Congenital goitre and respiratory distress as rare presenting features of congenital hypothyroidism in the newborns

Khalid A. Makki
MD, MRCPH, MB.BS, Paediatrics specialist, Ahmed Gasim Specialized Hospital for Children, Khartoum North, Sudan | Silla Hospital, SEHA, AL Dhafra region, UAE.

**Abstract**---In babies with congenital hypothyroidism, congenital goiter and respiratory distress (due to upper airway compression/hypothyroidism) are rarely encountered as the first presenting signs. Computerized Tomography of the neck revealed enlargement of all thyroid gland lobes and further laboratory revealed hypothyroidism. Thyroid hormone replacement therapy was initiated early, which resulted in a significant reduction in respiratory distress and goiter regression. With a high TSH, a low Tri-iodothyronin, no central line abnormality, no maternal history of drug/s use during pregnancy, and no familial history of hypothyroidism, Congenital Goitrous Primary Hypothyroidism is the most likely diagnosis.

**Keywords**---Goiter, respiratory distress, newborn, primary hypothyroidism.

**Introduction**

Sudan has yet to implement a nationwide newborn screening program for (CH), and home births are still common in many parts of the country, including the capital. Congenital primary hypothyroidism, occurring in approximately 1:2000 to 1:4000 newborns, is one of the most common preventable causes of intellectual disability worldwide. There is an inverse relationship between age at treatment initiation and intelligence quotient (IQ) later in life. (1)

Most newborns with Congenital Hypothyroidism (CH) have few or no clinical manifestations of thyroid hormone deficiency. In addition, the majority of cases are sporadic, so it is not possible to predict which infants are likely to be affected. For these reasons, newborn screening programs were developed to detect this condition as early as possible by measuring either thyroxin (T4) or thyrotropin (thyroid-stimulating hormone [TSH]) in heel-stick blood specimens. (2)
The most common causes of Primary Hypothyroidism (PH) are thyroid gland dysegenesis 85% and dyshormonogenesis (caused by one of the inborn errors in thyroid hormone synthesis and secretion) 15%. [3] Thyroid dysegenesis resulting from agenesis, hypoplasia, or ectopy. Thyroid ectopy accounts for two-thirds of the dysgenesis cases worldwide. [4] More than 95% of infants with congenital hypothyroidism have few, if any, clinical manifestations of hypothyroidism at birth. [5]

Infants born in regions of the world that lack newborn screening programs typically present with symptoms and signs of hypothyroidism that develop over the first few months of life, which include lethargy, hoarse cry, feeding problems, often needing to be awakened to nurse, constipation, puffy (myxedematous) and/or coarse facies, macroglossia, umbilical hernia, large fontanels, hypotonia, dry skin, hypothermia, and prolonged jaundice (primarily unconjugated hyperbilirubinemia). [6]

Newborn infants with thyroid dyshormonogenesis may have a goiter detected on prenatal ultrasound or on clinical examination of the neonate, while in others the goiter is discovered later in life. Palpable subcutaneous nodules (ossifications) may be a tip-off to congenital hypothyroidism caused by pseudohypoparathyroidism. [7] Clinical features related to central hypothyroidism, the clinical manifestations are often related to associated deficiencies of other pituitary hormones and include hypoglycemia, micropenis, undescended testes, and, least commonly, features of diabetes insipidus and hearing loss. [8]

Case report

Term baby boy, outcome of uneventful pregnancy, first born of consanguineous marriage to healthy parents with no medical history of thyroid disease/drugs. Antenatal care provided by the visiting registered (legal) midwife. Born through normal vaginal delivery at home labor assisted by registered (legal) midwife. Baby cried immediately after birth and was vigorous.

At the age of six hours, the patient was brought to the emergency room due to difficulty breathing and feeding, as well as swelling on the anterior aspect of the neck. (Figure 1). No family history of hyper/hypothyroidism and no history of congenital anomalies including deafness. The mother used only folic acid during pregnancy.
On admission, he was ill, distressed (RR=86/mn) and SPO2 (89%) at room air, Heart rate (190/mn), head pushed backward as spontaneous adaptation to the neck swelling, not dysmorphic (Figure 1.). 4*6 cm enlargement involving the anterior region of the neck, more to the right side, extending behind the sternum with ill-defined margins and smooth surface. Soft in consistency, neither pulsatile nor tender with normal skin over it. Trachea is difficult to palpate. Male external genitalia were normal and systemic examination was unremarkable. The anterior fontanel is open 4*3 cm and anthropometric measurements are within normal limits.

Patient’s preferred position (minimal neck hyperextension) was maintained, SPO2 was normalized (94-99 percent) with non-invasive breathing support (2-4 Lt/mn nasal prongs), septic screening was performed, and combination antibiotics were begun according to local protocol (Presence of two risk factors for sepsis, being delivered at home and the presented symptoms). After a period of NPO, oral feeding was gradually introduced as the respiratory distress improved.

