Triploid partial molar pregnancy and fetal neural tube defect detected through Down's syndrome screening

F. QUINTIERI - R. IDOTTA (*)

Summary: A third case of triploid partial molar pregnancy with fetal neural tube and abdominal-wall defect, detected through maternal serum alphafetoprotein and hCG screening for Down's syndrome is reported. A 23-year-old woman had elevated serum alphafetoprotein and hCG levels at routine screening at the 16th week of gestation. Ultrasound examination suggested a fetal neural tube and abdominal-wall defect with thickened placenta. Amniocentesis was performed. A triploid karyotype (69 XXY) was confirmed with standard cytogenetic analysis. Fetal neural tube and abdominal-wall defect was confirmed at the autopsy of the fetus. Placental histology was consistent with a hydatiform mole. When serum alphafetoprotein and hCG are elevated, during screening for Down's syndrome, the possibility of a partial molar pregnancy with fetal malformation should be added to the differential diagnosis.

Key words: Hydatiform mole; Partial mole; Down's syndrome screening.

INTRODUCTION

Often the classic ultrasound findings may not be present in the placenta of a partial molar pregnancy, especially in the first trimester of gestation. Consequently, prenatal diagnosis can be difficult. A third case of a triploid partial molar pregnancy with fetal neural tube and abdominal-wall defect more suspected initially, as levels were elevated serum alphafetoprotein and hCG. Ultrasound examination, amniocentesis and standard cytogenetic analysis confirmed the diagnosis.

Received February 12, 1996 from the
Clinica Ginecologica ed Ostetrica
Università di Padova
(*) Casa di Cura Villa Aurora, Reggio Calabria
Revised manuscript accepted for publication March 27, 1996.

CASE REPORT

A 23-year old woman, para 0000, at the 16th week of gestation underwent a maternal serum screening programme for Down's syndrome (measurement of unconjugated oestriol, human chorionic gonadotrophin and alphafetoprotein levels). The hCG level was 163.810 mIU (n.v. 6940 - 16320), alphafetoprotein 156 ng/ml (n.v. 18-32), oestriol unconjugated 1.3 ng/ml (n.v. 0.83-2.57). With the combination of these three parameters, the risk for Down's syndrome was 1:1933 while the risk for neural tube defect was 1:1. An ultrasound examination revealed a fetal neural tube and abdominal-wall defect. The placenta appeared thickened and contained multiple sonolucent areas. Amniocentesis was performed for chromosome analysis. Presumptive diagnosis of

partial hydatiform mole with fetal malformation was made. The pregnancy was terminated with prostaglandin vaginal suppositories, followed by curettage of the uterine cavity with no complications. Autopsy of the fetus showed hydrocefalia, omphalocele, syndactyly and pliable low-set ears. The heart presented a unique and dilated atrium. Placental histology was consistent with focal hydropic swelling of chorionic villi and moderate trophoblastic hyperplasia. Conventional cytogenetic analysis on cultured amniocytes revealed a 69 XXY karyotype. Quantitative hCG levels were followed weekly and the values decreased progressively until a zero value was reached after three weeks.

DISCUSSION

Molar pregnancy may be classified as either a complete or partial mole based on gross morphology, histopathology and karyotype ([1, 2, 3]).

Complete hydatiform moles, that occur in 1 per 1200 pregnancies, present with diffuse hydropic swelling of the chorionic villi and diffused trophoblastic hyperplasia in the absence of an identifiable embryo or fetus ([4]). Generally they have a 46 XX or 46 XY karyotype and the molar chromosomes are derived entirely from paternal origin ([5]). Partial moles occur in 1 per 10,000-100,000 pregnancies and are characterized by focal hydropic swelling of chorionic villi, moderate hyperplasia and identifiable embryos or fetal structures ([6]). Usually they have a triploid karyotype and the fetus presents congenital anomalies associated with triploidy, including syndactyly, neural tube and abdominal-wall defects ([7]). A correct diagnosis of partial hydatiform mole is made before evacuation in only about 10% of the cases. Prenatally the more common signs are vaginal bleeding and absent fetal heart beat (70%) ([5]). Specific ultrasound findings in the placenta may not always be present and levels of hCG have been reported as high, low, or normal ([8, 9]). In triploid pregnancies that are not partial moles, low levels of hCG have been attributed to placental insufficiency ([10]). In our case, the levels of hCG and alphafetoprotein were elevated and associated with a neural tube and abdominal-wall defect with triploidy, and histologic features of hydatiform mole in the placenta.

