

# Antenatal diagnosis of abdominal wall defects: a missed opportunity?

J P Roberts, D M Burge

## Abstract

A review of six years' experience with antenatal diagnosis of abdominal wall defects by ultrasound showed its impact to be limited by poor detection rates. Twenty infants with exomphalos and 20 with gastroschisis were recorded but only 25 (63%) were diagnosed antenatally. The ultrasound false negative rate was higher for exomphalos (35%) than for gastroschisis (22%).

No difference was detected in the incidence of associated abnormalities, premature gestation, primary closure rate, or mortality between the antenatally and postnatally diagnosed groups for either exomphalos or gastroschisis.

Antenatal diagnosis of gastroschisis has little effect on management but allows parental counselling and in utero transfer. The frequency of concomitant abnormalities in exomphalos profoundly affects prognosis and the detection of these is the major role of antenatal diagnosis in this condition.

Failure to detect abdominal wall defects by ultrasound may be a reflection of technique or equipment, but some gastroschises may be of perinatal onset and not detectable antenatally.

The advent of obstetric ultrasound has made the prenatal diagnosis of abdominal wall defects commonplace and allows rational antenatal and perinatal management. The high incidence of associated abnormalities in fetuses with exomphalos has resulted in the recommendation that full antenatal evaluation be performed in every case.<sup>1</sup> Although these recommendations need not apply to gastroschisis, where the incidence of associated anomalies other than gastrointestinal is extremely low, specific anatomical features may predict the postoperative course and enable more accurate antenatal counselling.

With improvements in the standards of antenatal ultrasound the opportunity exists for early diagnosis of all babies with abdominal wall defects and the detection of any associated lethal anomalies. We have undertaken this review of our recent experience with abdominal wall defects to assess the impact of antenatal diagnosis on management and outcome.

## Methods

Records of all patients with antenatally diagnosed gastroschisis or exomphalos managed at this centre from January 1982 to December 1988, were entered prospectively on to a com-

puter database. Over the same period, similar cases not antenatally diagnosed were identified from ward admission records and theatre registers. Information on prenatal ultrasound scans, place, mode and timing of delivery, associated abnormalities, operative findings, and outcome were obtained from the database, maternal and infant notes, and ultrasound records, for both the antenatally and the postnatally diagnosed groups.

Premature gestation was defined as less than 37 completed weeks of pregnancy by expected or scan dates.

It was our policy over this period to evaluate all antenatally diagnosed cases of exomphalos with karyotyping and detailed ultrasound. Echocardiography only became available in the latter part of the period.

## Results

Over the period studied a total of 40 infants with abdominal wall defects (20 with gastroschisis and 20 with exomphalos) were managed at this centre, including one case of Cantrell's pentalogy and one of cloacal extrophy.

Mortality rates were similar for those diagnosed antenatally and those in whom the diagnosis was made at birth, being in the order of 45% for exomphalos and 15% for gastroschisis (table 1). Of the 12 total deaths nine had associated abnormalities (seven of the nine in exomphalos and two of the three in gastroschisis).

There was a high incidence of premature delivery in both conditions, with seven infants with exomphalos and 10 with gastroschisis delivering before 37 weeks of completed pregnancy.

A high incidence of concomitant abnormalities was found in exomphalos (12 of 20, table 2), but these all remained undetected prenatally

Table 1 Outcome

	Exomphalos		Gastroschisis	
	Diagnosed antenatally at birth (n=11)	Diagnosed at birth (n=9)	Diagnosed antenatally at birth (n=14)	Diagnosed at birth (n=6)
Delivery:				
Preterm	6	3	8	2
>37 weeks	4	6	6	4
Termination	1			
Surgical closure:				
Primary	7	7	11	5
Staged	2	1	3	1
No surgery	2	1		
Mortality:	5*	4	2	1
Rate	45%		15%	

\*One termination.

