

Congenital Goiter, Is it a Medical or Surgical Disease?

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Presentation of congenital goiter (CG) with upper airway obstruction is uncommon. Limited literature is available with regards to the approach and outcome of such patients.

A two-month-old boy was born with diffuse goiter. He presented with stridor and respiratory distress requiring immediate intensive care unit (ICU) admission. Thyroid function tests, neck ultrasound, neck X-ray, CT neck and thyroid scan revealed diffuse goiter with retrosternal extension and airway obstruction. The neck swelling reduced and symptoms resolved following a conservative management.

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Congenital goiter (CG) is a rare entity. In the Middle East, a high incidence has been reported, 1 in 1,400 to 1 in 2,000¹. The causes of the condition include defective iodine uptake and transport, inborn errors of thyroid hormone production (dyshormonogenesis), transplacental passage of thyroid antibodies, maternal ingestion of antithyroid drugs or goitrogens and tumors^{2,3}. Dyshormonogenetic goiter represent 10-15% of all cases of congenital hypothyroidism (CH) and most neonates would exhibit a relatively large goiter⁴.

The aim of this presentation is to report a case of congenital goiter, the management options and the effectiveness of medical management.

THE CASE

A full-term baby boy, product of spontaneous vaginal delivery to a 31-year-old female, not known of any medical or thyroid disease and not on anti-thyroid medication. At birth, Apgar score was 9 and 9 at 1 and 5 minutes, respectively. On examination, the neonate was jaundiced with labored breathing, using the accessory muscles and with audible stridor. Neck examination revealed an anterior neck swelling, soft, cystic and non-tender. No signs of inflammation or lymphadenopathy. The abdomen was soft, lax, no guarding, rigidity or organomegaly. Systemic examination was unremarkable. Hearing assessment was normal.

Hemoglobin was 17.1 g/L, white blood cell count was 8.04 x10⁹/L and platelets were 210 x10⁹/L. Thyroid-stimulating hormone (TSH) was 100.00 uIU/ml, low serum free thyroxine (fT4) 7.51 pmol/l and low free serum triiodothyronine (fT3) 3.14 pmol/l. Glucose-6-phosphate dehydrogenase (G6PD) test revealed a hemizygous male. Coagulation profile was normal. Hemoglobin electrophoresis was negative.

Initial postnatal ultrasound revealed a normal size and echotexture of right and left thyroid lobes, no thyroid nodules

and no obvious soft tissue swelling. However, TC-99 MM thyroid scan reported prominent size thyroid gland with diffuse increased uptake. No hot or cold nodule. The total thyroid uptake was 42% distributed, 19.2% for right lobe and 22.8% for left lobe. High thyroid uptake suggested dyshormonogenesis. A diagnosis of CH with diffuse goiter due to dyshormonogenesis was contemplated. Thyroxine 25µg OD started on the seventh day of life and the patient was discharged.

At the age of two months, the patient suffered from recurrent cyanotic episodes with intermittent reduced feeding and activity. On examination, the neck swelling increased in size and the child was having labored breathing although hemodynamically stable. Oxygen saturation was maintained at 98% on room air, obvious biphasic stridor was heard, with signs of moderate respiratory distress using the accessory muscles, tracheal tag, pectus excavatum and intermittent circumoral cyanosis. Fiberoptic nasopharyngolaryngoscopy revealed a narrow hypopharynx, posterior prolapse of the tubular epiglottis and a short aryepiglottic fold. Vocal cords were not visualized. Instant high kilovoltage lateral and anteroposterior neck X-ray showed a significant narrowing of the airway, see figure 1.

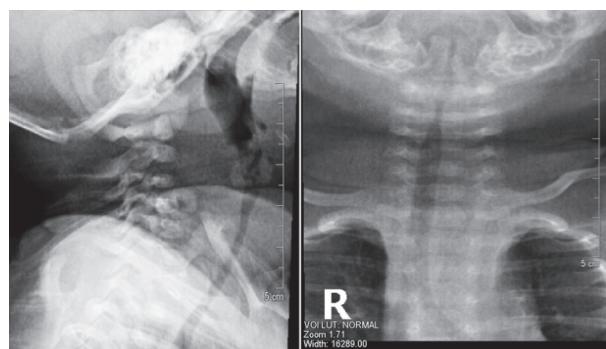


Figure 1: AP and Lateral Neck X-ray Showing Compromised Airway

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A repeat ultrasound revealed diffusely enlarged homogenous thyroid. The right lobe measured 1.5x2 cm and left lobe 2x2.8 cm. Slight left lobe retrosternal extension. The isthmus measured 6 mm. The tracheal transverse diameter was 4 mm and longitudinal measured was 6 mm, giving the impression of goiter with retrosternal extension and airway narrowing.

The patient was admitted to the ICU, kept on three liters of oxygen through a nasal cannula, dexamethasone and thyroxine 25 μ g. Forty-eight hours later, the oxygen was tapered off and the stridor had improved.

