

## Two cases of fetal goiter

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### ABSTRACT

**Introduction:** Anterior fetal neck masses are rarely encountered. Careful routine ultrasound screening can reveal intrauterine fetal goiters (FGs). The incidence of goitrous hypothyroidism is 1 in 30,000-50,000 live births. The consequences of both FG and impaired thyroid function are serious. **Aims and Objectives:** To emphasize role of ultrasound in both invasive and non-invasive management of FG. **Materials and Methods:** Two pregnant patients, during second trimester, underwent routine antenatal ultrasound revealing FG, were investigated and managed. **Results:** Case 1: Revealed FG with fetal hypothyroidism. Intra-amniotic injection l-thyroxine given. Follow-up ultrasound confirmed the reduction of the goiter size. At birth, thyroid dyshormogenesis was suspected and neonate discharged on 50 mcg levothyroxine/day with normal growth and development so far. Case 2: Hypothyroid mother with twin pregnancy revealed FG, in twin 1, confirmed on magnetic resonance imaging (1.5 × 1.63 cm). The other twin had no thyroid swelling. Cordocentesis confirmed hypothyroidism in twin 1. Maternal thyroxine dose increased as per biochemical parameters leading to reduction in FG size. Mother delivered preterm and none of the twins had thyroid swelling. Fetal euthyroidism was confirmed on biochemical screening. **Conclusion:** FG during pregnancy should be thoroughly evaluated, diagnosed and immediately treated; although in utero options for fetal hypothyroidism management are available, emphasis should be laid on non-invasive procedures. Newer and better resolution techniques in ultrasonography are more specific and at the same time are less harmful.

**Key words:** Fetal goiter, hypothyroidism

### INTRODUCTION

Fetal goiter (FG) is a rare, serious condition during pregnancy. Careful routine ultrasound screening can reveal intrauterine FGs, which usually appear as symmetrical homogeneous masses at the anterior of the neck. Primary congenital hypothyroidism is seen in 1/3,000-1/5,000 deliveries all over the world. Here we present two cases of FG.

### CASE REPORTS

#### Case 1

A 37-year-old at 23 weeks of gestation. A routine antenatal ultrasound examination indicated a large homogeneous,

bilobed mass in the anterior aspect of the fetal neck [Figure 1a-c]. The patient, clinically euthyroid, had no history of thyroid disease in the past. Pregnancy was confirmed at 12 weeks 5 days. Other fetal parameters for vitality and bone maturity were normal. The amniotic fluid volume appeared normal. A cordocentesis was performed at 27 weeks confirming fetal hypothyroidism with serum thyroid-stimulating hormone (TSH) >100 mIU/mL and fT4 was 0.2 ng/dL with low-serum total T3 and T4. Twenty-nine weeks of gestation onwards intra-amniotic injection l-thyroxine was given on weekly basis. After 4 weeks ultrasonographic studies confirmed the reduction of the goiter size. The patient underwent lower section cesarean section (LSCS) at 37.2 weeks and delivered a male infant weighing 2690 g, with APGAR score of 9 at 5 min. No evidence of airway obstruction was seen. The newborn thyroid volume was barely palpable. The neonate was screened for congenital hypothyroidism wherein ultrasound showed slightly enlarged goiter (possibly due to dyshormonogenesis) with no other congenital deformities. He was discharged on 50 mcg levothyroxine/day with normal growth and development so far.

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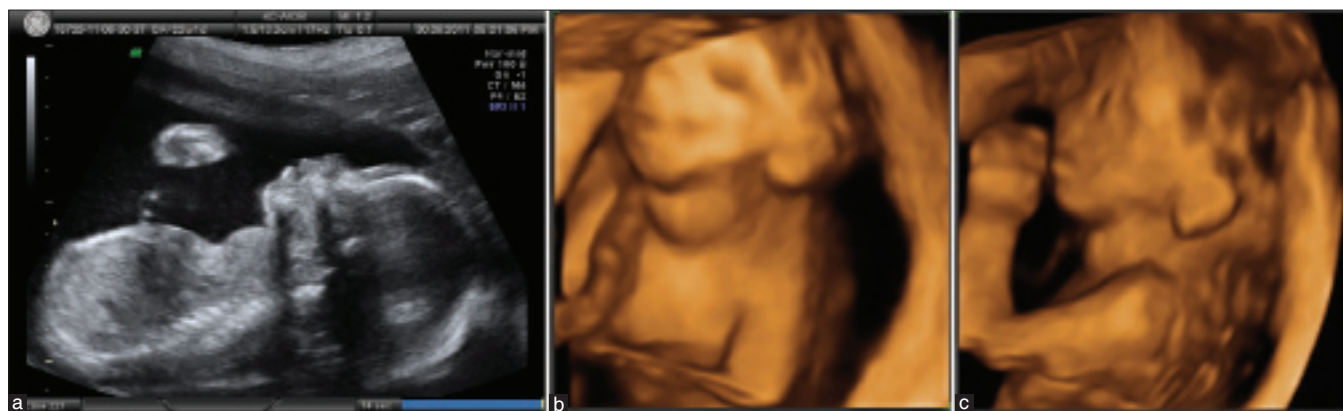
## Case 2

