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Fetoscopy in the Management of Twin Pregnancies Discordant for a Severe Abnormality

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Abstract. Selective feticide by fetoscopic air embolism was carried out in six twin pregnancies complicated by discordance for a severe abnormality. In three, a previous diagnostic fetoscopy had been performed. The procedure was effective and uncomplicated in all cases. One patient is undelivered and five have delivered healthy babies, three at term and two preterm. One of the latter, delivered at 28 weeks, died of complications in the neonatal period.

Key words: Prenatal diagnosis, Fetoscopy, Selective feticide, Twin pregnancy, Microcephaly, Spina bifida, Hemophilia A, Duchenne muscular dystrophy, Trisomy 21, Epidermolysis bullosa letalis

INTRODUCTION

Prenatal diagnostic techniques such as amniocentesis, ultrasound and fetoscopy are being increasingly applied to twin pregnancies. We have used fetoscopy in 22 twin pregnancies to obtain blood from fetuses at risk of hemoglobinopathies, hemophilia, and for confirmation of chromosome disorders by rapid karyotyping with fetal lymphocytes; to biopsy fetal skin for epidermolysis bullosa letalis; and for fetal visualisation.

The problem of discordancy poses a new clinical dilemma if one fetus has a severe disorder. To the alternatives of allowing the pregnancy to continue or of terminating it, a third possibility has been added, selective feticide (SF). This has been perfomed by hysterotomy [2], cardiac puncture [1,5], and intravascular air embolism [10]. The present paper reports on six twin pregnancies with this complication and in three of which a previous diagnostic fetoscopy had been performed.

METHODS

Diagnostic fetoscopy in singleton pregnancy [7] requires some modification for twin pregnancy. We have developed a method whereby a single uterine introduction of the trocar and cannula into one amniotic sac is made. Having sampled that twin, the needle is passed through the septum to sample the other, or, if it is too opaque, the endoscope is pushed trans-septally [9].

SF can be considered when one twin has a serious disease or abnormality, if the parents wish it, and if the pregnancy is thought to be dichorionic. Thickness of the septum on ultrasound scanning and opacity on fetoscopic visualisation provide good evidence for this. Great care must be taken to identify the affected fetus. Where there is a structural defect or a sex difference that can be detected by ultrasound or fetoscopy, this presents no difficulty. It is not straightforward, however, in the absence of a marker and reliance is placed on the topographical relationship of the fetuses and their sacs to the cervix. If the disease has been diagnosed from a sample of fetal blood or skin or amniotic fluid, this is repeated at SF to provide "posthumous" confirmation.

The fetoscope is introduced into the sac of the affected fetus at a site determined by real-time ultrasound scanning, so that placental damage is avoided and access is obtained to the umbilical cord insertion. When this has been seen and the umbilical vein identified the latter is punctured under direct vision. A fetal blood sample is aspirated and immediate confirmation is provided by a particle size analyser (Coulter Channelyzer) because fetal erythrocytes are larger than maternal. Bacterium-free air, which has been drawn through an epidural filter into a 20 ml syringe, is then slowly injected into the fetal circulation. After about 10 ml, asystole occurs and the fetal anatomical detail seen with ultrasound is lost due to an appearance of "white-out", caused by interference of the ultrasound waves by air in the fetal peripheral circulation. Circulatory arrest is also confirmed by seeing stationary air bubbles in the chorionic plate vessels. The fetal heart of the other twin remains undisturbed.

The patient usually leaves hospital on the following day, after a further ultrasound scan has been performed. Anti-D gammaglobulin is given to Rh-negative mothers and oral salbutamol is prescribed for one week.

PATIENTS

Six patients with discordant twin pregnancies have undergone this procedure. Three had a preceding diagnostic fetoscopy. Their histories are briefly summarised.

1. This patient's first child was microcephalic and in her second pregnancy she had twins, one of which was also found to be microcephalic by serial ultrasound measurements [3].
2. She was referred for prenatal diagnosis of hemophilia B, having one severely affected son. Amniocentesis of both sacs revealed two female fetuses, but one had a major spina bifida.
3. An obligate carrier of Duchenne muscular dystrophy, with two affected sons had had amniocentesis of both sacs in her country of origin. One fetus was female and the other was male. The latter therefore had a 1 in 2 chance of being affected. Since there was no sufficiently reliable prenatal test for the disease, she elected to have SF.

4. A carrier of hemophilia A with one severely affected son was referred. Following amniocentesis of both sacs, one male and one female fetus were found. Fetoscopy was performed on the male fetus and the factor VIIIC activity in a fetal blood sample was < lu/dl [6]. The result was the same in the sample taken at SF the following day.
5. A 40-year-old woman had amniocentesis of both sacs in her country of origin. Trisomy 21 was diagnosed in one fetus, both of which were female, and the procedure was repeated to obtain confirmation. When this was available, she was referred for SF. The diagram of the amniocentesis was not a reliable means of deciding which fetus was affected and although the ultrasound measurements of one were smaller than those of the other, this evidence was not regarded as conclusive. Fetal blood was taken by fetoscopy at 21 weeks from both twins and the lymhocytes cultured for 72 hours [4]. Karyotype analysis was thus available in a few days and showed that the smaller twin did indeed have trisomy 21. A futher blood sample at SF one week after the first fetoscopy produced the same result.
6. This patient's first child had died of epidermolysis bullosa letalis. In her second pregnancy, twins were diagnosed when she came for fetoscopy. Ultrasound scanning suggested that one fetus was male and the other was female and fetoscopic examination corroborated this. Skin biopsies were taken from each fetus as well as amniotic fluid. As for blood sampling, the second twin (in order of sampling) was approached by pushing the endoscope and biopsy forceps through a hole made in the septum because it was opaque. Ultrastructural studies showed the male fetus to be affected [8]. A further biopsy taken at SF one week later confirmed this. Chromosome analysis of cultured amniotic fluid cells agreed with the previous findings on fetal sex.

All these patients received prolonged counselling before SF. Four out of the six had little difficulty in deciding; the other two were mainly concerned about the thought of having a dead fetus inside them. The gestational age at SF ranged from 18 to 22 weeks and the procedure was uneventful and effective in all cases.

OUTCOME OF PREGNANCY

The antenatal care of all six patients continued at the hospital from which they were referred and one pregnancy is still progressing normally. Two patients experienced intermittent contractions and abdominal pains which required admission to hospital for a few days, but which did not proceed to premature labour. These patients were given β-mimetic agents to suppress uterine activity but it was felt that anxiety may have been a major component of their symptoms. It was suggested that clotting status be checked regularly and although this was not always done, no patient had complications indicative of a coagulopathy.

There were no abortions and five women delivered healthy babies as well as a mummified fetus. Two were born prematurely (< 37 weeks); one at 36 weeks had no problems. The other, born at 28 weeks did well initially, but, unfortunately, developed severe necrotising enterocolitis, had a colectomy and after prolonged postoperative complications, died at five weeks of age. The mother had had amniocentesis and two fetoscopies and these procedures could be related to the onset of spontaneous labour.

The remaining three women delivered at term (> 37 weeks), one by elective cesarean section and two vaginally.

CONCLUSIONS

These findings suggest that SF is an acceptable option for some parents faced with the difficult dilemma of a twin pregnancy discordant for serious abnormality. This is particularly so when there is already an affected child and the recurrence risk is high. A couple may then place extra "value" on the healthy twin.

The risks of SF by air embolism cannot yet be accurately determined but the present small series suggests that they may not be much higher than those normally accompanying twin pregnancy.

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