (5). Early treatment of CH should start

immediately upon confirmation of diagnosis to prevent adverse outcomes.

Detailed diagnostic evaluation in CH cases are essential to differentiate between permanent and transient forms. Alongside thyroid hormone profiling, high-resolution ultrasonography (HRUS) of the neck and a 99mTc thyroid scan have to be done for determining the etiology. If imaging reveals a normal thyroid gland, the case is likely transient CH , and lifelong treatment may not be required. Reassessment should be performed after the age of 3 years by discontinuing treatment for 4 weeks and repeating thyroid function tests. If imaging was not conducted at the time of diagnosis, it should be performed after the age of 3 years to confirm the underlying cause (6). In confirmed cases of thyroid dysgenesis identified via a 99mTc thyroid scan, screening of family members and offspring is recommended to detect potential familial predispositions.

CONCLUSION

CH is one of the preventable causes of intellectual disability. Early detection and prompt treatment initiation can lead to better neurocognitive outcomes. Without newborn screening early detection is very much difficult. As 2% cases are familial, screening of relatives and family members and offspring of affected individual is very important. Along with thyroid hormone profile, HRUS of neck and 99m Tc thyroid scan plays important role in etiological diagnosis of CH.

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