Thyroid function done and showed high Thyroid Stimulating Hormone (TSH) (> 100 micromol/l) (N: 0.25-5), low thyroxin T4 18.37 micromol/l (N: 66-181) and normal Tri-iodothyronine (T3) (1.16micromol/l) (N: 1-3). Maternal Thyroid function test is normal. Lateral neck x-ray showed soft tissue swelling and tracheal deviation and compression (Figure 2). Ultrasound neck done showed picture of congenital lobulated goiter. Echocardiography, normal heart structure and function. Computed Tomography (CT)-Neck and chest: markedly lobulated homogenous enlargement of all thyroid gland lobes extending retrosternally and compressing the trachea. Figures (3)
Oral thyroid hormone replacement therapy (Levothyroxine 25 μg/each morning), initiated during the first week, noted that respiratory distress improved with initiation of Levothyroxine although the size of the neck swelling was slightly reduced. By the fifth day the patient had been weaned off oxygen, was being fed well orally and the antibiotics were discontinued by the sixth day due to negative septic screening. A paediatric endocrinologist was involved in the management plan early on, and the mother was well informed and taught about her child’s condition and the need to continue hormone replacement therapy along with regular follow-up at AHMED GASIM SPECIALIZED HOSPITAL FOR CHILDREN.

At the three-month follow-up, the thyroid swelling had subsided and was no longer visible on clinical examination, the patient's development was appropriate for his age, the neurological examination was normal, he was gaining weight (weighing 5 kg at the time on exclusive breast feeding), he had no coarse facial features, he passed hearing testing, and the mother was satisfied with the treatment and had good compliance (Figure 4). Repeated Thyroid function showed normal level of **TSH**, **T4** and normal **T3**. Levothyroxine dose increased to 50 μg/each morning. At 6 and 12 months of age, the patient was examined again and determined to have normal growth, development, and neurological exams.

Figure 2. Lateral neck X-ray.
Figure 3. CT-Neck and chest
Discussion

Prenatal diagnosis of congenital anomalies remains a challenge in low resource countries, and in many areas of Sudan, prenatal care is provided by midwives or sometimes by General practitioners (GPs) which make the prenatal radiological diagnoses of such congenital anomalies and treatment very difficult afterward. Congenital goitrous primary hypothyroidism suggests transient hypothyroidism or intrinsic defect in thyroid hormone synthesis (8). Also congenital goiter associated with in utero exposure to maternal anti-thyroid antibodies, or maternal ingestion of anti-thyroid drugs and other goitrogens. The infant may have associated hypo/hyperthyroidism, or have normal thyroid function.

Congenital goiter in newborns is not commonly seen in practice, which makes working towards exclusion / proof of surgical neck lumps more reasonable. The first impression in such neck swelling will be cystic hygroma or lymphangioma or hematoma or branchial cleft cysts. The diagnosis was obtained for this patient by first imaging and thyroid function subsequently.

The clinical manifestation of a goiter varies depending on its size, ranging from asymptomatic in a small goiter to respiratory discomfort and cyanosis in a large goiter (due to compression of the upper airways). Due to the implementation of a congenital hypothyroidism screening program, classic features of congenital hypothyroidism such as hoarseness, feeding problems, dry skin, constipation,
puffy (myxedematous) and/or coarse facies, macroglossia, umbilical hernia, large fontanels, hypotonia, hypothermia, and prolonged jaundice are no longer commonly seen. In most cases the diagnosis of hypothyroidism can be confirmed or excluded by results of serum tests of thyroid function, informing the decision to start thyroid hormone treatment.

Additional testing may be helpful in specific situation, like measurement of TSH-receptor blocking antibodies (in the mother and/or fetus) may be useful in diagnosing transient congenital hypothyroidism. Thyroid imaging like ultrasonography or radionuclide uptake measurements may provide information about the underlying etiology, eg, thyroid dysgenesis or one of the types of dyshormonogenesis \(^9, 10\). Other studies, such as serum thyroglobulin assay, or urinary iodine excretion, may be performed to identify the cause. These tests usually do not alter treatment; thus, they are considered optional and do not change management and can be considered optional.

The overall goals of management are early detection of congenital hypothyroidism and early initiation of thyroid hormone replacement therapy to restore serum fT4 (or T4) and TSH concentrations to the normal range as rapidly as possible, and this may avoid brain damage ensuring normal growth and neurodevelopmental outcome.

**Conclusion**

Huge goiter and reparatory distress can be presenting features of congenital hypothyroidism, and early initiation of thyroid replacement therapy can improve respiratory distress as well as a rapid decrease in the size of the goiter.

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**Reference**