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Fetal goiter diagnosed on routine ultrasonographic assessment – case report

Wole u płodu wykryte w trakcie przesiewowych badań ultrasonograficznych – opis przypadku

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Keywords

fetal goiter, hypothyroidism, l-thyroxine treatment

Słowa kluczowe

wole płodowe, hipotyreoza, leczenie l-tyroksyną

Summary

Fetal goiter is a rare condition usually caused by maternal Graves' disease and transplacental passage of thyroid stimulating antibodies or treatment with antithyroid drugs, lithium, amiodarone or Lugol's solution. Occasionally fetal goiter may be caused by congenital disorders in thyroid hormone synthesis. The prompt diagnosis and treatment is needed because fetal goiter is always accompanied by fetal thyroid dysfunction: hyper- or hypothyroidism – impairment of intellectual and somatic development may be the final consequence of the both. Here we present the case of fetal goiter which was diagnosed on routine ultrasonographic assessment on the 23rd week of gestation. The mother, 20-year old healthy woman was euthyroid and negative for anti-TSH receptor antibodies. She did not take any medicines except for prenatal multivitamins containing 200 μg of iodine per tablet. The analysis of parenteral data and TSH, fT4, fT3 concentrations from umbilical cord sampling led to the diagnosis of hypothyroid fetal goiter probably dyshormonogenetic. Treatment with l-thyroxine given intraamniotically and into umbilical vein was successfully undertaken. The patient is still under observation.

Streszczenie

Wole u płodu jest rzadką patologią, najczęściej spowodowaną chorobą Gravesa i Basedowa u matki i przechodzeniem przez łożysko przeciwciał stymulujących receptor TSH lub przyjmowaniem przez ciężarną leków: tyreostatyków, węglanu litu, amiodaronu lub płynu Lugola. Rzadszą przyczyną jest choroba dziecka – genetycznie uwarunkowane zaburzenia biosyntezy hormonów tarczycy, np. defekt syntezy peroksydazy tarczycowej lub tyreoglobuliny. Wole u płodu jest ważnym objawem, ponieważ zawsze towarzyszy mu dysfunkcja tarczycy: nadczynność lub niedoczynność, wiodące do zaburzeń rozwoju somatycznego oraz umysłowego dziecka. Przedstawiamy przypadek wykrycia wola u płodu w przesiewowym badaniu ultrasonograficznym wykonanym w 23. tygodniu ciąży. U 20-letniej matki, dotychczas zdrowej, nie stwierdzono patologii tarczycy, nie przyjmowała również żadnych leków poza preparatami wielowitaminowymi zawierającymi 200 μg jodu przeznaczonymi dla kobiet ciężarnych. Wykonano kordocentezę i na podstawie badania stężenia TSH, fT4 i fT3 w krwi pępowinowej oraz analizy matczynych czynników ryzyka rozpoznano u dziecka hipotyreozę i wole prawdopodobnie dyshormonogenetyczne. Podjęto leczenie l-tyroksyną, podając lek początkowo wyłącznie doowodniowo, później także do żyły pępowinowej. Uzyskano zmniejszenie nasilenia niedoczynności tarczycy i wielkości wola. Ciężarna pozostaje w obserwacji.

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INTRODUCTION

Fetal goiter is very rare pathology, which always indicates fetal thyroid dysfunction. Thyroid hormones excess, and particularly thyroxine deficiency during intrauterine life interferes with the child's development, particularly the brain. We would like to present a case of fetal goiter diagnosed on routine ultrasonographic assessment (USG) carried out in at 23rd week of gestation.

CASE REPORT

The ultrasound imaging, performed on a 20-year old healthy woman at the 23rd week of her first gestation, revealed the presence of fetal goiter. The patient was referred to the Gynecological and Prenatal Ultrasound Diagnostic Unit of Gynaecology and Obstetrics Clinic CMKP (Centre of Postgraduate Medical Education) in Bielański Hospital in Warsaw, where the presence of extensively vascularised fetal goiter with the features of trachea compression was confirmed, with an accompanying cardiomegaly (fig. 1A-C) (1).

Performed examinations of the pregnant woman and the child's father showed normal thyroid hormone function, negative thyroid antibodies: anti-thyroperoxidase (anti-TPO) and anti-thyroglobulin (anti-Tg), and thyrotrophin receptor antibodies (TRAb), and absence of goiter.

The cordocentesis was performed, and on the basis of concentration of TSH, fT4 and fT3 in the cord blood, the primary hypothyroidism of the fetus was diagnosed. Table 1 presents the preliminary results of laboratory tests of the mother and child (2-4).

