Trisomy 21 fetus co-existent with a partial molar pregnancy: case report

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Summary

Background: Approximately 1 in 1,000 pregnancies in the United States are complicated by the presence of a hydatidiform mole. A Medline search revealed no reported cases of a trisomic fetus co-existent from 1966-1998. We present the case of a patient, initially found to have hypertension, edema, and proteinuria in the first trimester, and later found to have a partial molar gestation co-existent with a trisomy 21 infant.

Case Report: A 31-year-old female presented to her family practitioner in the first trimester and was found to have hypertension and proteinuria. A thorough work-up by a nephrologist revealed no cause. The patient was transferred to the Maternal-Fetal Medicine Service at 26 weeks’ and 1 day estimated gestational age. An amniocentesis revealed the presence of a fetus with trisomy 21, At 27 weeks’ and 3 days estimated gestational age, the patient underwent a cesarean delivery for a non-reassuring fetal heart rate. Pathologic examination of the placenta revealed the presence of a partial hydatidiform molar pregnancy.

Conclusion: The present account represents the first reported case of a fetus with trisomy 21 co-existent with a partial hydatidiform mole.

Key words: Trisomy 21; Partial hydatidiform mole.

Introduction

Approximately 1 in 1,000 pregnancies in the United States are complicated by the presence of a hydatidiform mole. In Japan the incidence is at least twice as high [1]. Ultrasonography can detect hydatidiform changes in only 75% of later confirmed hydatidiform moles [1]. Pircon et al. found that although all pregnancies complicated by molar gestations in their study had abnormalities discovered by ultrasonography, there were no consistent abnormalities detected [2]. Although the most common presenting sign is vaginal bleeding, occasionally a patient will present with pregnancy induced hypertension or preeclampsia.

Earlier diagnosis has led to a decrease in the rate of preeclampsia being a presenting sign in patients with a molar gestation [1]. The average estimated gestational age (EGA) at diagnosis has decreased from 16 to 12 weeks EGA. In earlier studies, as high as 27% of patients presented with preeclampsia but in more recent studies only 1.3% of patients presented with preeclampsia [1, 3, 4].

We present the case of a patient, initially found to have hypertension, edema, and proteinuria in the first trimester, who was later found to have a partial molar gestation co-existent with a trisomy 21 infant.

Case Report

A 31-year-old female presented to her family practitioner at 7 weeks’ estimated gestational age (EGA) and was found to have hypertension and proteinuria. During her last non-pre-

Revised manuscript accepted for publication April 15, 1999

Discussion

Molar gestations are not common, occurring in approximately 1 in 1,000 pregnancies in the western hemisphere [1]. Partial moles comprise 25-40% of molar
gestations [5]. While the most common karyotype for a complete mole is 46,XX (all parental), partial molar gestations usually have a triploid karyotype (46,XXX; 46,XXY; or 46,XYY). Although characteristically a fetus is present in a partial molar gestation, it typically dies during the first trimester. Only rarely does a fetus survive into the second or third trimester [1].

A twin gestation consisting of a complete molar gestation co-existent with a normal fetus occurs in 1 in 22,000-100,000 pregnancies [6]. These pregnancies are at increased risk for hemorrhage and persistent trophoblastic neoplasia [7]. Only 3 cases of partial hydatidiform mole co-existent with a twin gestation have been reported [6, 8, 9]. Two of these had 46,XX karyotypes and one of them had a triploid karyotype [6, 8, 9].

In the current case, we have a histologically proven partial hydatidiform mole co-existent with a fetus having a trisomy 21 chromosomal anomaly. A Medline literature search was performed and no other cases of a trisomic fetus co-existent with a partial molar pregnancy were found. Two possible explanations exist for the current case’s findings. Either this case represents a singleton gestation partial hydatidiform mole (with two different distinct chromosomal anomalies) or this case represents a fourth reported partial hydatidiform mole twin gestation. This patient did not have an extensive ultrasound performed until transferred from her primary care physician to the St. Vincent Hospital Maternal-Fetal Medicine Service in her third trimester. No evidence of a second fetus was found on ultrasonographic evaluation. Amniocentesis revealed a male fetus with trisomy 21. No evidence was found on pathologic evaluation of a second fetus when the placenta was evaluated after delivery. Therefore, we conclude that this case represents the first case of a trisomic fetus co-existent with a partial hydatidiform mole.

References


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