Wessex Regional Centre for Paediatric Surgery, Southampton General Hospital  
J P Roberts  
D M Burge

Correspondence to:  
Mr J P Roberts,  
Surgical Unit,  
The London Hospital,  
Whitechapel E1 1BB.

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Table 2 Associated anomalies in exomphalos

Diagnosed antenatally (n=11)	Diagnosed at birth (n=9)
Cleft lip	Beckwith's syndrome
Patent ductus arteriosus	Cantrell's pentalogy*
Patent urachus+duplex kidneys	Cloacal extrophy*
Trisomy 13+ileal atresia*	Multiple anomalies*
Phocomelia+cleft lip and palate	
Trisomy 18+cardiac anomalies*	
Trisomy 18 (termination)*	
Beckwith's syndrome+pulmonary hypoplasia*	

\*Died.

apart from the patent urachus and a case of trisomy 18. This contrasts with an incidence of three associated abnormalities, all of which were gastrointestinal, in a total of 20 gastroschisis. Of the three cases of trisomy associated with antenatally diagnosed exomphalos, one was terminated, one was misdiagnosed as gastroschisis (see discussion), and one presented too late to consider termination.

Thirteen (52%) of those diagnosed antenatally were diagnosed at less than 20 weeks' gestation. Of the 11 infants with exomphalos, the mean gestational age was 20 weeks (range 15–35) and of the 14 with gastroschisis, the mean gestational age was 24 weeks (range 18–37). In most cases in which the diagnosis had not been made until after 30 weeks' gestation, earlier scans had not been performed. Unfortunately the indication for scanning in most of this group was unascertainable.

Two infants had normal scans at 18 and 14 weeks only to have an abdominal wall defect detected on subsequent scans later in pregnancy, one at 35 weeks (exomphalos) and one at 37 weeks (gastroschisis), respectively.

Of the 15 patients diagnosed at birth, 10 had antenatal ultrasound scans performed (table 3). The indication for scanning in these 10 was routine (for dates) (n=5), maternal age (n=1), uncertain lie (n=1), and unknown (n=3). In six a scan had been performed after 20 weeks' gestation, and in four after 30 weeks. None of these scans detected the abdominal wall defects.

Overall 10 (29%) of the abdominal wall defects (six (35%) with exomphalos and four (22%) with gastroschisis) in this series were not diagnosed antenatally despite having had scans performed.

The mode of delivery was altered in seven of 11 with exomphalos and five of 14 with gastroschisis resulting in elective termination (n=1), caesarean section (n=9), and induction (n=2). The decision to alter the mode of delivery was made by the obstetrician without reference to the paediatric surgeon.

Table 3 Undetected abdominal wall defects in infants diagnosed at birth

	Exomphalos (n=9)	Gastroschisis (n=6)
No antenatal scan	3	2
Negative scans	6	4
Gestational age at last scan (weeks):		
<20	2	2
20–30	1	1
>30	3	1

The false negative rate was 35% for exomphalos and 22% for gastroschisis.

The site of delivery was altered by in utero transfer in 12 of the infants diagnosed antenatally to enable the mother to be close to the baby after delivery and to avoid the risks of neonatal transfer. In eight in utero transfer was not necessary as the family lived within the catchment area of the unit. In the remainder premature delivery took place at the centre of origin before transfer could be arranged. The timing of delivery was altered in three instances by termination (n=1) and induction (n=2).

Primary surgical repair was achieved in 14 of 20 infants with exomphalos and 16 of 20 with gastroschisis. Staged closure was required in three infants with exomphalos, of whom two died (pulmonary hypoplasia and venous infarction of bowel), and in four with gastroschisis, all of whom survived.

Counselling was given by a paediatric surgeon in all but six of the 25 abdominal wall defects detected antenatally. Those not counselled included three cases of exomphalos. Failure to counsel was due to premature delivery in two, counselling by outside sources in one, and was unclear in the remainder.

