

Twin pregnancy with triploidy and co-existing live twin progressing to viability: Case study and literature review

Abstract

Background: Twin pregnancies consisting of a triploid fetus with molar change and a healthy coexisting fetus is an extremely rare phenomenon. Triploidy rarely advances to the second trimester, most resulting in a miscarriage, termination or stillbirth. Only five cases of a triploid fetus and a healthy co-twin have been reported in literature. We present the first documented case of a live delivery of both triploid fetus and coexisting viable twin.

Case presentation: A 23-year old woman, gravida 2, para 1 was admitted at 26+4 weeks gestation with DCDA twins and a shortened cervix. Sonography revealed polyhydramnios, an abnormal cystic placenta, and small aortic valve for the presenting twin. Emergency caesarean section was performed at 28 weeks gestation for cord prolapse. Twin A was born a live infant with ambiguous genitalia, syndactyly, epicanthic folds and cerebral cysts. Twin B was born a healthy live male. Histopathology revealed features consistent with partial hydatidiform mole, and cytogenetics revealed a diandric triploidy in twin A. Twin A survived until day 8 days at which supportive care was withdrawn.

Conclusion: Twin pregnancies with a triploid fetus and healthy co-twin pose a significant challenge for the clinician given the range of antenatal complications. Historically, the management approach has been towards selective termination of the triploid twin before 20 weeks gestation to increase survival and decrease the risk of preterm delivery for the healthy co-twin. Our case is rare where both babies were delivered alive with a coexisting triploidy.

Keywords: triploidy, twin pregnancy, viable co-twin, partial hydatidiform mole.

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Introduction

Triploidy is one of the most common chromosomal abnormalities in humans, accounting for 1 to 3 percent of all conceptuses and approximately 20 percent of spontaneous miscarriages.¹ It is characterised by having an extra set of haploid chromosomes resulting in a total of 69 chromosomes.²

According to paternal origin, triploidy can be classified into two phenotypes with distinct placental and sonographic findings. The most common type is diandric triploidy where chromosomes are of paternal origin, 80% of which are monospermic and 20% are dispermic.³ It is associated with a relatively well-grown fetus with either proportionate head size or slight microcephaly, increased nuchal translucency, and an enlarged and partially multicystic placenta with elevated levels of maternal serum beta human chorionic gonadotropin (β -hCG).⁴ The second type is digynic triploidy where the additional chromosomes are of maternal origin. It is characterised by severe fetal growth restriction, relative macrocephaly, and a small non-cystic placenta with decreased levels of β -hCG.⁵ Common structural defects in both types are syndactyly, adrenal hypoplasia and cardiac defects.

Twin pregnancies consisting of a triploid fetus with molar change and a healthy coexisting fetus are an extremely rare phenomenon.^{6,7} Triploidy rarely advances to the second trimester, most resulting in a miscarriage, termination or stillbirth. Only five cases of a triploid fetus and a healthy co-twin have been reported in the literature. We present the first documented case of a live delivery of both triploid fetus and coexisting viable twin.

Case report

A 23 year old woman, gravida 2 para 1 presented to our hospital at 19 + 3 weeks gestation with a dichorionic diamniotic (DCDA) twin pregnancy, on a background of one uncomplicated spontaneous vaginal delivery at term three years ago. Sonography revealed twin A had multicystic placental anomalies. Invasive testing was discussed with the patient and declined. She was monitored with regular ultrasound scans.

She was admitted at 26+4 weeks gestation after a routine scan showed a shortened cervix (4mm). Twin A was presenting breech, and had polyhydramnios (deep venous pocket (DVP) 13cm) with an abnormally thickened placenta with cystic areas. Twin B was cephalic presenting with a normal AFI and placenta.

On admission, she had antenatal corticosteroids as per the hospital protocol and was commenced on progesterone pessaries. The neonatal intensive care team consulted to discuss the prognosis for preterm twins.

On day 3 of her admission, she reported abdominal tightenings with copious vaginal discharge, and a headache not responsive to analgesia with no visual changes. On examination, she was normotensive, tachycardic with a maternal pulse of 120bpm, but remained afebrile. A speculum exam revealed a cervix that was short and closed. The remainder of her examination was normal. She had a normal cardiotocography (CTG). Investigations for pre-eclampsia were negative. An iron transfusion was arranged for microcytic anaemia.

Sonography revealed a cervix of 15mm, Twin A DVP 14cm and Twin B DVP 3.2cm, both cephalic presentation. A fetal MRI was arranged secondary to polyhydramnios and showed twin A had a mild bilateral ventriculomegaly with symmetrical periventricular cystic change. At this stage, non specific differentials included non-recent ischaemia, metabolic anomaly or infection. Twin B appeared normal.

