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Case Report

Congenital Goiter Presenting as a Transient Neck Swelling in a Newborn: A Case Report

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ABSTRACT

Background: Goiter is not frequently seen in the neonatal period.
Case report: We report the case of a full-term neonate who was born with anterior neck swelling and observed to suffer from congenital goiter on examination. Thyroid function tests were within normal limits. Thyroid swelling gradually reduced in size during the 1st week of life. All the cases of congenital goiter that has been reported in the literature are associated with hypothyroidism. To the best of our knowledge, this study was the first case of congenital goiter with euthyroid status.
Conclusion: Goiter is rarely seen in newborn infants. All pediatricians who deal with neonates with this disease should recognize the disease, understand its cause and prognosis, and advise appropriate therapy.

Keywords: Congenital goiter, Congenital hypothyroidism, Case reports

Introduction

Congenital goiter is a rare cause of neck swelling in a newborn. Congenital goiter presents at birth either as diffuse or nodular enlargement of the thyroid gland. In this disorder, thyroid hormone secretion may decrease, increase, or be normal depending on the etiology. It may result from the maternal ingestion of antithyroid drugs or ingestion of goitrogens, transplacental passage of maternal antibodies, or inborn errors of thyroid hormone production (e.g., dyshormonogenesis). The diagnosis is made by confirming thyroid size with ultrasonography and hormonal assay. The treatment with thyroid hormone replacement is indicated when the congenital goiter is associated with hypothyroidism. Surgery is indicated once respiration or swallowing is impaired.

Case report

A full-term 3,200 g male baby was born of nonconsanguineous marriage to a 25-year-old primigravida mother at our hospital. The baby was delivered by the vertex vaginal delivery. The mother's antenatal history was insignificant with no history of drug or radiation exposure. The mother was clinically euthyroid during pregnancy with no history of any antithyroid medication ingestion. There was no history of hearing loss in the family. On admission, the baby had a heart rate of 143 beats/min, the respiratory rate was 68 breaths/min, SpO_2 was 97%, and the baby had inspiratory stridor. The patient was given oxygen support with nasal prongs. General physical examination revealed a bilateral swelling in the neck, which was soft, mobile, and noncystic, with no inflammatory signs, and no bruits were audible. There was no other abnormality on systemic examination.

The patient's complete hemogram showed a hemoglobin level of 15 gm/dl, total leukocyte count of 12000/mm^3, and platelet count of 2.5 lac. The patient C-reactive protein was negative, and the chest and abdomen X-ray did not reveal any abnormality. The ultrasound of the thyroid gland showed the diffuse enlargement of bilateral thyroid lobes (right: 4.0×3.6 cm, left: 4.2×3.4 cm) and isthmus with normal echotexture. Thyroid

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hormonal assays showed thyroid stimulating hormone (TSH) of 6.94 µIU/ml, total thyroxine

A B

Figure 1. A) Enlarged thyroid (1st day of life); B) significant reduction in size of swelling (8th day of life)

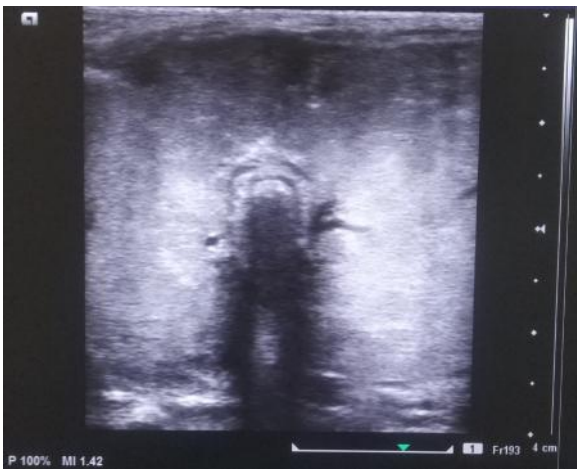

Figure 2. Thyroid ultrasonography showing enlargement of bilateral thyroid lobes (straight arrows) and trachea (curved arrow)

(T4) of 12.02 µg/dL, and total triiodothyronine (T3) of 3.26 ng/mL which were normal for the neonate age. (Figure 1, Figure 2)

The X-ray of the knee showed the absence of the distal femoral epiphysis. Further genetic tests were not performed due to financial constraints. As thyroid hormonal assay was within normal range, thyroid hormone replacement therapy was not started. In view of persistent stridor with the increased rate of breathing, surgical treatment was planned on the 3rd day of life, but the patient showed clinical improvement with a significant reduction in stridor and improvement in respiratory efforts. (Figure 3)