## Discussion

The antenatal diagnosis of abdominal wall defects by ultrasound is now widespread and usually straightforward. The criteria for ultrasound diagnosis are well reported.<sup>2,3</sup> Differentiation between exomphalos and gastroschisis is essential because of the association of chromosomal and lethal structural abnormalities in 30–80% of fetuses with exomphalos,<sup>1,4</sup> and the virtual absence of such abnormalities in fetuses with gastroschisis.<sup>1,5</sup> In one case in this series the anatomical features distinguishing these two conditions were not fully understood by the ultrasonologist and a mother carrying a baby with exomphalos and trisomy 13 was counselled antenatally as for gastroschisis, with disastrous results. All personal performing antenatal ultrasound scans should be fully conversant with the embryology, anatomy, and prognosis of all major structural abnormalities as recommended by the Royal Colleges of Obstetricians and Radiologists.<sup>6</sup> Access to specialists in the relevant postnatal field (for example, cardiologists, paediatric surgeons, etc) should be readily available to the ultrasonologist and obstetrician for accurate parental counselling.

The implications of an antenatal diagnosis of exomphalos have led to the recommendation of full evaluation of the fetus by detailed ultrasound, karyotyping, and echocardiography to enable rational decisions about the fate of the pregnancy and perinatal management.<sup>1,7,8</sup> In our series one pregnancy was terminated, and in a further case described above, termination would have been requested if the associated trisomy had been detected antenatally. In other reports the incidence of aborted fetuses (spontaneous or induced) is high.<sup>4</sup> Our rate is undoubtedly an underestimate as it takes no account of pregnancies managed without our knowledge.

Apart from a recent report<sup>9</sup> there is general agreement that caesarean delivery for abdomi-

nal wall defects has no beneficial effect on survival.<sup>1 7 8 10</sup> We do not advise caesarean section for those defects antenatally diagnosed, but there remains an increased use of this mode of delivery. This may reflect the increased complications associated with premature delivery, but may in part be due to a reduced threshold for caesarean section in these babies among obstetricians managing such pregnancies. All postnatally diagnosed babies were delivered vaginally without any apparent deleterious effects.

Antenatal ultrasound does not appear as efficient in detecting abdominal wall defects as might be anticipated. Of the 17 babies with exomphalos who were scanned, the defect was noted only in 11 (65%). This seems rather a low number but is similar to a previous report.<sup>7</sup> The detection rate was better for gastroschisis (78%), an improvement on the previous reported rate of 34%.<sup>7</sup> However, ultrasound failed to detect abdominal wall defects even well into the third trimester. This may be partly due to errors in technique, quality of equipment, and operator inexperience. Because of the rarity of abdominal wall defects, with an incidence of about 1:2550 pregnancies,<sup>4</sup> individual experience with these conditions will be small. Routine scanning performed for gestational age assessment is less likely to detect the defect than is detailed scanning performed for obstetric abnormalities such as a raised  $\alpha$  fetoprotein concentration or polyhydramnios. All three of the missed exomphalos cases scanned after 30 weeks were routine scans for dates or fetal lie (two of the defects were minor). There is also the possibility that some abdominal wall defects, particularly gastroschisis, may arise late in gestation or even perinatally.<sup>5</sup> One of our cases had a detailed anomaly scan, including fetal abdominal circumference measurement, performed by an experienced fetal ultrasonologist at 33 weeks' gestation with no abnormality detected, yet the baby was born with gastroschisis. Similar cases have been reported.<sup>11</sup>

Our overall mortality of 15% for gastroschisis and 45% for exomphalos is in keeping with other series,<sup>3 4</sup> although the mortality rate may rise as high as 86% in exomphalos if associated with cardiac defects.<sup>4</sup>