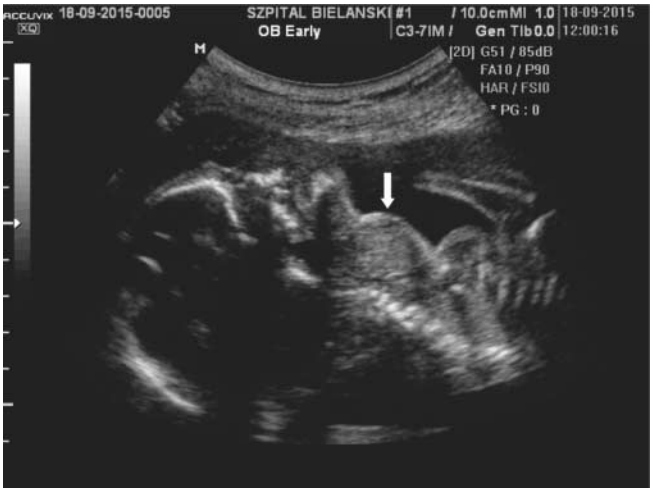

Fig. 1A. Fetal goiter (marked with a white arrow), 23rd week of gestation.

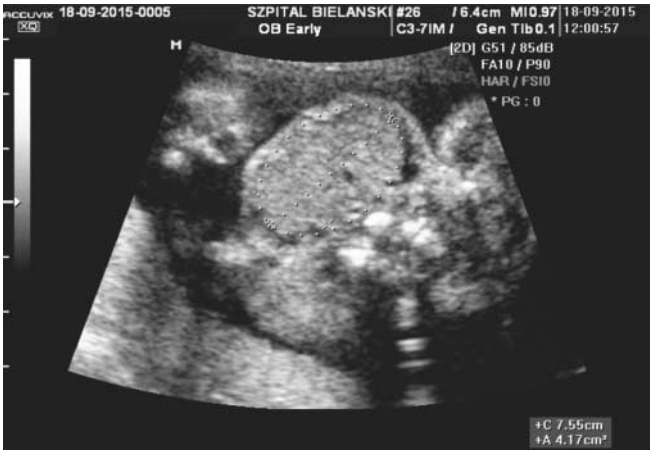

Fig. 1B. Fetal goiter in cross-section, features of tracheal stenosis, 23rd week of gestation. The cross sectional diameter of goiter – 2.75 cm (n. up to 1.74 cm), the cross-sectional circumference of goiter – 7.55 cm (n. up to 4.75 cm), the surface area of goiter – 4.17 cm^2 (n. up to 1.65 cm^2). Normal sizes of the fetal thyroid, depending on the gestational age, are shown as 97 percentile (1).

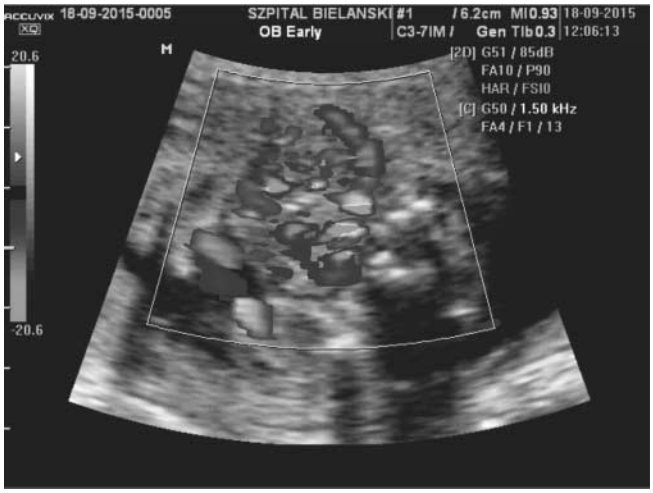

Fig. 1C. Fetal goiter in cross section: extensive vascularization in colour Doppler examination, 23rd week of gestation.

Table 1. Preliminary results of laboratory tests of the mother and child (2-4).

Tests	Mother	Child
TSH mIU/ml	2.3 (n. 0.05-3.4[2])	> 1000 (n. 6.8 ± 2.93[3])
fT4 pmol/l	11.5 (n. 10.46-16.67[2])	2.0 (n. 16.5 ± 5.3[3])
fT3 pg/ml	3.6 (n. 3.29-5.45[2])	0.87 (n. 0.2-0.5[4])
aTPO IU/ml	< 35 (n. < 60)	< 35
aTg IU/ml	< 25 (n. < 60)	< 25
TRAb IU/l	0.42 (n. < 1.8)	0.48
Tg ng/ml	-(n. 0-55)	> 1500
NT-proBNP ng/ml	-(n. 0.0-125)	1247.0

