

The chorionic bump associated with acrania – case report

Współwystępowanie guza kosmówki i beczaszki – opis przypadku

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Abstract

The chorionic bump is a rare abnormality of the gestational sac, presenting as a convex bulge from the choriodecidual surface into the sac, correlated with poor prognosis for the pregnancy.

We report a case of a 36-year-old pregnant woman, with a history of spontaneous abortion, who presented for an early scan at 6 weeks and 4 days of gestation. The pregnancy was spontaneous and unplanned. The patient conceived in less than 3 months after discontinuing oral contraceptives. No folic acid was taken before or in the pregnancy. An ultrasound scan revealed a chorionic bump with a hypoechoic center and echogenic border, measuring 18.3 x 14.7 x 21.9 mm. No motion within the chorionic bump was detected upon color and power Doppler examination. The second scan was performed a week later, at 7+4 wks. The chorionic bump had not changed in terms of size and sonographic appearance. An acranial fetus of CRL 45.5 mm was diagnosed at 11+2 wks. The concentration of free β -hCG was 17.2 IU/L, corresponding to 0.37 MoM and PAPP-A levels were 1.31 IU/L, corresponding to 0.82 MoM. After counseling the patient opted for termination of pregnancy.

Very few cases of chorionic bumps have been described so far and, to the best of our knowledge, its coexistence with neural tube defects has been reported for the first time. We postulate a possibility of an underlying pathological mechanism for such coexistence. The chorionic bump is a focal convex bulge with irregular borders, protruding from the choriodecidual surface into the gestational sac and with different degrees of echogenicity, usually a hypoechoic middle and echogenic border. The chorionic bump might represent the following: a hematoma, an area of hemorrhage, a non-embryonic gestation, or a demise of an embryo in a twin pregnancy. The presence of the bump is associated with a four-fold increase in the spontaneous abortion rate as compared with the general population.

Decreased folate levels increase the incidence of neural tube defects. Oxidative stress resulting from folic acid deficiency may be responsible for neural tube defects through impairment of factors inhibiting apoptosis in the neuroepithelium. Fetuses with neural tube defects are at an increased risk of being aborted spontaneously. Furthermore, women who deliver children with neural tube defects frequently have a history of miscarriage. Our patient did not take any folic acid and also had a history of spontaneous miscarriage.

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In the case we herein presented, the coexistence of acrania and placental pathology could be attributed to folate deficiency. Such coexistence is described for the first time and could be accidental, but there is possible theoretical association between these two pathologies.

Key words: **chorionic bump / acrania / first trimester scan / folate deficiency /**

Streszczenie

Guz kosmówki jest rzadką patologią jaja płodowego związaną z niekorzystnym rokowaniem dla rozwoju ciąży. U badaniu ultrasonograficznym daje obraz guzowatej zmiany uwypuklającej się z powierzchni kosmówki do wewnątrz jaja płodowego.

Autorzy opisują przypadek 36-letniej kobiety, z jednym poronieniem w wywiadzie, która zgłosiła się na badanie USG w 6+4 t.c. Ciąża nie była planowana i pacjentka nie stosowała suplementacji kwasem foliowym.

W czasie badania USG uwidoczniono guz kosmówki o wym. 18.3 x 14.7 x 21.9 mm, o zwiększonej echogeniczności i z hipoechogeniczną częścią centralną. Nie uwidoczniono przepływu w badaniu dopplerowskim. Kolejne badanie wykonano w 7+4 t.c. Obraz ultrasonograficzny guza nie uległ zmianie. W 11+2 t.c., przy CRL=45,5 mm rozpoznano płód beczaszkowy. Poziom β -hCG wynosił 17,2 IU/l (0,37 MoM) a PAPP-A = 1.31 IU/l (0.82 MoM). Pacjentka podjęła decyzję o terminacji ciąży.

Do tej pory opisano bardzo niewiele przypadków guza kosmówki, a jego współistnienie z płodem beczaszkowym opisujemy po raz pierwszy. Guz kosmówki może być objawem krwaka kosmówki lub obumarcia jednego z zarodków w ciąży bliźniaczej. Jego obecność wiąże się z czterokrotnym wzrostem ryzyka poronienia.

Niedobór kwasu foliowego wiąże się ze zwiększonym ryzykiem otwartych wad ośrodkowego układu nerwowego. Płody z takimi wadami obciążone są większym ryzykiem samoistnego poronienia. Kobiety, które rodzą dzieci z otwartymi wadami OUN, częściej mają w wywiadzie poronienia samoistne. W opisywanym przypadku pacjentka nie stosowała kwasu foliowego i miała poronienie w wywiadzie.

Współistnienie tych dwóch patologii może być przypadkowe ale może istnieć wspólny mechanizm związany z niedoborem kwasu foliowego prowadzący do tych dwóch patologii.

