Congenital Neck Mass: A Case Report

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ABSTRACT

Congenital goiter is a very uncommon cause of neonatal neck mass. Administration of antithyroid drugs or radiation exposure during pregnancy may result in reduced synthesis of thyroxine and cause congenital goiter with or without thyroid dysfunction. We report a case of a full term, newborn male baby who had normal thyroid functions but was born with a neck swelling and no apparent abnormalities. Maternal investigations for thyroid function tests were found normal as well.

Key words: Goiter, Congenital neck mass, Thyroid function tests

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INTRODUCTION

Congenital goiter is a rare cause of a neonatal neck mass. The majority of cases of congenital hypothyroidism (CH) are not hereditary and are caused by thyroid dysgenesis, maternal antithyroid drug ingestion, goitrogens, transplacental passage of maternal antibodies, tumors, and rare causes such as activated mutations of the TSH receptor and G-protein -subunit (McCune Albright syndrome). Even in the hereditary forms of goiter and thyroid dysfunction that often accompany it, it may not be evident at birth (1).

The majority of case reports on neonatal goiter in the literature were based on the recording of fetal goiter by antenatal scans, which needed the expertise of the radiologists. Furthermore, there is a lack of ultrasound machines and qualified manpower to carry out routine antenatal scans in resource-poor nations. Some of the highest incidences (1 in 1400 to 1 in 2000) have been reported from various locations in the Middle East (2). Implementation of a pilot newborn screening program is the primary goal for early detection (3).

CASE REPORT

A full-term male baby of 25 days, first born of a non-consanguineous parents was referred to NINMAS for thyroid function tests after the mother noticed an enlarged anterior neck mass on the seventh post-partum day. There was no family history of thyroid disease or hearing impairment. Apparently healthy, non-diabetic mother stated that her pregnancy period was uneventful with no previous history of radiation exposure, tuberculosis or miscarriage. The baby was delivered by C-section due to delayed progression of labor with normal birthweight (2.8 kilograms). Non-smoker mother of the baby had no history of taking anti-thyroid medications, iodine-containing drugs, goitrogens, or radioiodine therapy. She consumed commercially available iodized salt and no known thyroid disease. Antenatal thyroid function tests including antibody levels were normal.

On examination, the baby boy was apparently healthy without any feeding difficulties, excessive crying, or any evidence of hypoglycemia, jaundice, or other noticeable symptoms. No evidence of tongue protrusion or umbilical hernia existed either. Bladder and bowel habits were normal with no history of constipation since birth. Clinical examination revealed a swelling on the left side of the neck (Figure 1).

Figure 1: Visible swelling (arrow) in the left side of upper neck of 25 days old neonate

The mass was soft, mobile, non-tender and cystic. There were no inflammatory symptoms or audible bruit. The remaining part of the examination was unremarkable.

On the 20th day of the baby's life, a high-resolution ultrasound (HRUS) of the neck was performed. HRUS revealed that the right thyroid lobe was normal in size, shape, and position, but the area of the left lobe was occupied by a nearly rounded, well-defined, cystic mass measuring about 31 X 31 mm (Figure 2).

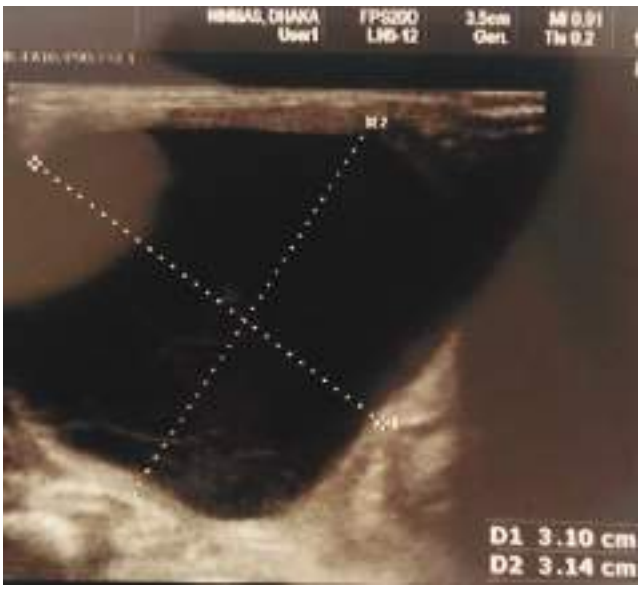

Figure 2: High resolution neck ultrasound image of 25 days old baby showing a fairly big, cystic lesion occupying the left lobe region of thyroid gland measuring about 31X 31 mm.