A treatment was initiated, which initially consisted in intra-amniotic administration of two doses of l-thyroxine: 500 μg and 250 μg in 14 days' interval. Each time, before the drug administration, blood was sampled from the umbilical vein in order to measure the level of TSH, fT4 and fT3. Because of absence of a satisfactory contraction of the goiter, and persistence of high levels of TSH in the cord blood, further 3 treatments were performed every week which consisted in administration of 20 mcg of l-thyroxine into umbilical vein and 230 mcg of l-thyroxine intraamniotically. As a result of those proceedings the laboratory tests results of the child performed at the 31st week of gestation were as follows: TSH – 61.8 mIU/ml (n. 8.0 ± 5.12 mIU/ml), fT4 – 9.8 pmol/l (n. 19.3 ± 4.3 pmol/l) (3), fT3 – 3.08 pmol/l (n. to 1.8 pmol/l) (4), and in the ultrasound examination the cross-sectional diameter of goiter was 3.1 cm (n. to 2.26 cm) (1).

The persistence of cardiomegaly was observed, but at the same time the regular heart action and normal intrauterine fetal growth were noted. There were no complications in regard to treatment, nor any significant changes in the concentrations of maternal TSH, fT4 and fT3.

DISCUSSION

Fetal goiter is very rare condition and its causes are usually attributable to the mother:

- maternal Graves' disease and trans-placental passage of TSH- receptor stimulating antibodies which results in fetal goiter and hyperthyroidism,
- medications and preparations taken by pregnant woman, which may cause fetal goiter and hypothyroidism, i.e.: antithyroid drugs, lithium salts, amiodarone, Lugol's solution, iodine contrast agents,
- severe deficiency of iodine in endemic areas, where daily supply of this element is < 20 μg, which causes hypothyroidism in the mother and child. Nowadays in Poland the iodine supply in adults is 150 micrograms per day.

Fetal goiter is much less associated with a thyroid disease of the child involving a genetically conditioned disorders in transportation and organification of iodine, thyroglobulin synthesis or iodotyrosine deiodination.

In the presented case, the mother did not suffer from thyroid diseases, she denied taking any drugs that affect the thyroid gland functioning, did not have any diagnostic tests with the use of contrast agents. During her pregnancy she only took multivitamins containing 200 μg of iodine per tablet. The child therefore was initially diagnosed with dyshormonogenetic goiter and hypothyroidism, but not associated with the disorder of thyroglobulin synthesis, because its concentration in the cord blood was very high. Given the potentially disastrous effects of hypothyroidism for the development of the child, an immediate treatment was initiated consisting in intra-amniotic administration of l-thyroxine. In the medical literature, there are descriptions of such a procedures implemented mainly in cases where the fetal goiter and hypothyroidism was caused by the treatment of pregnant woman with antithyroid drugs or Lugol's solution, and occasionally dyshormonogenezis in the child (5-8). Due to the rarity of this pathology there are no guidelines for dosing l-thyroxine, optimal frequency of dosing or the way of its administration. Ribault et al. published multi-centre results of treatments with intra-amniotic injections of l-thyroxine in 12 cases of fetal dyshormonogenetic hypothyreosis; they administered from 1 to 6 injections in doses of 150 to 800 μg per injection in 1-4 weeks intervals. In all newborns the TSH level after their birth was abnormal and ranged from 38 to 450 mIU/ml, and only in 2 of 12 children the levels of T4 fell within the normal range (9). Despite hypothyroidism during intrauterine life, the development of children evaluated at the school age was normal. Agrawal et al. described the treatment of one such case consisting in intra-amniotic administration – between the 29th and 36th week of gestation – of 3 doses of l-triiodothyronine: 60, 60 and 120 μg, and 2 doses of l-thyroxine of 150 and 300 μg. The child had moderate hypothyroidism and a goiter after birth (10). In the case presented by us, we decided on concurrent intravenous and intra-amniotic administration of l-thyroxine, following lack of satisfactory results after intra-amniotic administration of the first 2 doses of the drug and probable compression of the oesophagus that inhibited the free swallowing of amniotic fluid. We concidered that the administration of the drug into the umbilical vein will lower the TSH level and reduce the size of the goiter faster, which consequently shall enable free swallowing. We found out that such method of treatment was significantly more effective and safe at the same time. The observed cardiomegaly, accompanied by biochemical features of heart failure expressed by a high concentration of N-terminal type B natriuretic propeptide – NT-proBNP, was probably linked to both the hyperkinetic circulation caused by extensive vascularization of goiter, and thyroid hormone deficiency. The presented case has a preliminary report character, however it shows the safety and efficacy of fetal dyshormonogenetic goiter treatment through the concurrent intravenous and intra-amniotic administration of l-thyroxine.

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