West Midlands Congenital Anomaly Register

Anterior Abdominal Wall Defects 1995-1996

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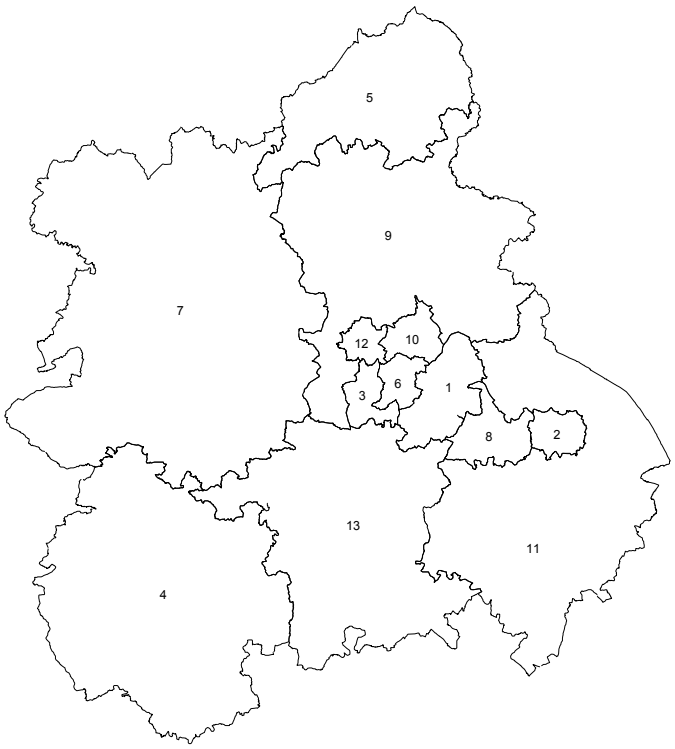
District Health Authorities in the West Midlands Region



KEY

1 BIRMINGHAM
2 COVENTRY
3 DUDLEY
4 HEREFORDSHIRE
5 NORTH STAFFS
6 SANDWELL
7 SHROPSHIRE
8 SOLIHULL
9 SOUTH STAFFS
10 WALSALL
11 WARWICKSHIRE
12 WOLVERHAMPTON
13 WORCESTERSHIRE

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PREFACE

This report is the second report on a major group of anomalies by the West Midlands Congenital Anomaly Register. By collaborating with staff at the Birmingham Children's Hospital it is the first report to include information on paediatric outcomes of surviving cases.

Abdominal wall defects are rare, making tertiary referral centres the major focus for gathering clinical experience. The spectrum of abdominal wall defects ranges from uniformly lethal lesions associated with other congenital anomalies to defects that can have an excellent prognosis.

This report details the prenatal diagnosis, pregnancy and immediate paediatric outcomes for West Midlands cases born in 1995 and 1996. The continued identification of new cases and follow up of those identified will allow better understanding, more accurate information and, therefore, improvements in the care given throughout pregnancy and into childhood.

Mike Wyldes
September 1998

TABLE OF CONTENTS

CLINICAL BACKGROUND OF ABDOMINAL WALL DEFECTS

Normal developmental embryology

Figure 1 - Rotation

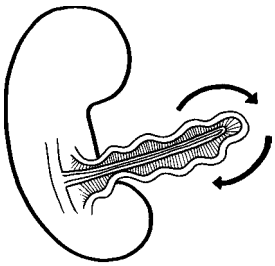


Figure 2 - Internalisation

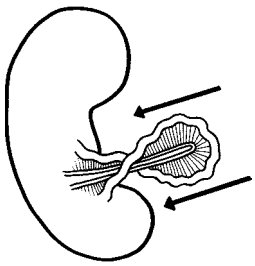
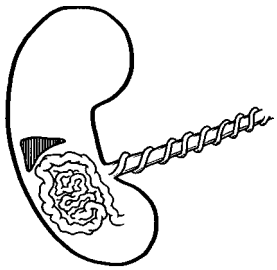


Figure 3 - Normal abdominal wall formation



In the third week of development the conceptus is a small disc of tissue. The edges of the disc become the front wall of the abdomen. The normal development of the bowel involves a period of exteriorisation followed by rotation, internalisation and finally closure of the umbilical ring by 10 weeks gestation. The mechanisms that drive the folding and then the narrowing of the umbilical ring are poorly understood.

