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## Fetal neck tumor – differentiation from thyroid goiter

## Guz szyi u płodu wymagający różnicowania z wolem

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**S u m m a r y**

Fetal anterior neck tumor is a rare condition and differential diagnosis includes goiter, teratoma, thyroglossal duct cyst, cystic hygroma, branchial cleft cyst, lymphangioma/hemangioma and neuroblastoma. The case of fetal anterior cervical tumor was diagnosed on routine ultrasonographic assessment on the 22<sup>nd</sup> week of gestation. Tumor was mixed solid and cystic with moderate vascularization, exhibited rapid growth causing neck hyperextension, esophageal compression and polyhydramnion. The both parents were healthy, their serum thyrotropin, thyroid hormones results and thyroid ultrasonography were normal, antithyroid antibodies negative. There was no history of excessive iodine intake by the mother. Fetal blood sampling was performed and normal fetal thyroid function was documented. Thus the diagnosis of fetal goiter was rejected. The tumor course and its ultrasonographic presentation indicated cervical teratoma. Pregnancy was terminated because of the poor prognosis for the fetus and diagnosis of teratoma was confirmed by pathological examination.

**S t r e s z c z e n i e**

Prezentujemy przypadek płodu z guzem szyi wykrytym w 22. tygodniu ciąży w trakcie rutynowego badania ultrasonograficznego. Guz wymagał różnicowania pomiędzy wolem, potwornikiem, nerwiakiem niedojrzałym, torbielą przewodu tarczycowo-językowego, torbielakiem surowiczym i naczyniakiem. Badanie ultrasonograficzne wykazało litotorbielowaty charakter guza, umiarkowane unaczynienie, gwałtowny wzrost, odgięcie głowy, ucisk przełyku, a w konsekwencji wielowodzie. Obydwoje rodzice byli zdrowi, nie przyjmowali żadnych leków, ocenione stężenia TSH, fT4 i fT3 były prawidłowe, przeciwciała tarczycowe negatywne, obraz ultrasonograficzny tarczycy nie wykazywał nieprawidłowości. Wykonano kordocentezę i stwierdzono prawidłową czynność hormonalną tarczycy płodu, co ostatecznie wykluczyło obecność wola. Obserwowane gwałtowne powiększanie się guza oraz jego obraz ultrasonograficzny nasuwały podejrzenie potwornika. Nastąpiła terminacja ciąży, a badanie histopatologiczne potwierdziło obecność potwornika.

**INTRODUCTION**

Ultrasonographic (US) assessment of the fetus is routinely recommended between 11-14, 18-23 and 30-33 week of gestation. We present a case of a fetus with cervical tumor diagnosed in the 22<sup>nd</sup> week of gestation which was differentiated from thyroid goiter.

**CASE STUDY**

Fetal tumor localized in left anterior part of the neck was diagnosed on routine US assessment. Its size at the time of diagnosis was 54 x 50 x 47 mm, it

was mostly solid with small cystic areas, with moderate vascularization and no calcifications. Normal fetal thyroid was not visible (fig. 1-3). Both parents were healthy with no family nor personal history of thyroid diseases, they had no palpable goiter and they did not take any medications. The mother had no history of excessive iodine intake nor medical procedures with iodine contrast media prior to the pregnancy. Parental serum thyrotropin (TSH), free T3 (fT3), free T4 (fT4) were within normal range, thyroid-peroxidase antibodies (TPOAbs), thyroglobulin antibodies (TGAb) and

TSH receptor antibodies (TRAbs) were negative and their thyroids US performance was normal. Cordocentesis was performed to evaluate fetal thyroid function and exclude fetal goiter. The results of the fetal blood examination were within the normal range for gestational age: TSH 8.4 mU/L (normal: 2.0-9.5 mU/L), fT4 – 8.38 pmol/l (normal: 3.0-9.0 pmol/L), fT3 < 1 pmol/L (normal: < 1.0 pmol/L) (1), TPOAb – 15.6 IU/mL (normal: < 60 IU/mL), TgAb – < 20 IU/mL (normal: < 60 IU/mL), TRAb – 0.8 IU/mL (normal: < 1.8 IU/mL). In the next US examination performed one week later rapid tumor growth was observed: its size enlarged to 68 x 62 x 56 mm causing significant hyperextension of the fetal neck and polyhydramnios resulting from esophageal obstruction. Fetal cervical teratoma was suspected and pregnancy was terminated because of the poor prognosis for the fetus. The pathological examination confirmed initial diagnosis: the tumor consisted of cartilaginous, intestinal and neural tissue, it adhered strictly to the tissues of the oral cavity, compressed the throat and larynx causing lesion of the jaw.



