Maternal Health, Neonatology, and Perinatology

CASE REPORT

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Diploid fetus with partially triploid placenta: case presentation and management strategy

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Abstract

Multiple placental cysts are a common finding in obstetric ultrasound imaging. Although they have benign differential diagnoses, such as hydropic degeneration of the placenta or placental mesenchymal dysplasia, it's important to consider significant pathologies, such as benign gestational trophoblastic disease or hydatidiform mole. A challenging issue in obstetrics is pregnancies with a placenta that has a bipartite texture. This means that one side of the placenta is normal, but the other side is full of cystic formations, and only one fetus is visualized. The main critical concern is the presence of a molar pregnancy because of its catastrophic consequences. Here, we report a rare case in which the gravid uterus had a normal diploid fetus but had a bipartite placenta, which was triploid in the hydropic part, revealing a unique genetic pattern.

Keywords Placental cyst, Bipartite placenta, Hydropic placenta, Twin molar pregnancy

Introduction

Multiple placental cysts are a common finding in obstetric ultrasound imaging. Although they have benign differential diagnoses, such as hydropic degeneration of the placenta or placental mesenchymal dysplasia, their significant pathologies, such as benign gestational trophoblastic disease or molar pregnancy, should always be considered [1, 2].

A challenging issue in obstetrics is pregnancies with a placenta with a bipartite texture. This means that one side of the placenta is normal, but the other has cystic formation, and just one fetus is visualized (Fig. 1). A possible explanation could be twin pregnancies, with one normal pregnancy in one sac and a complete molar cotwin in another. These pregnancies are at increased risk of serious complications. Although ultrasound imaging features such as identifying a lambda sign can help recognize this kind of pregnancy, they can be inconclusive in some cases. On the other hand, in any pregnancy with a bipartite placenta and only one fetus, placental mesenchymal dysplasia and also partial molar pregnancies should always be considered. Placental mesenchymal dysplasia is a rare, benign placental malformation identified with placentomegaly and grape-like vesicles, mimicking molar pregnancy on ultrasound imaging.

Prenatal recognition of placental mesenchymal dysplasia during early and late gestation could affect the future of pregnancy and avoid unnecessary pregnancy

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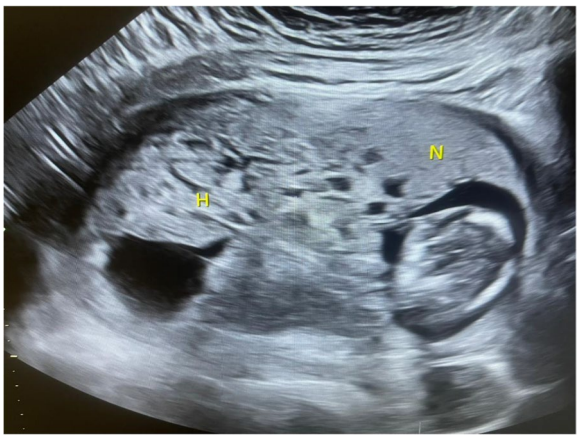

Fig. 1 placental ultrasound imaging in the first trimester revealed a bipartite placenta with two distinct zones: N (normal appearance) and H (hydropic appearance)

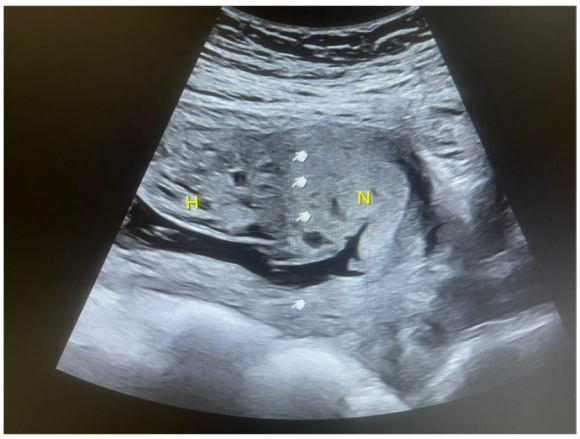

Fig. 2 Placental appearance in the 16th week of pregnancy. The normal part (N) was distinct from the hydropic part (H)

termination [3, 4]. Moreover, visualization of the abnormal fetus can lead to a partial molar pregnancy diagnosis, but it can still be challenging in some cases.

Here, we report a rare case in which the gravid uterus had a normal diploid fetus but had a bipartite placenta, which was triploid in the hydropic part, revealing a unique genetic pattern. Our management approach for this case can suggest a possible strategy that can be used as guidance in managing similar cases.

