Selective second trimester fetal reduction due to 46XY, 10q+ fetus

B. KAPLAN - D. RABINERSON - M. HIRSCH - Y. PELED - A. NERI

Summary: Second-trimester amniocentesis of a twin gestation revealed one normal karyotype, whereas the other had a 46XY, 10 q+, indicating an excess of genetic material on chromosome number 10.

A selective reduction of the affected twin was carried out on the 20th week of gestation; the outcome of pregnancy and delivery at term of the normal twin were both uneventful.

Key words: Chromosomal abnormality; Fetal reduction; Twin pregnancy.

INTRODUCTION

Prenatal genetic fetal assessment is a well established procedure. When the fetus is affected by a genetic abnormality, termination of the pregnancy may be indicated. In the case of twin gestation, when only one fetus is affected by genetic abnormality, selective fetal reduction allows continuation of pregnancy with the healthy fetus and avoids either immediate termination of the pregnancy with both fetuses, or the undesirable option of delivering both twins at term.

Received December 28, 1995 from the
Department of Obstetrics and Gynecology,
Beilinson Medical Center Petach-Tikva,
Sackler Faculty of Medicine,
Tel Aviv, University, Israel
Revised manuscript accepted for publication February 2, 1996.

CASE REPORT

A 31-year-old patient, primigravida, had been treated with clomiphene citrate to induce ovulation, resulting in a twin pregnancy. At week 16 of gestation amniocentesis was performed, due to maternal anxiety, revealing a normal 46 XX karyotype for one of the twins and a 46 XY, 10 q+ karyotype for the other twin (e.g. an excess of genetic material on chromosome number 10). No abnormality was detected in the karyotype of the parents, indicating that the aberration in the affected fetus was new and would bear no consequence in future pregnancies.

After obtaining the patient's written consent, ultrasound-guided cardiocentesis and injection of KCl solution into the malformed male fetus was performed on the 20th week of gestation. The procedure was well tolerated by the second fetus.

Follow-up visits included bidimensional ultrasound assessment of fetal growth and biophysical profile. Doppler assessment of umbilical blood flow and biweekly nonstress test were all within normal range. Weekly evaluation of maternal coagulation function did not demonstrate signs of intravascular coagulation.

At 40 weeks of gestation, the patient spontaneously delivered a normal female child with birth weight of 3,600 g, Apgar score of 10 at 1 and 5 minutes, and umbilical vein blood pH

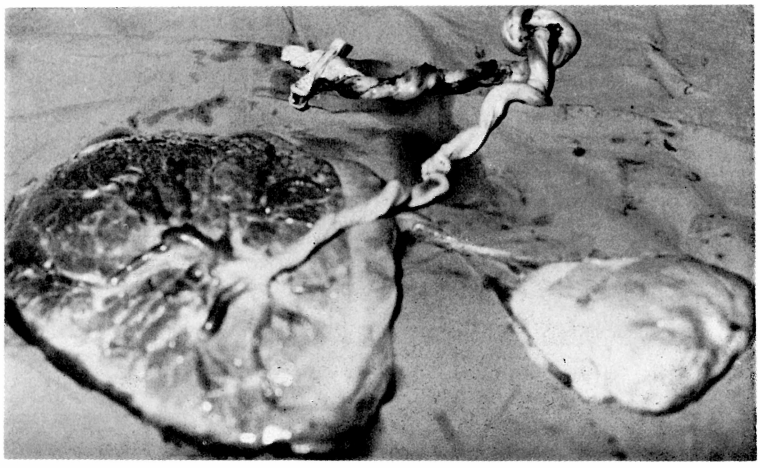

Fig. 1. — The normal placenta, together with the partially macerated fetus.

of 7.34. The placenta was spontaneously expelled, together with a partially macerated fetus (Fig. 1). Mother and newborn infant were discharged from hospital at normal time.

DISCUSSION

Prenatal genetic fetal assessment is an established procedure. Major indications for determining the fetal karyotype are maternal age ([1]), a previous child with chromosomal abnormality, parental chromosomal rearrangement, sex determination ([2]), or even parental anxiety about a possibly malformed fetus. If a prenatal genetic diagnosis is indicated in a twin gestation, both amniotic sacs should be sampled.

In cases where malformation of one fetus is confirmed, selective reduction of the affected fetus is indicated, to allow the birth of a healthy infant without the birth of a congenitally abnormal coexisting fetus and to preserve a singleton pregnancy, when the woman would otherwise need to have the whole pregnancy terminated ([3]).

When a genetic defect is detected by amniocentesis it may be difficult at later date to determine which infant carries the abnormality ([4]). This difficulty is reduced when the twins are of different sexes, as they can be easily distinguihed by ultrasound.

Embryo reduction (selective fetocide), after obtaining approval by a legal committee, is carried out during the second trimester of pregnancy, via transabdominal approach. Under ultrasound guidance, potassium chloride is injected into the fetal cardiac chamber (cardiocentesis) in order to kill the abnormal fetus ([5]). Management of the remaining live fetus requires the following: frequent antenatal surveillance; ultrasound evaluation of growth and well-being ([6]); determination of chorionicity, if possible, to predict potential risk of thrombotic morbidity to the surviving twin; serial ultrasound evaluation every 2-3 weeks for fetal growth, amniotic fluid volume, central nervous system and renal anatomy; Doppler assessment of the discordant twin's umbilical blood flow; weekly or biweekly nonstress tests or biophysical profiles; and weekly evaluation of maternal clotting factors (prothrombin time, partial thromboplastin time, fibrinogen, fibrin split products and platelets) ([7]). The above, coupled with active intervention, have been shown to improve the outcome ([8]).

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Address reprint requests to:
B. Kaplan, MD
14 Montefiore St.,
Kiriat-Ono 55226, Israel