Thyroid hormone levels repeated on the 5th day of life showed TSH of 7.23 µIU/ml, total T4 of 14.30 µg/dL, and total T3 of 5.65 ng/mL which were also within normal limits. By the 8th day of life, there was a significant reduction in the size of the thyroid gland. Repeated thyroid hormonal levels at the time of discharge were also within normal limits (i.e., TSH of 6.70 µIU/ml, T3 of 4.14 ng/mL, and T4 of 11.32 µg/dL). The baby was

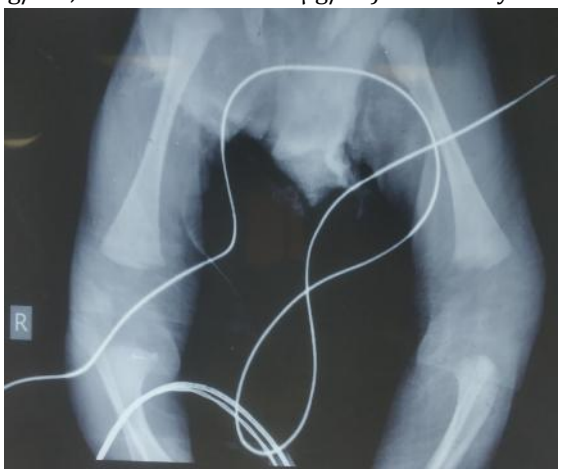

Figure 3. X-ray of knee joint showing absent distal femoral epiphysis

discharged on the 14th day of life and was regularly under follow-up.

Discussion

The differential diagnosis of neck mass in a newborn includes cystic hygroma, thyroglossal duct cyst, branchial cleft cyst, sternocleidomastoid tumors, hematoma, and enlarged thyroid. The differentiation of neck masses can be done based on the location of swelling. Congenital goiter is an infrequent cause of neck masses in neonates (1). It can be caused by a variety of inborn errors in thyroid hormone production (e.g., dyshormonogenesis), anomalies of thyroid embryology, in utero exposure to maternal antithyroid antibodies, and maternal ingestion of antithyroid drugs or other goitrogens.

Infants with congenital goiter might be associated hypothyroidism, hyperthyroidism, or normal thyroid function (2). A euthyroid goiter

presenting at birth is usually secondary to maternal antithyroid drug intake, transplacental passage of antibodies, and ingestion of goitrogens during pregnancy that can induce thyroid hyperplasia in a fetus (3). In the index case, the thyroid function tests of the mother were normal, and there was no history of antithyroid drug intake during pregnancy in mother. Further molecular analysis could not be done due to financial constraints.

The clinical findings presenting with congenital goiter vary from a completely asymptomatic neonate to enlarged thyroid volume causing stridor, respiratory distress, and cyanosis by airway compression that can require intubation and mechanical ventilation. In the index case, the patient had respiratory distress and inspiratory stridors. The patient required oxygen support in the form of nasal prongs.

Congenital goiter is usually associated with hypothyroidism in newborns. The prevalence of congenital hypothyroidism is approximately 1 in 4,000 births. It is an important preventable cause of mental retardation. A hypothyroid neonatal goiter presenting at birth is usually secondary to inborn errors in thyroid hormone production (e.g., dyshormonogenesis). Less commonly, the transplacental passage of maternal medications or maternal blocking antibodies can result in a hypothyroid neonatal goiter. Hearing screening must be done in every newborn with defects in thyroid hormone synthesis to rule out Pendred syndrome.

The optimal neurodevelopmental outcome depends on the early adequate treatment of congenital hypothyroidism. The overall goal of treatment should be to assure the adequate growth and mental development of the child as close as possible to their genetic potential. In newborns with congenital hypothyroidism, the use of L-thyroxine should be started at a dose of 10 to 15 µg/kg/day, with higher doses used for infants with the lowest T4 and highest TSH levels (4).

Ideally, the T4 level and TSH level will be normalized within 1 week and 2 weeks of starting therapy, respectively. Repeating T4 and TSH measurements should be performed 1 week after starting the therapy, every 1 to 2 weeks until thyroid hormone levels have been normalized, 2 to 4 weeks after any dose change, and every 1 to 2 months in the first year of life (5). Nonadherence to treatment can have serious permanent neurodevelopmental consequences for the infant and should always be considered when thyroid function tests fail to be normalized with treatment. Surgery is required when there is tracheal compression leading to respiratory compromise.

Conclusion

Goiter is rarely seen in newborn infants. All pediatricians who deal with neonates with this disease should recognize the disease, understand its cause and prognosis, and advise appropriate therapy.

Acknowledgments

None

Conflicts of interests

The Authors declare that there is no conflict of interest.

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