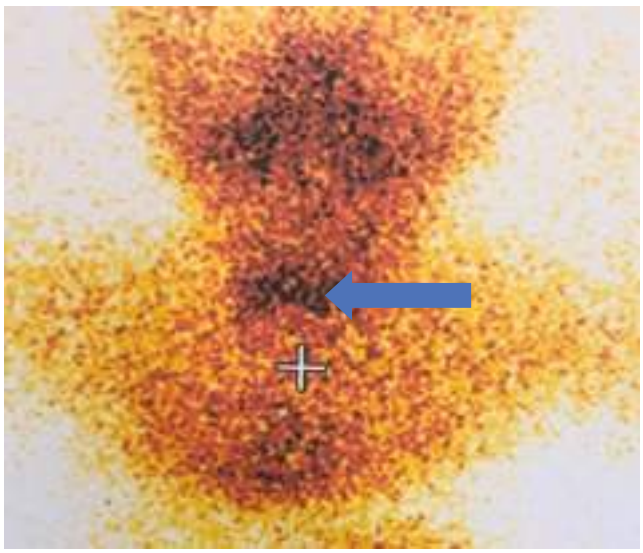

Figure 3: ^{99m}-Tc thyroid scan image of the same patient, showing decreased and patchy tracer concentration in left thyroid bed region.

Biochemical investigations showed, Hb= 9.8 gm/dl, PCV= 28%, TC= 11000/cumm, Platelet count= 254000/cumm, RBS= 150 mg/dl with negative C-reactive protein and normal chest radiograph. Thyroid scintigraphy with 01mCi intravenous injection of ^{99m}-Technetium revealed a decreased and patchy radiotracer concentration in the left lobe region (Figure 3). The right lobe of the thyroid gland was normal in size with a uniform radiotracer concentration. Serum thyroxine (T4 = 18.30 µg/dL) and thyroid stimulating hormone (TSH = 2.37µIU/mL) were found normal, suggesting a normally functioning thyroid gland. Hearing tests were normal as well. Further genetic tests were not done due to lack of genetic testing facility. Because the baby appeared to be healthy and thyroid function tests were within normal limits, he was not given medication and was monitored instead.

After 18 days, repeat HRUS of the thyroid gland showed regression of the size (about 21X14mm) of the left lobe nodule (Figure 4). Now the course of the nodule is uneventful though the baby is still under observation.

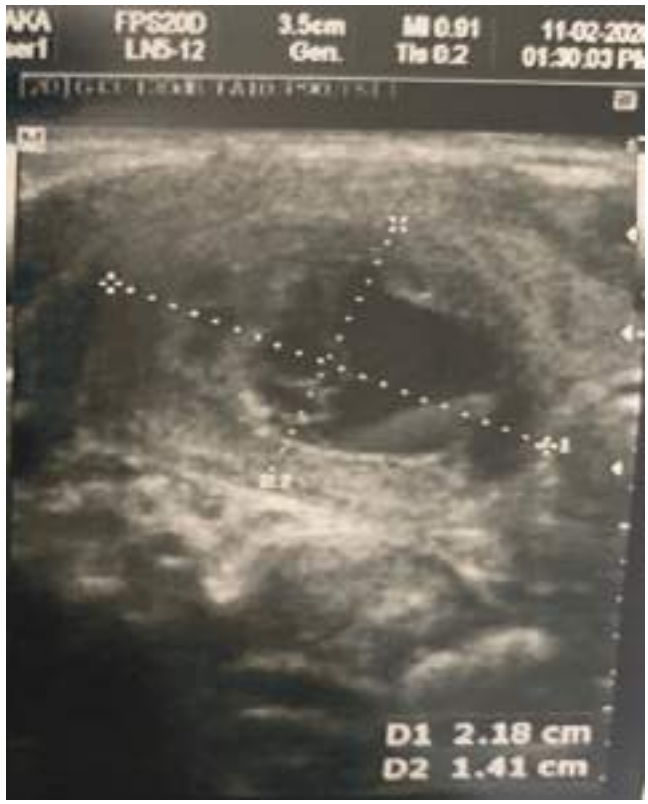

Figure 4: Repeated ultrasound image of same patient after 18 days, showing regression of size of the nodule, measuring about 21.8 X 14.1 mm.

DISCUSSION

Neonatal neck masses can be diagnosed clinically and with the help of imaging. Recognizing a mass may not be easy due to the difficulty of examining the neck of neonates. The most common causes of neck masses in newborns are cystic hygroma, lymphangioma, branchial cleft cyst, thyroglossal cyst, hematoma etc. These can be differentiated by their location, like cystic hygroma, which is the most common lymphatic malformation in children and usually presents as a painless, cystic, trans illuminated mass located superior to the clavicle; branchial cleft cysts found along the anterior margin of the sternocleidomastoid muscle; thyroglossal duct cyst, or enlarged thyroid, that may present with a midline neck; and hematomas, which may be the cause of a mass in the lower portion of the neck (4).