EXOMPHALOS

Figure 4 - Small exomphalos

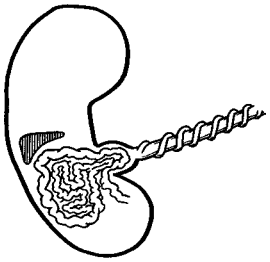
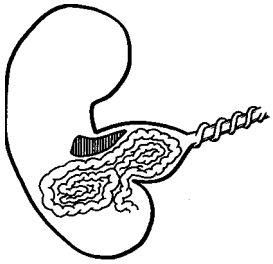


Figure 5 - Large exomphalos



Exomphalos is also called omphalocele. In an exomphalos some of the abdominal contents are found outside the abdomen in a thin clear sac to which the baby's umbilical cord is attached. The sac consists of amnion and parietal peritoneum with some mesenchyme between. A small exomphalos may contain only a Meckel's diverticulum whilst a large defect may contain the stomach, liver and bladder. Non-rotation of the intestines is commonly seen.

There are two rare related defects. Sometimes the diaphragm and sternum are also abnormal and the heart lies outside the chest (pentalogy of Cantrell). Another serious association is cloacal exstrophy where the hind gut is open on the surface of the abdomen between two hemi-bladders and below a broad low lying exomphalos.

Aetiology

Exomphalos is frequently associated with major abnormalities of other systems suggesting that this anomaly is not a simple failure of umbilical ring closure. The primary defect is likely to occur in early development. Most cases of exomphalos are sporadic, however the condition is often associated with aneuploidy and familial occurrence has been described.

Exomphalos associated with macroglossia and macrosomia is known as Beckwith-Wiedemann syndrome. These cases are sporadic and have a predisposition to renal hyperplasia and Wilms' tumour (duplication of 11p).

It is possible that exomphalos, like other anomalies, may be the final manifestation of a number of diverse primary developmental defects.

Prenatal diagnosis and pregnancy management

Most cases are identified by routine fetal anomaly scans and may be associated with elevated maternal serum alphafetoprotein (AFP) levels.

On ultrasound the normal cord insertion is not seen but is replaced by a mass representing the bowel or liver herniating into the base of the umbilical cord. The mass encapsulated by a membrane is seen to be attached to the cord.

Careful ultrasound examination will usually identify coexistent structural anomalies (especially cardiac anomalies) which exist in 70-85 % of fetuses. Karyotyping should always be offered as at least 35% of fetuses have a chromosome abnormality[1]. The presence of other abnormalities including aneuploidy will largely determine the prognosis. Frequently, a poor prognosis is predicted and termination of pregnancy requires discussion. Serial ultrasound measurements should be performed noting fetal growth if the pregnancy is continued.

In cases with a normal karyotype and no major associated anomalies a vaginal delivery may be appropriate. Large lesions, especially containing liver are delivered by caesarean section. Detailed prenatal ultrasound may not identify all potential problems making a careful neonatal assessment vital in these cases. In choosing the place of delivery, consideration should be given to the availability of fetal medicine, neonatal and specialist surgical expertise.

Paediatric management and surgery

With a small or medium sized exomphalos one operation to close the abdomen is all that is usually required. Sometimes the skin but not the muscles can be closed over an exomphalos sac and later operations are needed to close the muscles. If the sac is large, a silastic pouch can be used to allow progressive reduction of the bowel over a number of days before closure. Very large sacs can also be managed without operation. The sac can be painted with agents that lead to a scar forming, over which skin slowly grows. If skin alone has been used to cover the defect a ventral hernia results, and subsequent procedures are required to prepare this. Survival depends on the other defects and varies from 30-70%.

