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NECK SWELLING IN A NEWBORN-A CASE OF CONGENITALGOITER

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ABSTRACT Congenital hypothyroidism (CH) occurs in approximately 1:2,000 to 1:4,000 newborns. The clinical manifestations are often subtle or not present at birth. This is likely due to trans-placental passage of some maternal thyroid hormone, while many infants have some thyroid production of their own . In case of maternal normal thyroid levels, dyshormonogenesis was considered to be the most probable cause of hypothyroidism. Congenital hypothyroidism presenting as goiter in the newborn period is very rare. We present a case of primary hypothyroidism presenting as congenital goiter in the newborn period with symptoms in the immediate postnatal life. Hormone replacement therapy was started leading to normal levels of free thyroxine and triiodothyronine.

KEYWORDS :

INTRODUCTION

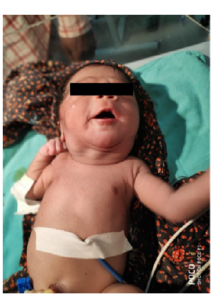
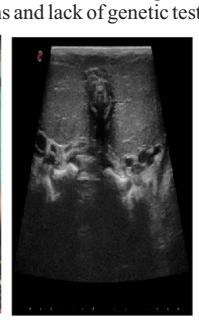
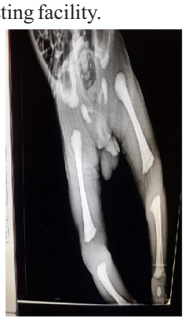
Congenital goiter is a rare cause of neonatal neck mass. Most cases of congenital hypothyroidism (CH) are not hereditary and results from thyroid dysgenesis, maternal ingestion of antithyroid drugs, goiterogens, transplacental passage of maternal antibodies and rare causes like activating mutations of the TSH receptor, activating mutations of the G-proteinα-subunit (McCune Albright syndrome), tumors. Even in the hereditary forms of goiter and thyroid dysfunction that often accompanies it may not be evident at birth. The prevalence of the CH based on neonatal screening is 1/4000 infants worldwide. Twice as many girls as boys are affected. Dyshormonogenesis represents 10-15% of all the causes of CH and most neonates would exhibit a relatively large goiter. Majority of case reports on neonatal goiter in literature were based on recording of fetal goiter by antenatal scans which is dependent on radiologist`s expertise.

CASE REPORT

A 3 days old term male baby born to a 25 year old mother ,second order in a nonconsanginous marriage having normal antenatal course, clinically euthyroid with no past history of thyroid disease, delivered by LSCS due to delayed progression of labour with birth weight of 2.8 kg, presented with swelling over the neck since birth ,delayed passage of meconium, poor activity and feeding difficulty.

On admission baby had stable vitals and general examination revealed midline neck swelling which is soft, mobile, noncystic, without inflammatory signs and no bruit. On per abdomen examination, there was soft distension, without hepatosplenomegaly or ascites. Rest of the Systemic examination was normal except for generalized hypotonia.

Investigations revealed Hb 15.9 gm%, PCV 49.9, TC 3730, Platelet count 3.4lakhs, RBS 150 mg/dl, CRP negative, free T3 40 IU/ml (normal 2.6±1.8 IU/ml) ,TSH >150 µIU/ml , X-ray abdomen normal , X-ray of knee showing absence of the distal femoral epiphysis, postnatal ultrasound of thyroid showed diffuse enlargement of thyroid gland. Hearing tests were normal. Further genetic tests were not done due to financial constrains and lack of genetic testing facility.

