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ANY PLACE FOR ANTENATAL DIAGNOSIS IN THE THIRD TRIMESTER OF PREGNANCY?

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Summary: The need for a means of antenatal diagnosis of chromosomal abnormalities in the last trimester of pregnancy to avoid unnecessary intervention for the sake of the fetus is illustrated by a case in which polyhydramnios and suspected fetal hydrocephalus were found by ultrasound at 36 weeks gestation. The mother decided against early caesarian section for possible neurosurgery for the fetus. Later, she delivered vaginally of a baby with multiple abnormalities and trisomy 18.

INTRODUCTION

The appearance of third trimester complications in an otherwise uneventful pregnancy often presents a management dilemma, as it is possible that the fetus may have a lethal chromosomal abnormality.

CASE SUMMARY

A 34 year old para 1 woman was referred at 36 weeks. Polyhydramnios and dilatation of

the cerebral ventricles were detected by ultrasound 2 weeks before. Her last pregnancy was also complicated by polyhydramnios and delivery at 32 weeks of a stillborn female in China. On examination, no medical or infectious conditions were detected. The only fetal abnormality detected by a repeat ultrasound scan was asymmetric cerebral ventricular dilatation. The mother preferred vaginal delivery despite the possibility that immediate Caesarean section might give the fetus a better chance. Labour was induced and 3.5 litres of clear liquor were drained. A male baby weighing 1.49 kg was delivered after 4 1/2 hours and died three hours

Table 1. — *Abnormalities found on postmortum examination.*

Holoprocencephaly
Cleft lip and palate
Bilateral absent radii
Atrial septal defect
Ventricular septal defect
Patent ductus arteriosus

later. The father remarked on the similar features shared by this two babies. Multiple abnormalities were found (table 1). Karyotyping revealed trisomy 18.

DISCUSSION

In retrospect, this woman had recurrent pregnancies with trisomy 18 fetuses, and could have benefited from early antenatal diagnosis by chorion biopsy (Niazi *et al.*, 1981) or amniocentesis (Martin, 1980) if these were available. However, unsuspected or undiagnosed cases presenting later with complications like polyhydramnios, fetal growth retardation or fetal distress, which are common in fetuses with congenital abnormalities, often lead to Caesarean section for the sake of the fetus (Cassidy *et al.*, 1981, Schneider *et al.*, 1981). Even when fetal abnormalities are detected, the full extent is difficult to assess, and the decision is either early delivery for surgical correction or

to avoid Caesarean section because of the likelihood of lethal malformations especially associated with chromosomal abnormalities.

While the association of congenital malformations in trisomic syndromes with fetal growth retardation and polyhydramnios has been stressed, there is as yet no certain means of diagnosis in late pregnancy. Ultrasound has limitations. Fetoscopy and amniography are not widely available. Chromosome analysis on cells obtained by amniocentesis is crucial in these cases, but full karyotyping is difficult in the third trimester because of the high proportion of non-viable cells. The successful use of uncultured cells in the second trimester for fetal sexing had been reported (Nadler and Gerbie, 1970). We are now looking into the feasibility of using this method for detecting chromosomal abnormalities in the third trimester for similar situations in the future.

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