At 27+5 weeks, she presented in preterm labour and examination revealed she was 6cm dilated and had membranes on view with cord presenting. A category 1 emergency caesarean section was performed. Twin A was delivered in cephalic presentation and neonatal examination showed a live infant with ambiguous genitalia, syndactyly and epicanthic folds. Twin B was a live male infant delivered in breech presentation. Post-operative period was uncomplicated.

Both twins were in neonatal intensive care unit. Twin A deceased on day 8 when care was withdrawn after consultation with patient and partner. Twin B remained healthy and well.

Fluorescence in situ hybridisation (FISH) and cytogenetics of twin A showed an abnormal result with male diandric triploidy. Histology of the placenta showed stem villous hydrops and cystic change in twin A. Twin B had normal results.

The patient was followed up by the Gestational Trophoblastic Centre. She became pregnant within 6 months and delivered a healthy live baby through an elective lower uterine segment caesarean section (Figures 1–3).



Figure 1 3D USS Twin A – fetal triploidy.

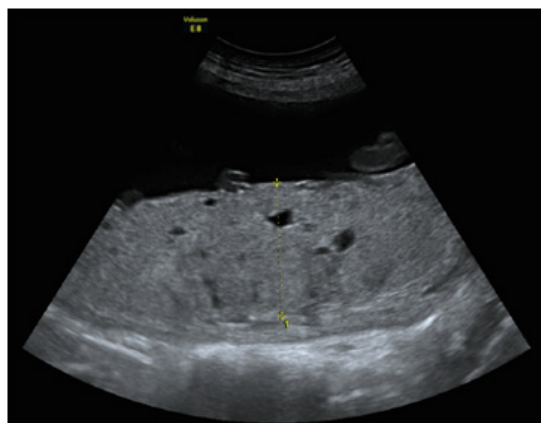


Figure 2 USS showing thickened cystic placenta for Twin A.

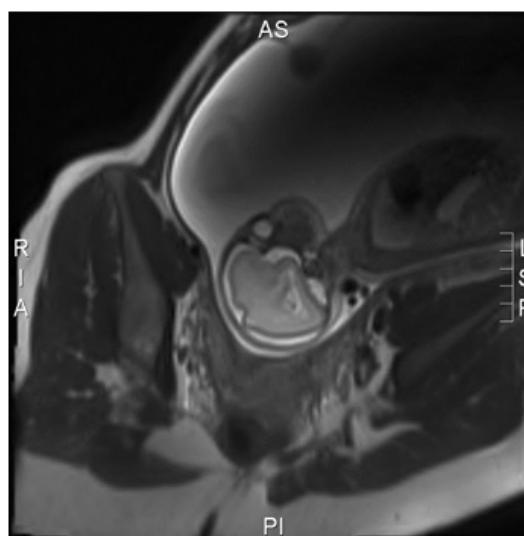


Figure 3 MRI showing bilateral ventriculomegaly and polyhydramnios in Twin A.

Discussion

To the best of our knowledge, our case may be the first to be reported where there is a concordant twin pregnancy with a live normal twin and a live triploid fetus. The features observed in the course of this twin pregnancy that correlate with the diandric triploidy phenotype include the large multicystic placenta, syndactyly and cardiac anomalies. This was confirmed on FISH and cytogenetic studies.

The management of twin pregnancies with severe genetic defect of one and regular genome represents a significant challenge. If a live fetus is present, chorionic villous sampling (CVS) or amniocentesis can be performed to confirm the diagnosis and offer selective termination of the triploid twin to ensure the best possible outcome for the healthy twin.⁸

Some of the maternal complications associated with twin pregnancies with a triploid fetus and healthy co-twin include per vaginal bleeding or haemorrhage, hypertensive disorders of pregnancy, hyperemesis, thyrotoxicosis, pulmonary oedema and thromboembolic phenomena. The risks to the fetus when pregnancy continues include miscarriage, intrauterine fetal demise, preterm labour, malpresentation and fetal growth restriction.⁹ Some of the complications demonstrated during the pregnancy in our case included preterm labour, fetal anomalies and polyhydramnios.

Conclusion

Twin pregnancies with a triploid fetus and healthy co-twin pose a significant challenge for clinicians, given the range of antenatal complications. Historically, the management approach has been towards selective termination of the triploid twin to increase survival and decrease the risk of preterm delivery for the healthy co-twin. Our case is rare as both babies were delivered alive with a coexisting triploidy.

Acknowledgments

None.

Conflict of interests

Authors declare that there is no conflict of interest.

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