DISCUSSION

Although the diagnosis of neonatal neck mass can be made on clinical grounds and with imaging, recognizing a mass may not be easy due to difficulty of examining the neck of neonates and insidious growth of some lesions. Neck masses in the newborns may be differentiated by their location and include the following: Cystic hygroma; lymphangioma that is the most common lymphatic malformation in children, typically presents as a painless, transilluminated, soft mass located superior to the clavicle; Branchial cleft cysts, palpated along the anterior margin of the sternocleidomastoid muscle; Hematomas, which may be the cause of mass in the lower portion of the neck; Thyroglossal cyst or enlarged thyroid that may present with a midline neck mass.The clinical findings presenting with goiter vary from asymptomatic to enlarged thyroid volume causing stridor, cyanosis and respiratory distress by airway obstruction that can be a serious emergency .

Thyroid dysgenesis (aplasia, hypoplasia or an ectopic gland) is the most common cause of CH, accounting for 85% of cases; 10% are caused by an inborn errors of thyroxin synthesis, and 5% are the result of transplacental thyrotropin receptor blocking antibodies (TRBAb). Transient CH may occur when drugs prescribed for the mother such as propylthiouracil, methimazole or iodides cross the placenta and block the fetal thyroid gland.A variety of defects in the biosynthesis of thyroid hormone results in CH; these are detected in 1/30,000-1/50,000 live births in the neonatal screening programs. These defects are transmitted in an autosomal recessive manner. Defects may be of iodide transport, organification and coupling, deiodination and thyroid hormone transport. The exact cause of thyroid dysgenesis is unknown in most cases. Thyroid dysgenesis occurs sporadically, but familial cases occasionally have been reported. Three transcription factors TTF-1, FOXE 1, and PAX-8 are important for thyroid morphogenesis and differentiation; mutations in these genes are associated with thyroid dysgenesis. Another transcription factor NKX2.1 has been reported to result in CH with persistent neurological problems including ataxia, despite early thyroid hormone treatment.

The initial dose of thyroxine in a term infant is 50 µg daily for the first 1-2 weeks and should be started promptly at the initial visit when screening results are abnormal and serum sample have been sent for confirmatory tests. At the end of 2nd to 4th week, serum T4 and TSH values should be measured to determine that the amount of L-Thyroxine is adequate but not excessive. Therapy should be adjusted to maintain the serum T4 levels during infancy in the upper half of the normal range to optimize developmental outcome. Concomitant administration of soy formula, calcium, iron and high fiber foods may interfere with absorption of the L-Thyroxine and should be avoided when possible. Discontinuation of L-Thyroxine therapy for 4 weeks

duration sometime after 3 years of age is a way of testing for transient CH. Although some experts suggest the use of radioisotope studies for all infants with suspected CH, others do not, and the thyroid scan is listed as an optional diagnostic study on the most recent American Academy of Pediatrics (AAP) guidelines.As per the 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. The presence of goiter in a newborn with primary hypothyroidism suggests transient hypothyroidism or intrinsic defect in thyroid hormone synthesis.Since mother is euthyroid and not on any antithyroid drugs, dyshormonogenesis was considered to be the most probable cause of the hypothyroidism. In this case, diagnosis and treatment of goiter due to hypothyroidism occurred even earlier than most of other reported cases due to unusual presentation of thyroid mass in the neck. The long-term follow-up in children with levothyroxine have shown normal mean IQ values, satisfactory school performance and minimal motor dysfunction. However, speech defects and minimal CNS defects have been reported. Infants who are treated adequately for CH since the first month of age have an excellent prognosis for normal intellectual function and linear growth. Infants who have prolonged fetal hypothyroidism, delayed skeletal maturation and low T4 values are most likely to have neurocognitive problems.

CONCLUSION

The overall goals of treatment are to assure normal growth and development and psychometric outcome similar to genetic potential, by restoring the serum T4 concentration as rapidly as possible to a normal range followed by continued clinical and biochemical euthyroidism. Compliance to treatment plan, periodic follow-up care and adjustment of therapy are essential for a good outcome. Goiter in newborn infants are not seen frequently but all pediatricians who deal with neonates should be in a position to recognize the syndrome, understand its cause and prognosis and to advise therapy.

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