The baby was euthyroid in this reported case, whereas other studies have shown that a thyroid nodule in a newborn usually presents with congenital hypothyroidism. Clinical manifestations of goiter range from asymptomatic to an enlarged thyroid volume causing stridor, cyanosis, and respiratory distress due to airway obstruction, which may be life-threatening (5). Sometimes recognizing a neck mass may not be easy due to the difficulty of examining the neck of neonates and the insidious growth of some lesions that remain unnoticed.

Etiologically, congenital goiters develop as colloid goiters from iodine deficiency (less common in iodine-sufficient areas of the world) and a variety of inborn errors of thyroid hormone production. These defects are inherited as autosomal recessive traits, resulting in varying degrees of hypothyroidism. Programs for newborn screening (NBS) are important for promoting public health and are regarded as the most comprehensive preventive medicine system. The neonates are examined for certain conditions that are not immediately symptomatic at delivery but that, if left untreated, could have a long-term negative impact on the baby's health (6). So newborn screening is an important tool for detection of thyroid disorder.

Reported case showed a cystic mass in HRUS of neck, whereas other studies found the neck swelling to be non-cystic, almost solid, and mobile, with no inflammatory signs. Authors also reported slow regression of neck nodule after levothyroxine replacement but this 25 days old baby showed spontaneous regression of the cystic neck swelling. In a study of 28 patients, by Youn SY et al. eight infants had thyroid nodules and among them five (62.5%) had their cystic thyroid nodules disappear after thyroxine replacement and one infant (12.5%) showed a decrease in size and no change in two (25%) (7).

The thyroid nodules are classified as either cystic, mixed, or solid depending on their interior texture. Although it is known that 15%–30% of all thyroid nodules evaluated by palpation are cystic, many of the non–palpable thyroid nodules were found to be cystic. (7, 8). In this study, the baby showed that the size and cystic consistency of the thyroid nodule changed within 18 days after diagnosis which might be due to degeneration as there was no treatment done due to biochemical euthyroid status. Youn SY et al. (7) documented congenital hypothyroidism (CH) of the affected children with thyroid nodules. CH affects roughly 1 in every 4,000 newborns and is a significant preventable cause of mental retardation. The most common causes of permanent CH are dysgenesis or agenesis of the thyroid or a deficiency in thyroid hormonogenesis pathway. Less frequently, the altered neonatal thyroid function is temporary and is caused by a) iodine excess or shortage b) maternal blocking antibodies or c) the transplacental transfer of maternal medications. (9). In a newborn with a defect in thyroid hormone synthesis, a hearing screening must be performed to rule out Pendred syndrome i.e. iodide organification defect and deafness (10).

Levothyroxine should be started with an initial dosage of 10-14 micrograms/kg per day immediately after the diagnosis of CH. According to AAP guidelines, child is monitored for T4 and TSH values every 1-2 months for up to 6 months, every 3-4 months from 6-12 months and every 6-12 months from 3 years to completion of the growth (11). This reported patient presented with thyroid nodule and euthyroid status but he is still under observant follow up to avoid symptoms related to hypothyroidism or thyroid nodules (i.e., dysphagia, dysphonia, discomfort, and dyspnea by local pressure).

Despite being underreported, cystic degeneration could result from thyroid dyshormonogenesis. We advise more frequent evaluation when nodules appear in these patients to avoid possible complications like CH or hemorrhagic degeneration of a cystic nodule (7).

The presence of goiter in a newborn with primary hypothyroidism suggests transient hypothyroidism or an intrinsic defect in thyroid hormone synthesis (11). The overall goals of treatment are to assure normal growth and development by restoring the serum T4 concentration. Compliance with the treatment plan, periodic follow-up, and adjustment of therapy are essential for a good outcome. Goiter in newborn infants is not seen frequently, but all pediatricians who deal with neonates should be in a position to recognize the syndrome, understand its cause, prognosis, and advise therapy (12).

CONCLUSION

An appropriate algorithm should be followed while evaluating thyroid nodules or incidentalomas in children. Thyroid function tests and autoantibody titers are the primary investigations. HRUS is a bedside, non-invasive test that is acceptable to children, but the complementary role of thyroid scintigraphy should be noteworthy as well. Ultrasound-guided FNA remains the gold standard, but if not possible, frequent control visits (at least once every 6 months) should be ensured while treating congenital thyroid nodules.

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