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-pregnant visit to her doctor's office, the patient had a blood pressure of 130/80 and was told she had "borderline" hypertension. The patient was followed by her primary care physician, and then at 15 weeks' EGA she noted increasing urine protein on dipstick, so a 24-hour urine study was done which revealed 408 mg of protein. A thorough work-up for secondary hypertension done at this time, including studies for pheochromocytoma, primary aldosteronism, Cushing's disease and systemic lupus erythematosus revealed no cause. Methyldopa was started at 16 weeks' EGA. An echocardiogram performed in the late second trimester was within normal limits except for mild left ventricular hypertrophy. Both maternal kidneys appeared normal by ultrasound. Labetalol was added at 23 weeks' EGA when the hypertension became worse.

The patient was transferred to the Maternal-Fetal Medicine Service at 26 weeks' and 1 day estimated gestational age because of worsening hypertension and suspected intrauterine growth restriction. An ultrasound performed at this time revealed a fetus below the 5th percentile for growth. Amniocentesis revealed the presence of trisomy 21. At 27 weeks' and 3 days EGA, the patient underwent a cesarean delivery for a non-reassuring fetal heart rate and an abnormal biophysical profile. A 530 gm male was delivered with APGARs of 5 at 1 minute and 9 at 5 minutes. The infant was admitted to the Newborn Intensive Care Unit and survived 3 days. Pathologic examination of the placenta revealed the presence of a partial hydatidiform molar pregnancy. The patient had serum ßhCG levels drawn weekly until two consecutive were <2 mIU and is being followed with bimonthly serum ßhCG levels for one year.

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

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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.

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