**Fig. 1.** Fetal examination on 2-dimensional ultrasonography. Giant anterior neck tumor with head hyperextension and polyhydramnion.



**Fig. 2.** Fetal neck tumor on 2-dimensional ultrasonography.



**Fig. 3.** Fetal examination on 3-dimensional ultrasonography. Giant anterior neck tumor causing head deviation.

**DISCUSSION**

Fetal anterior neck tumor is a rare condition and differential diagnosis includes goiter, teratoma, thyroglossal duct cyst, cystic hygroma, branchial cleft cyst, lymphangioma/hemangioma and neuroblastoma. We considered a goiter first of all because it can be quite easily diagnosed and often successfully treated. Fetal goiter is always accompanied by fetal thyroid dysfunction: hyper- or hypothyroidism (2). Fetal hyperthyroid state is commonly related to maternal Graves’ disease and transplacental passage of TSH receptor stimulating antibodies (3). The other extremely rare causes of fetal hyperthyroidism include activating mutations of the stimulatory G protein in McCune-Albright syndrome and activating mutations of the thyrotropin (TSH) receptor (4).

Fetal hypothyroid goiter is usually related to maternal factors: antithyroid treatment for hyperthyroidism, iodine deficiency (endemic goiter) or iodine excess (dietary excessive intake, Lugol’s or antiseptic solutions, amiodaron, iodine containing contrast media) and medications affecting thyroid function such as lithium (5). The very rare causes are congenital disorders in thyroid hormone synthesis, especially defective synthesis and secretion of thyroglobulin and defective iodine organification frequently caused by mutations in TPO gene (6).

On US goiter presents as a anterior neck mass which is solid, homogenous and maintains characteristic lobular shape. Central hypervascularization of the goiter can be seen on Doppler examination in fetal hyperthyroidism and peripheral hypervascularization in fetal hypothyroidism (2, 7). Esophagus and trachea can be compressed causing polyhydramnion and airway compromise at birth.

The other rare US signs of fetal thyroid dysfunction include abnormalities of bone maturation and heart rate, cardiac failure, hydrops and intrauterine growth restriction.

Fetal goiter and other signs of fetal thyroid disorders can diminish or subside after proper treatment including intraamniotic L-thyroxine injections for fetal hypothyroidism (8-11).

The parental history and fetal presentation on US made the diagnosis of fetal goiter very unlikely and this suspicion was definitely rejected after obtaining normal fetal TSH and free hormone results.

The possibility of the thyroglossal duct cyst, cystic hygroma, branchial cleft cyst, lymphangioma/hemangioma was also unlikely because the nature of the observed tumor was predominantly solid.

Diagnosis of cervical teratoma seemed the most probable because the tumor exhibited rapid growth to massive size, caused typically head hyperextension and deviation, jaw and ear distortion and esophageal obstruction. As majority of teratomas the observed tumor was mixed solid and cystic and exhibited moderate hypervascularization. The pathognomonic signs of teratomas observed in 50% of cases – macrocalcifications were unfortunately absent. The pathological result confirmed the diagnosis: tumor was composed of ectodermal (neural tissue), mesoderm (cartilage) and endoderm elements (intestinal tissue).

The neck and head teratomas are of poor prognosis: they can be malignant, may cause high-output cardiac failure and hydrops because of vascular shunts within the tumor, head hyperextension precludes vaginal delivery, and airway compression are the often cause of hypoxia, acidosis and brain anoxic injuries after delivery. Mortality for neck and head teratoma is 80-100% (12, 13). In some cases of massive cervical tumors causing tracheal compression EXIT procedures are offered in tertiary care hospitals. EXIT procedure (ex utero intrapartum treatment) consists in performing cesarean section and delivering baby partially; while keeping it attached by umbilical cord to the placenta and maintaining placental support tracheostomy, intubation or resection of the neck mass is undertaken.

## CONCLUSIONS

Fetal neck tumor observed during routine ultrasonographic assessment was diagnosed as teratoma and differentiated from goiter basing on parental history, fetal US presentation and normal thyrotropin and thyroid hormone results obtained from fetal blood sampling.

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