Case presentation

A 36-year-old primigravid woman with no abnormal medical, surgical, or drug history was referred to our clinic for performing her first-trimester screening ultrasound imaging in the 12th week of gestation. The placenta had a biphasic appearance during this examination (Fig. 1). There was a hypoechoic cystic appearance in the upper part of the placenta, but the lower part had a homogenous standard view. Due to the fetus' normal anatomical survey and nuchal translucency in the ultrasound imaging assessment, the first-trimester biochemistry screening test (β-hCG anPAPP-A) was suggested, revealing a low-risk pregnancy. β-hCG titer was 152,000 Miu/MI. At this stage, the differential diagnoses were:

1- Twin dichorionic pregnancy with a complete molar in one sac and one normal placenta and fetus in another. However, we could not reach a definite diagnosis since the ultrasound imaging did not show the lambda sign.
2- Partial molar pregnancy. Since the fetus had a normal anatomical survey, partial molar pregnancy was less likely as most of the fetuses in partial molar pregnancies are triploid and malformed.
3- Normal pregnancy with mesenchymal dysplasia of the placenta.

In the 16th week of pregnancy, she did another ultrasound imaging. The placenta still had the mentioned distinct pattern, and the fetus was normal (Fig. 2). After counseling and discussing differential diagnosis, the patient decided to undergo a genetic study to confirm the normal fetal karyotype for reaching a definite diagnosis and also ruling out molar pregnancy as a primary critical differential diagnosis. Chorionic villus sampling (CVS) from the placenta's hydropic part and amniocentesis were performed simultaneously. We did QF-PCR and standard GTG-banding karyotype on both samples. The result was a normal diploid fetus, and the hydropic part of the placenta was triploid.

Considering the live fetus with normal condition, the pregnancy continued under close observation. All of her routine laboratory tests were normal during pregnancy. β-hCG titer was 130,000–150,000 MIU/ML in three consecutive assessments. Two ultrasound fetal structural surveys revealed normal development in the 18th and 24th weeks of pregnancy. The fetus followed the normal growth curvature and gained appropriate weight during pregnancy. The pregnancy was terminated in the 38th week due to increased maternal blood pressure (160/90 mmHg) and 2+proteinuria in random urine analysis on the day of admission. She underwent a cesarean section because of fetal breech presentation after maternal stabilization by controlling blood pressure using Labetolol and administrating Mgso4. Lab tests, including liver function tests, platelet count, hemoglobin, and creatinine levels, showed average results.

Apgar scores of the newborn were 9 and 10 in the 1st and 5th minutes. The umbilical artery pH was 7.28,

and the birth weight was 2750 g (13% percentile due to Hadlock growth curvature). Macroscopically, the placenta had two distinct parts, discoid creamy-gray friable soft tissue measuring 60∗50∗20 mm, which was fused to the normal-appearing placenta. The patient refused to undergo a placental pathological examination. In the postpartum period, she received anti-hypertensive drugs for two days to control her blood pressure and intravenous MgSO4 as seizure prophylaxis. She was discharged after three days with a normal health condition. Her β-hCG levels in the three-month follow-up were also normal.

Discussion

Cystic formation in the confined area of the placenta, along with a normal appearing texture in the other parts, should raise concern about whether it is a twin pregnancy with one normal fetus and complete molar pregnancy in co-twin, partial molar pregnancy or placental mesenchymal dysplasia. Although in most of these pregnancies, the diagnosis can be straightforward according to ultrasound imaging features, in some borderline cases, reaching a definite diagnosis can be challenging, especially in the early stages of pregnancy.

The observation of a bipartite placenta with cystic areas beside a normal fetus may raise critical concerns regarding the pregnancy outcome. The primary differential diagnoses are: 1- dichorionic twin with a normal fetus in one sac and a complete mole in another, 2- dichorionic twin with a normal fetus accompanied by partial mole of the missed fetus, 3- mesenchymal dysplasia, and 4- very rare event of a normal fetus and confined placental partial molar pregnancy.

The primary critical diagnosis is the presence of complete molar pregnancy because of catastrophic consequences. The karyotype in complete molar pregnancy is 46,XX, but all the chromosomes are from the paternal origin. In the clinical setting, instead of a genetic study, the diagnosis is established by observing the pathognomonic appearance in ultrasound imaging, pathological examination, and gross visualization of the molar tissue. However, in cases such as pregnancies with normal fetuses and a cystic pattern of the placenta, a way to reach a definite diagnosis should be granted.