CT neck and upper chest with intravenous contrast revealed marked thyromegaly measuring approximately 45.5x42.2x22.7 mm (height, width and depth) with retrosternal extension and narrowing of the air space opposite the laryngeal region, see figure 2.

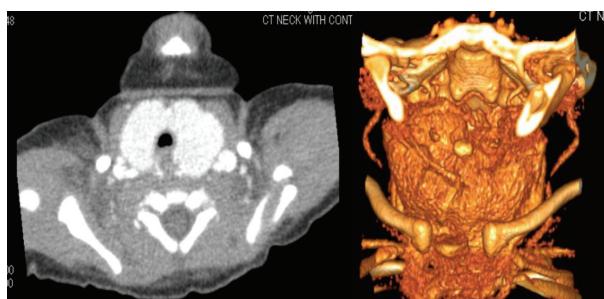


Figure 2: 3D CT Scan Showing Diffuse Thyroid Enlargement and Airway Narrowing

Due to potential airway obstruction with hemorrhage, fine needle aspiration was not performed.

TSH decreased to 0.33 μ IU/ml on serial testing, see figure 3. The mother was not complying with the administration of his thyroxine doses, therefore, a social worker was involved. The importance of compliance to medication was emphasized, and complications were thoroughly explained to the mother and maternal psychiatric support was provided. The patient was discharged on thyroxine 50 μ g.

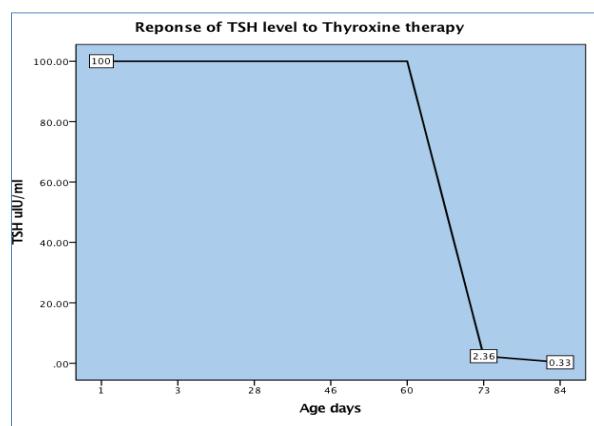


Figure 3: Response of TSH Level to Thyroxine Therapy

At the age of 5 months, the patient's developmental milestones are corresponding to his age. He has no palpable goiter and is in good condition.

DISCUSSION

CG presents as a neck mass at birth or afterbirth, occasionally accompanied by CH. It is a preventable cause of neurologic impairments with a prevalence of 1:4000 births³. In our case, CH with diffuse goiter was attributed to dyshormonogenesis. The two predominant causes of dyshormonogenesis are the defective organification of iodine and the defective synthesis and secretion of thyroglobulin synthesis. Hence, impaired thyroid hormone synthesis increases TSH secretion resulting in a compensatory goiter².

Presentation varies from asymptomatic to enlarged thyroid with stridor, cyanosis and respiratory distress by airway obstruction³. Large goiter can compress the trachea and obstruct respiration².

American Academy of Pediatrics (AAP) recommends that any infant with low thyroxine (T4) and TSH >40 mU/L is considered to have primary hypothyroidism and replacement with levothyroxine (L-T4) should be initiated as soon as confirmatory tests are obtained. Soy formula, calcium, iron and high fiber foods must be avoided as they may interfere with thyroxine absorption⁴. Thyroid ultrasonography, Iodine-123 or Technetium-99m scans are optional and treatment must not be delayed to perform radiological imaging⁴. A hearing assessment must be performed to rule out Pendred syndrome³.

An initial dose of 10-15 μ g/kg of thyroxine has been recommended and is the treatment of choice⁴. Occasionally, this disorder may be identified prenatally and treated by injecting T4 into the amniotic fluid³.

Pediatric endocrine diseases requiring operative interventions are relatively rare⁵. Surgical intervention for CG is considered if compressive symptoms present, including discomfort, pain, dysphagia, dysphonia or difficulty of breathing when lying flat. Uninodular goiters are amenable to lobectomy while multinodular goiters require near-total or total thyroidectomy⁵. A multidisciplinary approach that involves pediatric endocrinologists, pediatricians, surgeons, nuclear medicine physicians, anesthesiologists, and pathologists is an essential part of the long-term management of patients who undergo thyroidectomy⁵. Early detection of CH with goiter is of paramount importance to initiate the management process as soon as possible.

A case was reported of a preterm male infant with antenatally detected goiter presenting as a neck mass with CH, similar to our case; medical therapy resulted in mass resolution and normalization of thyroid hormones⁶. Another case reported a patient with CH having a large intrathoracic goiter compressing the trachea causing dyspnea and stridor⁷.

CONCLUSION

Pressure symptoms from CG could take place. On such occasions both conservative and surgical correction options should be explored. The conservative choice is favored when surgical intervention is thought to increase the morbidity and cause further complications.

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