Only two other reported cases of a partial molar pregnancy with an open neural tube defect have been identified through high levels of alphafetoprotein and hCG during Down's syndrome screening ([11, 12]).

Many molar pregnancies, complete or partial, that have not presented clinically before 15 weeks are detected through Down's syndrome screening ([13]). In conclusion, when serum alphafetoprotein and hCG are elevated during screening for Down's syndrome, in absence of other clinical signs or symptoms, a partial mole with neural tube and/or abdominal-wall defect should be suspected and included in the differential diagnosis.

REFERENCES

1) "Gestational Trophoblastic Disease". Report of WHO Scientific Group, Technical Report Series 692, 1983, WHO Geneve.
2) Szulman A. E., Surti U.: "The syndromes of hydatiform mole. I. Cytogenetic and morphologic correlations". *Am. J. Obstet. Gynecol.*, 1978, *131*, 665.
3) Szulman A. E., Surti U.: "The syndromes of hydatiform mole. II. Morphologic evolution of the complete and partial mole". *Am. J. Obstet. Gynecol.*, 1978, *132*, 20.
4) Atrash H. K., Hogue C. J., Grimes D. A.: "Epidemiology of hydatiform mole during early gestation". *Am. J. Obstet. Gynecol.*, 1986, *154*, 906.
5) Berkowitz R. S., Goldstein D. P., Bernstein M. R.: "Natural history of partial molar pregnancy". *Obstet. Gynecol.*, 1985, *66*, 5, 677-681.
6) Watson E. J., Hernandez E., Miyazawa K.: "Partial hydatiform moles: a review". *Obstet. Gynecol. Surv.*, 1987, *42*, 9, 540-544.
7) Doshi N., Surti U., Szulman A. E.: "Morphologic anomalies in triploid liveborn fetuses". *Hum. Pathol.*, 1983, *14*, 716.
8) Oyer C. E., Canik J. A.: "Maternal serum hCG levels in triploidy: variability and need to consider molar tissue". *Prenat. Diagn.*, 1992, *12*, 7-9.

9) Romero R., Ghidini A., Sirtori M., Cullen M., Fisher N., Hobbins J. C.: "First trimester diagnosis of a partial mole with the combined use of ultrasound and chorionic villus sampling". *Am. J. Perinatol.*, 1989, *6*, 314-315.
10) Fejgin M., Amiel A., Goldberger S., Barnes I., Zer T., Kohn G.: "Placental insufficiency as a possible cause of low maternal serum human chorionic gonadotropin and low maternal serum unconjugated estriol levels in triploidy". *Am. J. Obstet. Gynecol.*, 1992, *167*, 766-7.
11) Zalel Y., Weiner E., Zabari A., Shalev E.: "High levels of maternal serum alphafetoprotein and human chorionic gonadotropins leading to the diagnosis of combined neural tube defect and partial mole". *Prenat. Diagn.*, 1992, *12*, 305-307.
12) Harper M. A., Ruiz C., Pettenati M. J., Rao N.: "Tripoid partial molar pregnancy detected through maternal serum alphafetoprotein and hCG screening". *Obstet. Gynecol.*, 1994, *83*, 5, 844-846.
13) Cuckle H. S., Densem J. W., Wald N. J.: "Detection of hydatiform mole in maternal serum screening programmes for Down's syndrome". *Br. J. Obst. Gynecol.*, 1992, *99*, 495-97.

Address reprint requests to:
F. QUINTIERI
Ist. Ginecologia e Ostetricia "G. B. Revoltella",
Clinica Ostetrica e Ginecologica
Via Giustiniani, 3
35128 Padova (Italy)