

# Fetal Goiter in the Absence of Maternal Thyroid Disease: A Case Report

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## Abstract

**Objective:** Most believe that it is important to be able to recognize and treat the fetal hypothyroidism in order to get the most out of growth and intellectual development in affected fetuses.

**Case:** A case has been introduced dealing with a woman contracted with fetal goiter that was identified by ultrasonography at 30 weeks of gestation. In Doppler examinations, it was realized that the thyroid gland was highly vascularized and diffusely enlarged. Under these circumstances, fetal goiter may only have something to do with fetal hypothyroidism. The patient was offered to get through amniotic fluid sampling via amniocentesis or cord blood sampling via cordocentesis, but she rejected the performance of these procedures.

**Conclusion:** Our priority is, whenever the situation permits, to trust in the ultrasonographic measurement of goitre size and color doppler signal, since it is of vital importance to be able to recognize and observe the fetal goiter based on ultrasound and Doppler examination.

**Keywords:** Fetal goiter, ultrasonography, prenatal diagnosis.

## Maternal tiroid hastalığı yokluğunda fetal guatr: bir olgu sunumu

**Amaç:** Etkilenen fetusların çoğunda fiziksel ve zihinsel gelişme geriliklerine yol açabildiğinden, fetal hipotiroidizmin tanınması ve tedavi edilmesinin oldukça önemli olduğu düşünülmektedir.

**Olgu:** Bu yazıda 30 haftalık iken ultrasonografi ile tespit edilmiş bir fetal guatr olgusu sunulmuştur. Hastanın tiroid hastalığı öyküsü mevcut değildi ve tiroid otoantikolarının negatif oluşu da dahil tüm tiroid fonksiyon testleri normaldi. Dopler ultrasonografide diffüz olarak büyümüş ve yüksek oranda kanlanan tiroid bezi izlendi. Bu durumda fetal guatrın sadece fetal hipotiroidizme bağlı olabileceği düşünüldü. Kesin tanı için amniosentez veya kordosentez yaptırması önerilen hasta bunu kabul etmedi.

**Sonuç:** Bu durumda önceliğimiz, klinik durumun tanı ve takibinin öneminden dolayı, ultrason bulguları ve dopler ölçümlerine güvenmek oldu.

**Anahtar Sözcükler:** Fetal guatr, ultrasonografi, prenatal tanı.

## Introduction

Thyroids disorders are common endocrine disorders encountered during the perinatal period. It is very difficult to identify and diagnose the fetal goiter, while maternal thyroid

abnormalities can be easily diagnosed applying maternal serum testing.<sup>1</sup> Different biochemical defects in thyroid hormone synthesis, or maternal autoimmune thyroid disease might cause fetal goiter.<sup>2</sup> Goiter might have association with fetal hypothyroidism or hyperthyroidism. Many

authors, on this issue, believe that fetal thyroid function has to be determined for beginning the early treatment.<sup>3</sup> Weiner et al reported that they diagnosed the fetal goiter by means of pre-natal sonography for the first time in 1980.<sup>4</sup> A very large goiter inside uterus might cause polyhydramnios because of esophageal and tracheal compression, and distocia as well, leading to hyperextension in the neck.<sup>5</sup>

### Case

The patient was a 28 year-old primigravida who referred to our hospital at 30 weeks of gestation for having ultrasound examination. The ultrasound examination indicated a large homogeneous mass in the anterior aspect of the fetal neck. The mother was married to a second-degree cousin. There was no family history of thyroid or autoimmune diseases and no maternal history of a past thyroid disease. There was no known guatrogens including iodine or thyroid medications. Following ultrasonography a wide, symmetrical bilobated thyroidal mass and a mild polyhydramnios, compressing the fetal trachea in the anterior portion of the neck, were detected. The mass appeared highly vascularized during the Power Doppler examination (Figure 2). After birth, color Doppler examination showed a diffusely enlarged thyroid gland (Figure 3).