[1] Snijders R J M and Nicolaides K H (1996). *Frontiers in Fetal Medicine Series: Ultrasound markers for fetal chromosomal defects*. The Parthenon Publishing Group.

GASTROSCHISIS

Gastroschisis is the intrauterine evisceration of fetal intestine through a paraumbilical anterior abdominal wall defect, nearly always on the right side of the umbilicus.

Figure 6 - Gastroschisis

In addition to the evisceration of the intestine the stomach, bladder and gonads are often extra-abdominal. The liver does not herniate from the defect. There is no surrounding sac and so the intestines are exposed to the amniotic fluid during pregnancy. The bowel usually becomes shortened, thickened and dilated and is often matted together with adhesions. Intestinal atresias are seen in 5-15% of cases. The mechanism of the atresia is thought to be due to vascular compromise due to rotation or volvulus of the exteriorised intestine or kinking of vessels over the edge of the fascial defect. All affected children have non-rotation of the intestine.

Aetiology

Gastroschisis is thought to originate from a relatively late event in development since there are few associated anomalies. It may arise from an isolated vascular event involving the right side of the abdominal wall. This abnormality occurs sporadically and has a low recurrence rate although some familial cases are reported[2]. It is therefore extremely doubtful that a genetic cause is responsible for gastroschisis so the possibility of a nutritional or environmental aetiology remains.

Associations with young mothers[3] and low social class[4] are established but not understood. Smoking has been suggested as a possible risk factor and an increased risk for gastroschisis has been described in women using recreational drugs[5] before or in early pregnancy. There has been an increase in the incidence of gastroschisis over the last several years, with a clustering of cases in the under 20 year age group. Over the same period a relative increase in the incidence of cigarette smoking by pregnant teenagers compared to older women is reported. This suggests an association between recreational drug and cigarette consumption with gastroschisis, possibly due to an interruption of fetal omphalomesenteric arterial blood supply.

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[2] Torfs C P and Curry C J (1993). Familial cases of gastroschisis in a population-based registry. *American Journal of Medical Genetics* **45(4)**, 465-467.

[3] Roeper P J, Harris J, Lee G and Neutra R (1987). Secular rates and correlates for gastroschisis in California (1968-1977). *Teratology* **35(2)**, 203-210.

[4] Hemminki K, Saloniemi I, Kyyronen P and Kekomaki M (1982). Gastroschisis and omphalocele in Finland in the 1970s: prevalence at birth and its correlates. *Journal of Epidemiology & Community Health* **36(4)**, 289-293.

[5] Torfs C P, Katz E A, Bateson T F, Lam P K, Curry C J (1996). Maternal medications and environmental exposures as risk factors for gastroschisis. *Teratology* **54**, 84-92.

Prenatal diagnosis and pregnancy management

In gastroschisis the maternal serum AFP level is elevated in approximately 75% of cases, values of 4-5 multiples of the median are common.

The diagnosis is made on ultrasound by visualising the free loops of bowel that herniate through the anterior abdominal wall into the amniotic fluid. The umbilical vessels at the cord insertion can be seen to the left of the exiting bowel.

Although the risk of aneuploidy is low, a detailed ultrasound examination should be performed with early karyotyping if indicated. Cases of ruptured exomphalos have been reported and can be confused with gastroschisis. Counselling following diagnosis should include discussions of the prognosis and the likely surgical and medical support required. Discussions should involve both a neonatologist and a paediatric surgeon.

Serial ultrasonography allows the measurement of fetal growth and intestinal assessment looking for dilatation and abnormal peristalsis. The amniotic fluid is either normal or slightly diminished unless there is associated gastrointestinal atresia when polyhydramnios may develop. Consideration should be given to fetal assessment with umbilical artery Doppler velocimetry because of the association with stillbirth.

The choice of timing, mode and unit of delivery are controversial. There is a balance between the ultrasound findings of the bowel and indices of fetal well-being with the risks associated with preterm delivery. A vaginal delivery should be contemplated unless there is an obstetric contraindication. The same considerations in selecting place of delivery apply to cases of both exomphalos and gastroschisis.