In our experience antenatal diagnosis does not appreciably alter the management or outcome in gastroschisis. It does allow in utero transfer of most babies for delivery at an obstetric unit with immediate access to paediatric surgery, and prenatal parental counselling. The value of counselling in this situation has yet, however, to be fully assessed. Some indication of likely outcome and severity of the postnatal course in gastroschisis, may be determined from

ultrasonically detectable features such as bowel wall thickness and gut loop distention.<sup>12</sup>

The presence of associated anomalies with antenatally diagnosed exomphalos, profoundly affects management and outcome. In cases with associated lethal anomalies termination may be recommended, but the low mortality for isolated exomphalos<sup>13</sup> should be appreciated and therapeutic termination cannot be justified in this situation.

Ultrasound should detect the presence of liver or spleen in the sac; this is associated with high mortality.<sup>7 13</sup> Some of this excess mortality is secondary to pulmonary hypoplasia with related chest deformity<sup>13</sup> and chest wall diameter may prove to be a further ultrasound predictive factor of outcome in exomphalos.

Because of the low rate of detection, antenatal diagnosis has had only a limited impact on the management of abdominal wall defects. Increased awareness of abdominal wall defects and use of detailed antenatal ultrasound performed as a screening procedure in all pregnancies may improve detection. The rarity of the condition and the volume of work presenting to obstetric ultrasound departments, however, are likely to result in a substantial proportion of abdominal wall defects remaining undiagnosed during pregnancy for the foreseeable future.

- Davidson JM, Johnson TRB, Rigdon DT, Thompson BH. Gastroschisis and omphalocele: prenatal diagnosis and perinatal management. *Prenat Diagn* 1984;4:355-63.
- Redford HA, McNay MB, Whittle MJ. Gastroschisis and exomphalos: precise diagnosis by mid-pregnancy ultrasound. *Br J Obstet Gynaecol* 1985;92:54-9.
- Nielson LB, Bang J, Norgaard-Pedersen B. Prenatal diagnosis of omphalocele and gastroschisis by ultrasonography. *Prenat Diagn* 1985;5:381-92.
- Mann L, Ferguson-Smith MA. Prenatal assessment of anterior abdominal wall defects and their prognosis. *Prenat Diagn* 1984;4:427-35.
- Moore TC. Gastroschisis with antenatal evisceration of intestines and urinary bladder. *Ann Surg* 1963;158:263-9.
- Royal College of Obstetricians and Gynaecologists and Royal College of Radiologists. *Report of joint ultrasound group of the Royal College of Obstetricians and Gynaecologists and Royal College of Radiologists*. London: RCOG, March 1987:9-15.
- Sermer M, Benzie RJ, Pitson L, Carr M, Skidmore M. Prenatal diagnosis and management of congenital defects of the anterior abdominal wall. *Am J Obstet Gynecol* 1987;156:308-12.
- Nakayama K, Harrison MR, Gross BH, et al. Management of the fetus with an abdominal wall defect. *J Pediatr Surg* 1984;19:408-13.
- Lenke RR, Hatch EI. Fetal gastroschisis: a preliminary report advocating the use of cesarian section. *Obstet Gynecol* 1986;67:395-8.
- Kirk EP, Wah RM. Obstetric management of the fetus with omphalocele or gastroschisis: a review and report of one hundred twelve cases. *Am J Obstet Gynecol* 1983;146:512-6.
- Knott PD, Colley NV. Can fetal gastroschisis always be diagnosed prenatally? *Prenat Diagn* 1987;7:607-10.
- Bond SJ, Harrison MR, Filly RA, Callen PW, Anderson RA, Golbus MS. Severity of intestinal damage in gastroschisis: correlation with prenatal sonographic findings. *J Pediatr Surg* 1988;23:520-5.
- Hershenson MB, Brouillette RT, Klempka L, Raffensperger JD, Posnanski AK, Hunt CE. Respiratory insufficiency in newborns with abdominal wall defects. *J Pediatr Surg* 1985;20:348-53.