A 27-year-old hypothyroid woman on thyroxine supplementation (75 mcg) having dichorionic diamniotic pregnancy underwent a routine antenatal ultrasound at 24 weeks of gestation, which revealed a large, butterfly shaped, uniformly echogenic solid mass ( $1.5 \times 2.6$  cm) suggestive of FG, in twin 1 [Figure 2a and b]. The thyroid swelling was confirmed on fetal magnetic resonance imaging (MRI) which showed a  $1.5 \times 1.63$  cm sized neck swelling. The other twin had no thyroid swelling. Cordocentesis was done and fetal free T4 values were 0.29 in twin 1 and 0.77 in twin 2. Free T3 values were 0.45 and 0.98 in twin 1 and 2, respectively. Mothers thyroid function showed a TSH of 8.3 and a free T4 of 0.6 and a very high titer of anti-thyroperoxidase (TPO) Ab ( $>3000$ ). Dose of thyroxine was increased to 200 mcg and free T4 after 2 weeks was 1.4 mcg. Repeated sonography was done to monitor FG size, evidence of fetal cardiac dysfunction, delayed bone maturity and polyhydroamnios. Sonography confirmed reduction in size of goiter. Mother delivered preterm at 28 weeks and surprisingly, none of the twins had thyroid swelling, which was confirmed on fetal ultrasound. Fetal euthyroidism was confirmed on biochemical screening.

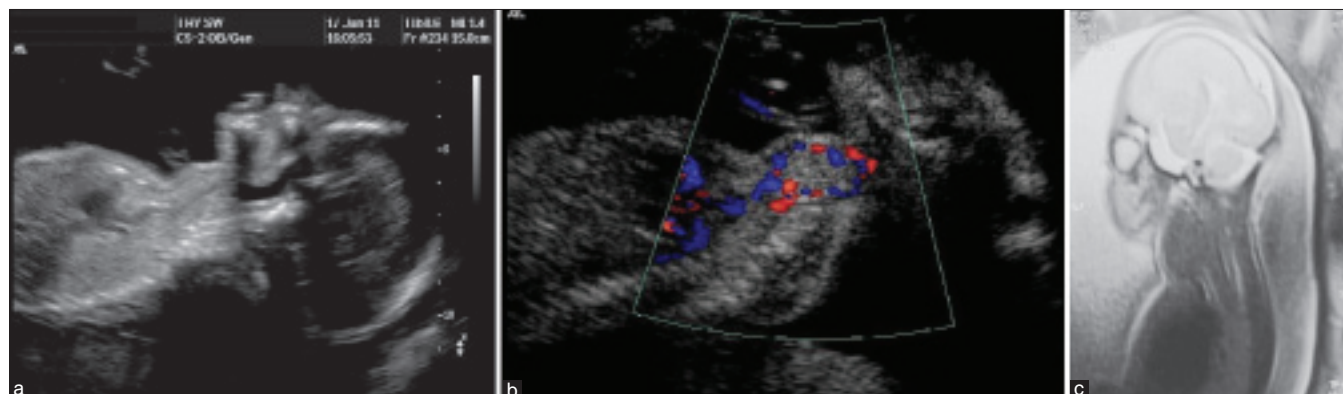
Anterior fetal neck masses are rarely encountered during the second or third trimester of pregnancy. Ultrasound screening can reveal intrauterine FGs, along with anatomical assessment for size, volume and compression of neck structures. MRI may be done for better details.

Usually, dysmorphogenesis is responsible for FG. Blockage of hormone biosynthesis increases fetal TSH levels, and this leads to FG. FG can lead to hyperextension of neck, polyhydroamnios, and compression of vascular structures in neck and edema. Mental and motor retardation also have been reported.<sup>[1]</sup> These reasons make fetal hypothyroidism important for early diagnosis and appropriate hormone-replacement treatment.

An ultrasonography (USG) score is also proposed including fetal heart rate (FHR), fetal movement, Colour Doppler ultrasound (CDU) vascularity and bone maturation.<sup>[2]</sup> The color Doppler indirectly indicates fetal thyroid function with the pattern of vascularity. Peripheral vascularity is suggestive of a nonfunctional hypertrophied gland, and central vascularization is suggestive of the overactive hyperfunctioning gland.



**Figure 1:** (a) Ultrasound image of case 1 (b) Ultrasound image of case 1 (c) Ultrasound image of case 1



**Figure 2:** (a) Ultrasound image of case 2 (b) Doppler image of case 2 (c) Magnetic resonance image of case 2

Maternal anti-TPO, anti-Tg titers suggest transient suppression of thyroid hormone synthesis in the fetus. Maternal thyroid stimulating hormone- receptors (TSH-R), thyroid receptors antibody (TRAb) can stimulate a fetal thyroid to hypertrophy and produces goitrous hyperthyroidism.

If the maternal thyroid functions are in the normal range, amniotic fluid sampling for fetal TSH, FT3 and FT4 levels should be done to detect fetal hypothyroidism. If fetal hypothyroidism is diagnosed, intra-amniotic thyroxine injections (200-600 µg/week) are administered, and serial amniocenteses or preferentially cordocenteses are performed to follow the changes in the fetal thyroid metabolism.<sup>[3]</sup>

Lately non-invasive techniques can be tried in cases of non-obstructing FG by just monitoring thyroid size on USG and titration of maternal thyroxine dose in case of maternal hypothyroidism.

To conclude, FG is a condition which can be noticed on routine ultrasonography, if carefully done. A high index of suspicion is required. Its diagnosis is important, as it is one of the treatable cause for mental and motor retardation.

Fetal hypothyroidism should be thoroughly evaluated, diagnosed and immediately treated. Although in utero options for fetal hypothyroidism management are available, emphasis should be laid on non-invasive procedures with newer and better resolution techniques in ultrasonography, which are less harmful.

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