Paediatric management and surgery

Immediate postnatal treatment includes resuscitation, transfer and operative reduction. Resuscitation requires large bore nasogastric tube drainage of the stomach, intravenous colloid and keeping the baby warm. The intestines should be supported to avoid vascular kinking. Often the best position is with the baby lying on its right hand side with the intestines also lying on this side. Wrapping in cling film or a bowel bag incorporating the legs helps reduce heat loss whilst maintaining good visibility of the bowel.

Transfer should be as prompt as emergency facilities allow. An appropriate neonatal incubator and a trained paediatrician and nurse are required.

In gastroschisis, survival is around 90% and at least 80% have a single operation to repair the abdomen. The umbilicus is usually preserved. Some procedures facilitate reduction such as anal dilatation with or without colonic washouts and abdominal wall stretching.

Forcing the intestines into too small an abdominal cavity can affect ventilation, vascular blood supply and renal perfusion. If this is the case a silo is fashioned and delayed closure performed after gradual reduction over 3-10 days. This is necessary if the abdomen is small particularly in the baby with intrauterine growth retardation. If an atresia is found, the bowel wall thickening usually precludes anastomosis at the time of closure. Either the atresia is left alone and the abdomen is closed or an ileostomy is fashioned. If the abdomen is simply closed a second procedure is planned for 3-6 weeks of age. Unfortunately these babies have greater morbidity and a higher mortality than those without an atresia. Necrotising enterocolitis is the commonest intestinal complication and intestinal dysmotility can be problematic. Further surgery may be needed for the repair of umbilical hernias, adhesion obstruction and undescended testes.

Patients require intravenous nutrition with normal feeding established in most cases at between 20 and 40 days but support may last for 6 or more months. Cisapride, a prokinetic agent reduces the time to the first sustained enteral feed and its use in gastroschisis is the subject of a British Association of Paediatric Surgeons multi-centre research project.

METHODS

West Midlands Congenital Anomaly Register

The West Midlands Congenital Anomaly Register (CAR) was set up in June 1994 and is administered by the West Midlands Perinatal Audit. The register aims to collect information on the occurrence of suspected and confirmed congenital anomalies of West Midlands residents, detected before and after birth. A number of minor anomalies are excluded from the register.

Reporting

Notifications are received by 2 methods. The first method is a notification card (Appendix A), which is used to notify the register of suspected anomalies. The card includes details of the type of anomaly and the estimated date of delivery and is most often completed by ultrasound departments. The second method is through a notification form (Appendix B), which contains much of the data set used for the Confidential Enquiry into Stillbirths and Deaths in Infancy (CESDI) but has additional details relating to the date that the anomaly is first suspected and the final postnatal diagnosis. The notification forms are usually completed by midwifes, obstetricians and paediatricians.

The Congenital Anomaly Register is maintained on the same database as the register of CESDI notifications of fetal and infant deaths. In this way the number of infants with lethal fetal anomalies can be validated. All anomalies are coded using the International Classification of Disease version 10 (ICD 10).

Additional information is also received from cytogenetics laboratories and Departments of Public Health. Inpatient episode data of infants with anomalies are also received from hospital information departments. These extra data are matched to the existing notifications and additional clinical information is added in some cases.

Definition of abdominal wall defect

ICD 10 provides 4 classifications of abdominal wall defect; Q79.2 (exomphalos/omphalocele), Q79.3 (gastroschisis), Q79.4 (prune belly syndrome) and Q79.5 (other congenital malformations of abdominal wall). Cases of umbilical hernia are excluded from this group of anomalies. ICD 10 code Q79.5 is used by the Congenital Anomaly Register for 2 groups of anomalies. The first group are cases notified to the register only as abdominal wall defects. Additional data from clinical notes or postmortem investigations would otherwise allow reclassification to the more specific codes Q79.2-Q79.4. The second group do not fit easily into any ICD 10 code since they are not seen in term births, these are cases of body stalk anomalies that are generally aborted. For the purposes of this report an abdominal wall defect refers to the two conditions of exomphalos and gastroschisis.