Słowa kluczowe: **guz kosmówki / beczaszkiwiec / badania ultrasonograficzne / pierwszy trymestr / niedobór kwasu foliowego /**

Case report

A 36-year-old woman with a history of spontaneous abortion presented for the first trimester pregnancy viability scan. The pregnancy was unplanned and spontaneous. The patient conceived in less than 3 months after discontinuing oral contraceptives. The patient denied periconceptional supplementation with folic acid. She was not a smoker and weighed 60 kg. A transvaginal ultrasound scan (Voluson 730 PRO, GE) revealed an intrauterine gestational sac (GS) with a live fetus with a crown-rump length (CRL) of 8 mm and fetal heart rate (FHR) of 162 beats per minute. The CRL measurement was consistent with the gestational age (GA) of 6 weeks and 4 days. Additional color and power Doppler sonography showed a chorionic bump with a hypoechoic center and echogenic borders, measuring 18.3 x 14.7 x 21.9 mm (Figure 1), but no motion was detected within it. The β -hCG level measured at that time was 36.1 IU/L. The second scan was performed a week later at the GA of 7⁺⁴ weeks and revealed an unchanged chorionic bump (CRL 12.4 mm, FHR 152 bpm). The CRL, GS, yolk sac and amniotic sac measurements showed a consistent growth between the two examinations. Another sonography at the GA of 11⁺² weeks revealed an acranial fetus with the CRL of 45.5 mm. (Figure 2). Free β -hCG and PAPP-A levels were 17.2 IU/L (0.37 MoM) and 1.31 IU/L (0.82 MoM),

respectively. The sonographic data and pictures were analyzed using Astraia Software. The patient was counseled and opted for termination of the pregnancy.

Discussion

The chorionic bump is a focal convex bulge with irregular borders protruding from the choriodecidual surface into the gestational sac and with different degrees of echogenicity, usually with a hypoechoic middle and echogenic border [1-3]. It was first reported by Harris et al. (2006), who prospectively noted 15 cases with the chorionic bump in a total of 2178 patients (prevalence of 0.7%) [1]. The etiology remains unclear but it has been hypothesized that the chorionic bump might represent a non-embryonic gestation or a demise of a single embryo in a twin pregnancy. Most authors, however, agree that it more likely represents a hematoma or a focal hemorrhage [1,2]. The latter hypothesis was confirmed by a report of histopathological findings in an abortus with a chorionic bump revealing large foci of hemorrhage with erythrocytes between the chorionic villi [3]. Coagulation disorders may be responsible for decidual vascular changes and cause thrombosis in the placental vessels [4].

The presence of a chorionic bump is associated with a four-fold increase in the incidence of spontaneous abortion

as compared with the general population [1]. The association between the chorionic bump and acrania, two rare anomalies which coexisted in the case we report, remains unclear. The pathogenesis of acrania is multifactorial and teratogenic drug use, genetic syndromes, and amniotic band syndrome have been implicated. Many pregnancies affected by malformation-producing genetic syndromes or chromosomal defects tend to abort spontaneously and some of them may begin to miscarry, but the miscarriage is not complete. In the reported case of a major fetal malformation, a hemorrhage may have started, resulting in the formation of a chorionic bump, and then stopped.

Many features of an early ultrasound examination regarding the irregularity of the gestational sac, large, calcified or irregular yolk sac, low heartbeat rate or large CRL to GS discrepancies, have been associated with poor pregnancy prognosis [3, 5-7].

Acrania is typically diagnosed at nuchal scan between 11+0 and 13+6 wks. [8]. Its associations with chorionic bump remain unclear. There is a wide spectrum of factors connected with acrania, from teratogens and genetic syndromes to amniotic bands. Many pregnancies affected by genetic syndromes or chromosomal defects tend to abort spontaneously. However, a lot of the pregnancies with chromosomal, genetic or structural malformations tend to continue till birth. Possibly, a lot of these pregnancies begin to miscarry due to the malformation that occurred, however they do not miscarry fully. Thus, the miscarriage starts as a hemorrhage that is not strong enough to abort the pregnancy. In our case of a fetal abnormality, it might have been that the bleeding started, resulting in a chorionic bump, and then halted.

It is widely recognized that folic acid supplementation leads to a decrease in the incidence of open neural tube defects [9]. The placenta concentrates folates, thus increasing their supply to the fetus. Folate levels are higher in the intervillous space of the placenta than in the sera of newborns (1.3-fold) and mothers (4.5-fold). A significant positive correlation was also found between serum folate levels of mothers, newborns and placentas [10]. Women with low folate levels are at a significantly increased risk for spontaneous abortion, what could be attributed to early vascular defects related to folate deficiency [11]. There is some evidence that folic acid may play a crucial role in extravillous trophoblastic invasion angiogenesis and vasculogenesis, and its decreased level is associated with placental apoptosis [12, 13].

In our case, low free β -hCG (0.37 MoM) and PAPP-A (0.82 MoM) levels might be indicative of impaired placental function. Large areas of hemorrhage with erythrocytes between villus formations found in an abortus with a chorionic bump could be attributed to folate deficiency, leading to abnormal extravillous trophoblastic invasion angiogenesis and vasculogenesis [3, 11-13].

Decreased folate levels increase the incidence of neural tube defects. Oxidative stress resulting from folic acid deficiency may be responsible for neural tube defects through impairment of factors inhibiting apoptosis in the neuroepithelium [14]. Fetuses with neural tube defects are at increased risk for being aborted spontaneously [15]. Furthermore, women who give birth to children with neural tube defects frequently have a history of miscarriage [16]. Our patient had no periconceptional supplementation with folic acid and also had a history of a spontaneous miscarriage.



Figure 1. Chorionic bump at 6+4 wks.



Figure 2. Acranius at 11+2 wks.

The concomitant occurrence of acrania and placental pathology we reported could be attributed to folate deficiency. To the best of our knowledge, it has been the first such case described in the literature. It cannot be excluded that the reported coexistence of these two rare pathological conditions was accidental, but there is a possible theoretical explanation we wish to share. It may be speculated that periconceptional folate deficiency led to a fetal malformation that brought about an arrested spontaneous abortion, with the resulting focal hemorrhage and ultimately the chorionic bump.

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