The prevalence of complete molar pregnancy is 1- 6%, higher than that of a partial mole [5]. On the other hand, partial moles come from the dispermic fertilization of a normal haploid oocyte that generally generates a triploid set of chromosomes. In cases with a partial molar pregnancy, the triploid fetus can develop, but it is anomalous and non-viable. In our case, we had a normal diploid fetus with a triploid hydropic placenta on one side and an appearing placenta on the other side.

There are some case reports of partial molar pregnancy with a diploid fetus [5–11]. There are three types of molar pregnancies with a normal live fetus [8]. Molar hydatidiform should be considered in pregnancies with high levels of serum β-hCG serially [9]. In partial molar pregnancies usually, ultrasound imaging shows a honeycomb-like echo in the placenta; the borders between the normal placental tissue and the honeycomb echo are not clear, and most fetuses are dead or malformed [10]. In a case report by Hossain et al., the fetus could not continue to live for more than 21 weeks, and the pregnancy ended with intra-uterine fetal death [11]. Still, there have been cases, e.g., Zeng et al., in which a live baby was delivered [12]. In pregnancies with partial hydatidiform moles, only a few villous vesicular changes occur; the cellular proliferation is bolded, 90% of the fetal chromosome karyotypes are triploid, and most pregnancies end with abortion and fetal death.

To understand the unique genetic pattern in our case and the underlying reason for a diploid fetus accompanied by a triploid hydropic placenta, we should consider that the characteristic of triploidy is the existence of three (3n) instead of two (2n) haploid chromosome sets on the cellular level. There are two types of triploidy depending on the parental origin; "diandric triploidy" or "diandry," where the extra chromosome set comes from the paternal origin, and "digynic triploidy," or "digyny," where the extra chromosome set comes from the mother [13, 14]. Mosaic triploidy happens less frequently than complete triploidy. The mechanisms suggested for mosaic triploidy include the fusion of one normal zygote with one triploid that results in a chimeric fetus, delayed fertilization of a zygote with a second sperm, and reincorporation of the second polar body into the fertilized egg [15].

All the molecular markers used in the QF-PCR assay on both amniotic fluid and chorionic villus samples showed similar size, indicating that the fetus and molar placenta had originated from the same zygote and the extra chromosome set may come from the incorporation of the second polar body into the fertilized egg in very early stages of postzygotic mitotic cell divisions (Fig. 3).

In summary, it is crucial to reach a definite diagnosis whenever we encounter a bipartite placenta. We should assess whether the hydropic appearance of the placenta 1) is due to a molar pregnancy, which can have catastrophic consequences or 2) is due to mesenchymal dysplasia, which can lead to IUGR or fetal demise, or 3) is just simple hydropic changes in the placenta with no critical consequence. Genetic study can help in differentiating them. Besides molar pregnancies with unique genetic patterns, mesenchymal dysplasia can also have some genetic aberration, including triploidy. So, in confusing cases, the genetic study of the placenta and fetus

