Antenatal diagnosis of fetal duodenal atresia by ultrasonic scan

Fetal malformations of the central nervous system can be detected early by ultrasonic examination. Gastrointestinal lesions, however, many of which may require immediate surgical correction after delivery, are more difficult to find.

Case history

A healthy 26-year-old woman whose first pregnancy had been uncomplicated booked at 14 weeks and attended the antenatal clinic regularly. The uterine size was consistently two weeks too large for her dates. Ultrasound scan at 19 weeks and six days verified the given dates. All other findings were normal. At 30 weeks there was apparent polyhydramnios: the uterus was tense and four weeks too large for dates. X-ray examination at 33 weeks showed no fetal abnormality. At 34 weeks the patient was admitted in early premature labour. Intrauterine ritodrine was given to decrease uterine activity and betamethasone to reduce the possibility of neonatal respiratory distress. A further ultrasonic scan four days after admission showed a biparietal measurement of 93 cm — again consistent with the patient’s dates. The fetal head and spine were normal. On taking cross-sections of the upper fetal abdomen, however, double fluid-filled structures were clearly visible (see figure). Measurement of the total internal uterine volume (6942 cm³) confirmed the apparent polyhydramnios. Slight uterine activity persisted over the next two weeks. It increased at 36 weeks. The cervix was found to be 5 cm dilated. Labour was allowed to proceed, amniotomy was performed, and six hours later a 2150 g female infant was delivered normally. A nasogastric tube was passed immediately and a large amount of bile-stained fluid aspirated. A clinical diagnosis of Down’s syndrome was subsequently confirmed by chromosomal analysis. Erect x-ray examination of the infant at 12 hours showed the “double-bubble” shadow in her abdomen, confirming duodenal atresia. The parents refused surgical treatment. The baby died at 8 days. Permission for necropsy was not given.

Comment

Polyhydramnios, in the absence of a maternal cause, may be associated with fetal abnormality in as many as half the cases. Lloyd and Clatworthy found 43% abnormal fetuses in their series of 76 pregnancies complicated by polyhydramnios. High small-bowel obstruction, proximal to the ligament of Treitz, was associated with polyhydramnios in 47%, of 49 pregnancies. Obstruction distal to the ligament of Treitz did not appear to give rise to excess liquor. Fonkalrud et al in a review of 503 infants with congenital duodenal atresia or stenosis found additional congenital malformations in 48%. Polyhydramnios was present in 45% of these cases and prematurity or dysmaturity (defined as a birth weight below 2500 g) in 51%. Down’s syndrome was present in 30%.

Cross-section of upper fetal abdomen scanned by unfocused transducer at frequency of 5 MHz. Note double cystic structures corresponding to stomach (S), dilated proximal duodenum (D), and fetal spinal canal (C).

Serum 2-hydroxybutyrate dehydrogenase activity and ineffective erythropoiesis

High levels of plasma lactate dehydrogenase activity are commonly found in patients with megaloblastic anaemia. Imperfect erythroblast maturation in this disorder results in the destruction of vast numbers of developing red cells rich in the anionic lactate dehydrogenase (LDH) isoenzymes LDH1 and LDH2. This is believed to be the cause of the