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Could chorionic bump represent an early ultrasound marker of Klinefelter syndrome? A case report of our experience

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INTRODUCTION

Chorionic Bump (CB) is a rare first-trimester sonographic feature described as an irregular, convex bulge from the choriodecidual surface into the first-trimester gestational sac (GS). Reported incidence is 1.5-7 per 1000 cases [1]. Etiopathology is not clear. Although there is contradictory evidence about pregnancy outcomes, the presence of CB on early first trimester ultrasound increases three-fold the risk of miscarriage. Thus, it is considered as a marker of long-term compli-

ABSTRACT

Background. Chorionic Bump (CB) is a rare first-trimester sonographic feature described as an irregular, convex bulge from the choriodecidual surface into the first-trimester gestational sac (GS), with an incidence of 1.5-7 per 1,000 cases. Etiopathology and related pregnancy outcomes are not clear.

Case presentation. We present a case of a 37-year-old female with a transvaginal sonographic finding of a CB associated with a 47, XXY karyotype (Klinefelter syndrome) previously detected by non-invasive prenatal testing (NIPT) and confirmed by amniocentesis and definitive genetic examination at autopsy.

Conclusions. To the best of our knowledge, this is the first case of CB associated to Klinefelter syndrome.

cations [5], even if in some cases it seems to be an incidental finding with no clinical significance [6]. We present a case of a 37-year-old female with a transvaginal sonographic finding of a CB associated with Klinefelter syndrome, a previously unreported finding.

CASE PRESENTATION

A 37-year-old female, nullipara, came to our observation for sonographic evaluation of pregnancy

viability. Her gestational age was of 6 weeks and 6 days. She was a nonsmoker and had no history of gynaecological pathologies; she only reported breast fibroadenomas removed five years earlier by surgery. Transvaginal ultrasound examination with an endocavitary probe revealed a single gestational sac in the uterine cavity with an embryo showing cardiac activity. The crown-rump length (CRL) of 8.6 mm corresponded to an estimated gestational age of 6 weeks and 6 days. A regular yolk sac was visualized (**Figure 1**). Additionally, a chorionic bump of 15.5 × 13.9 mm was identified within the gestational sac (**Figures 1,2**). The chorionic bump had a spherical shape, with a hypoechoic centre, without signs of movement of the internal echoes or vascularization detected at colour Doppler examination; it appeared as a continuous image with the chorio-decidual surface (**Figure 2**).

At 12 weeks, the patient performed another ultrasound evaluation. The embryonic morphology was regular, and the thickness of the nuchal translucency measured 1.2 mm. The CB was still present, and measures were equal to the previous ultrasound examination.

The pregnant woman decided to undergo aneuploidy screening via cell-free DNA in maternal blood (NIPT-test), which resulted in high risk for 47, XXY karyotype, with a sensitivity of 98.9%. For this reason, after informed consent was acquired, the patient decided to undergo amniocentesis at 15 weeks and three days of gestational age. Amniotic fluid sample was sent to perform karyotype test. After a few days, a quantitative fluorescence polymerase chain reaction (QF-PCR) confirmed a 47, XXY karyotype (Klinefelter syndrome).

After adequate genetic counselling, the patient underwent therapeutic abortion at 16 weeks and 2 days of gestational age, by oral misoprostol induction.



Figure 1. Crown-rump length (CRL) of 86 mm, corresponding to a gestational age of 6 weeks and 6 days and chorionic bump, inside a regular yolk sac.



Figure 2. Chorionic bump of 15.5 × 13.9 mm identified within the gestational sac.

After the completion of the abortive birth, we sent the foetus for autopsy and definitive genetic examination. The definitive chromosomal arrangement confirmed the diagnosis of 47 XXY karyotype; the definitive histological analysis of the placenta described a “placental tissue consisting of immature and mature intermediate villi, with regular trophoblastic cover, vascularized with anucleate erythrocytes (compatible with the gestational age), some of them with a degenerative involution”.

DISCUSSION

Chorionic bump (CB) is a rare first-trimester sonographic feature. Etiopathology of CB is not clear. Several studies suggest that it represents a placental hematoma [2], other theories consider it as a non-embryonic gestation, or an anembryonic early miscarriage in a twin pregnancy [3]. However, histological examination of the conceptus after spontaneous miscarriage reported that CB and subchorionic hematoma are two different entities. Indeed, CB is an echolucent lesion arising within the chorion immediately under the chorionic membrane, whereas subchorionic hematoma is the presence of echolucent fluid represented by blood between the uterine wall and the chorion or within the endometrial cavity [4]. Regarding pregnancy outcomes, there is contradictory evidence. Several studies suggest that a CB detected during the 11- to 13-week scan could be considered a transient, incidental finding, with probably no clinical significance [7]. However, other studies demonstrated that the detection of CB on first trimester, increases three-fold the risk of miscarriage and vaginal bleeding. Indeed, it is considered as a marker of long-term complications, and it is associated with poor maternal and foetal outcomes

[5]. The size or localization of the bump seems not to have a correlation with pregnancy outcomes [6]. To the best of our knowledge, this is the first case of chorionic bump associated to Klinefelter syndrome. Coexistence of CB and Klinefelter syndrome could be accidental, but an association with foetal aneuploidies cannot be excluded. Indeed, a retrospective study of Wax *et al.* demonstrated that a sonographically isolated chorionic bump confers a further increase in the likelihood of a chromosomal abnormality in pregnancies already at increased aneuploidy risk [8]. Moreover, Wax *et al.* also provided an alternative aetiology for CB associated with aneuploidy. Indeed, according to them, CB in some abnormal pregnancies may be the US manifestation of focal placental pathology such as villus hydrops and stromal oedema [9]. Furthermore, it is important to remember that the presence of advanced maternal age itself (*i.e.*, more than 35 years old) could be considered a risk factor for congenital malformations [10] and having pre-existing insulin-independent Diabetes Mellitus has been shown to correlate with more foetal anomalies seen on ultrasound [11].

CONCLUSIONS

In conclusion, this is the first case of chorionic bump associated to Klinefelter syndrome. Even if this finding might be accidental and its meaning is still controversial, it might be consistent with the hypothesis of an association between chorionic bump and aneuploidies that has already been advanced by other authors. For this reason, even if the pregnancy continues (83%) [12], in the second trimester an invasive prenatal diagnosis could be necessary to confirm the normality of the foetal karyotype or to exclude foetal aneuploidies.

COMPLIANCE WITH ETHICAL STANDARDS

Authors contributions

L.L.F., P.R., R.D., F.C.: Conceptualization, investigation, writing – original draft, writing – review & editing.

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Study registration

N/A.

Disclosure of interests

The authors declare that they have no conflict of interests.

Ethical approval

The case report was considered exempt from formal IRB approval.

Informed consent

Written informed consent allowing data collection for research purposes was collected from the patient at time of examination.

Data sharing

N/A.

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