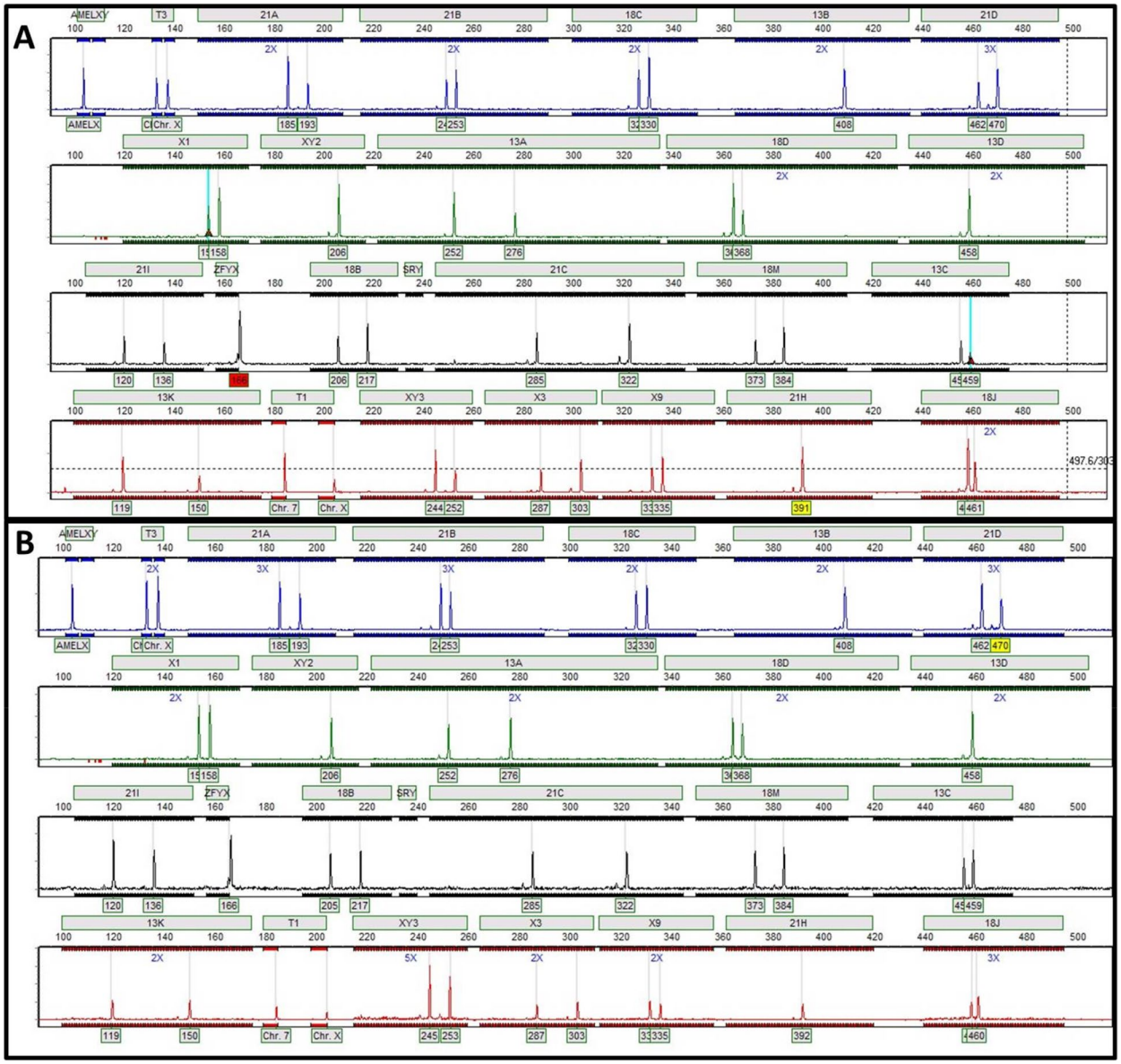

Fig. 3 The QF-PCR result. It shows a triploidy pattern in the CVS (A) and a normal diploid pattern in the AF (B). All the molecular markers had the same length in both samples, indicating that the extrachromosomal set in the triploid CVS has the same genetic constitution with one of the gametes, in this case, the ovum. Therefore, it can be inferred that the possible explanation of the occurrence of mosaic diploid/triploid, in this case, can be due to the incorporation of the second polar body into the fertilized egg in the early stages of postzygotic mitotic cell divisions

simultaneously can help in pregnancy management, counseling the family and clarifying the pregnancy outcome.

Conclusion

Based on our experience, genetic study of the hydropic part of the placenta and fetus simultaneously can help to clarify the diagnosis confidentially in similar cases, allowing the pregnancy to be continued or assisting the family in deciding to terminate it. We can say that the pregnancy is not a complete molar if we do not have a diploid genotype with entirely paternal origin in the hydropic part. In our case, a simultaneous amnion assay of the fetus revealed a normal diploid pattern, and the presence of a triploid genotype of the hydropic part of the placenta saved the pregnancy. However, in our experience and according to some other cases we encountered, sampling of chorionic villous in the hydropic placenta may fail due to its small amount. Further study and more cases are needed to have a decisive conclusion.

Our study shows that genetic assessment of the hydropic part of the placenta and amniotic fluid simultaneously, plus monitoring pregnancy with continuous follow-up of the patient's β-hCG titers and ultrasound imaging, can help determine the pregnancy's destiny.

Acknowledgements
The authors thank Muhammed Hussein Mousavinasab for editing this text. We appreciate this family for their valuable participation in this report.

Authors' contributions
the case managed by Dr.behrokh sahebdelDr.Zahra moghimi and Dr.Ehsan sobhanian wrote the main manuscript textDr.Ali Rashidi nejhad analized the genetics dataall other authors had a consulted during management of case.

Data availability
All data about this case is now available in the Tehran university of medical center - Maternal, Fetal and Neonatal Research Center data base.

Declarations

Competing interests
The authors declare no competing interests.

Received: 18 April 2024 Accepted: 9 October 2024
Published online: 02 December 2024

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