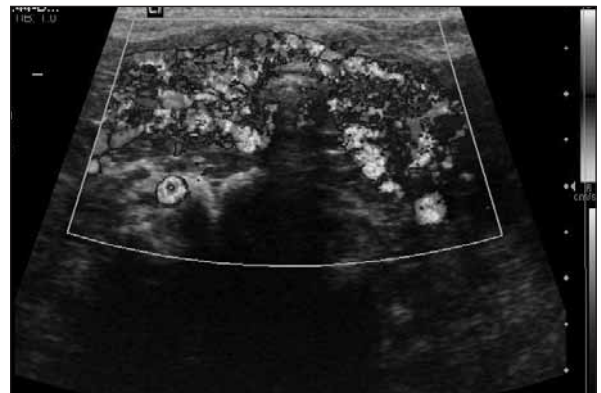
Fetal heart rate was about 220 beats/min. No other anomaly was noted in the fetus. Fetal growth and movements were normal. Maternal serum thyroid function test results were normal. Ultrasound of the maternal thyroid was usual. We suggested amniotic fluid sampling via amniocentesis or cord blood sampling via cordocentesis to the mother but she refused the suggestion. The patient was advised for bed rest to avoid premature labor. We did not attempt to obtain fetal blood to confirm fetal hypothyroidism, however we proposed intra-amniotic l-thyroxin injections for to start early treatment.



**Figure 1.** Symmetrical bilobated thyroidal mass.



**Figure 2.** Highly vascularized mass.



**Figure 3.** After birth, color Doppler examination showed a diffusely enlarged thyroid gland.

Both parents did not allow this procedure too. We tried to rely on ultrasonographic measurement of goiter size and color Doppler signal

when applicable. At 36 weeks of gestation the patient is hospitalized for premature rupture of membranes and delivered a male newborn, weighing 2600 g by caesarean section because of the narrow pelvis. His Apgar scores were 6 at 1 min and 7 at 5 min. Although a soft bilobated goiter was present, neonatal airway obstruction was not observed and resuscitation was not needed. He had no problem in respiratory adaptation postnatally. Measurement of TSH and iodothyronines in the cord blood confirmed the diagnosis of primary hypothyroidism. Thyroid hormone therapy commenced on the first day of life with a daily oral dose of 50 µg levothyroxine. The weight, height and psychomotor development of the child were normal at 6 months of age.

## Discussion

Incidence rate of congenital hypothyroidism is one out of 4000 live birth, causing the mental retardation that can most commonly be treated.<sup>6</sup> It is very seldom (one out of 4000) to encounter fetal goitrous hypothyroidism, constituting only 10 to 15% of all congenital hypothyroidism cases.<sup>7</sup> Because of the development of ultrasound technology, reports on the investigations of fetal goiter, despite being a rare incidence, has gradually been increasing.<sup>8</sup>

A large goiter may cause hyperextension of the fetal neck, resulting in malpresentation and complicating labor and delivery. Following the birth, the trachea may be blocked by goiter, which may cause asphyxia and death. During delivery, pediatric anesthesia and pediatric ear, nose and throat consultants have to be present in an adjoining theater with intubation and bronchoscopy equipment set up. Neonatal screening programs have successfully been used for diagnosing congenital hypothyroidism shortly after birth, and the prognosis for normal development has dramatically improved with earlier postnatal treatment. However some

infants exposed to congenital hypothyroidism have encountered difficulties and delays in neuromotor, perceptual and language abilities, despite early postnatal therapy. Therefore, antenatal treatment of congenital hypothyroidism has to be taken into account and given priority.<sup>9</sup>

Pathological cases, including the thyroid gland, can easily be distinguished from cases with other neck lesions detected at ultrasound. The differential considerations of fetal goiter should include all anomalies of the anterior and anterolateral nuchal region, including teratomas, thyroglossal duct cysts, cystic hygromas, lymphangiomas/hemangiomas, branchial cleft cysts and other developmental cystic lesions.<sup>10</sup> These lesions frequently appear as fluid-filled cystic masses. This finding enables the differentiation of the neck lesions from thyroid gland masses.<sup>11</sup>

Amniotic fluid concentrations of TSH accurately reflect fetal serum levels, but Bruner and Dellinger consider cord blood measurements more reliable, thus render evaluation through amniocentesis doubtful. Fetal thyroid function can be accurately assessed by fetal blood sampling, but this procedure is riskier with about 1 % fetal demise in experienced hands.<sup>12</sup>

We reported this situation to the family members but the parents rejected the suggestion of performing amniocentesis and/or fetal blood sampling. We observed the size and fetal development of the fetal goiter until delivery, using the ultrasound and Doppler examination.

## Conclusion

As a result, ultrasound and Doppler examination are of vital importance in recognition and observation of fetal goiter. Therefore, mental retardation and other developmental (evolutional) disorders could be prevented in the incidence where early diagnosis and treatment have already been utilized.

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