Denominators

The numerator comprises reported cases of abdominal wall defect. The denominator includes the numerator plus all babies who had any possibility of having an abdominal wall defect. As the number of cases of abdominal wall defect is small, the use of denominators in calculating incidence rates provides us with inter-district comparisons. Comprehensive clinical information is available for cases of fetal anomaly, however similar denominator data on all births are relatively difficult to obtain.

The appropriate denominator for calculating incidence rates is the total number of deliveries regardless of gestation but this information is unavailable. This report uses instead the sum of the number of births (live and stillborn), the number of terminations of pregnancy for fetal anomaly (less than 24 weeks) and any late fetal losses notified to the West Midlands Perinatal Audit. The denominator should also include all fetal losses under 24 weeks, however this information is not available. The numerator for incidence rates also includes cases of fetal loss due to abdominal wall defect.

Outcomes

This report divides outcomes of pregnancy into the following groups:

Late fetal loss less than 24 weeks (LFL),
Stillbirth 24 weeks or more (SB),
Neonatal death under 28 days (NND),
Postneonatal death 28 days up to 1 year of age (PNND),
Alive.

Termination of pregnancy (TOP) is defined as a therapeutic termination undertaken under the 1967 Abortion Act, and excludes situations of induction following spontaneous fetal death in utero. Some terminations of pregnancy may result in a registerable stillbirth, or indeed a live birth regardless of gestation.

Data Sources

Numerator data West Midlands Congenital Anomaly Register
Northern Region Congenital Anomaly Survey

Denominator data Office for National Statistics (ONS), registerable births
West Midlands Perinatal Audit, fetal losses above 20 weeks gestation
West Midlands Congenital Anomaly Register, fetal losses (all gestations) and terminations of pregnancy of fetal anomaly cases

Significance testing

All significance testing in this report was carried out by calculating the χ^2 statistic using 2 x 2 contingency (fourfold) tables. Yates's correction for continuity was incorporated into all χ^2 calculations. Significance was tested at a 95% confidence level i.e. $p < 0.05$ indicating a significant result. For small numbers, where an expected cell value was less than 5, a 2 tailed Fisher's exact test was used.

INCIDENCE RATES

There were 47 cases of exomphalos and 40 cases of gastroschisis born to West Midlands residents during 1995 and 1996.

The 40 cases of gastroschisis were all singleton pregnancies and among the cases of exomphalos there was one twin pregnancy. In this pregnancy one twin was a missed abortion and the other terminated at 19 weeks following diagnosis of trisomy 13, exomphalos and ventriculomegaly.

Table 1 - Abdominal wall defects: West Midlands residents 1995-96

	Exomphalos		Gastroschisis		Total		All
Year	n	rate	n	rate	n	rate	births
1995	26	3.82	16	2.35	**42**	**6.18**	68,004
1996	21	3.07	24	3.51	**45**	**6.58**	68,406
Total	**47**	**3.45**	**40**	**2.93**	**87**	**6.38**	**136,410**

Rate: rate per 10,000 births

Incidence rates for gastroschisis have traditionally been much lower than those for exomphalos. However, for the last two decades the birth incidence of gastroschisis reported nationally has risen with a small reduction in the number of babies born with exomphalos[6]. For West Midlands residents the incidence rates for exomphalos and gastroschisis are similar, but there was a 50% increase in gastroschisis cases between 1995 and 1996.

A 7 year retrospective analysis of ONS data on abdominal wall defects[5] reported an increased incidence of gastroschisis and exomphalos in Northern England and Scotland. Demographic differences may contribute to this difference as might the uptake of terminations following prenatal diagnosis.

There is an increased number of abortions in teenagers in whom there is an increased incidence of gastroschisis. In England & Wales the incidence of abdominal wall defects is similar to that of the incidence of abortions for fetal anomalies. It is therefore unlikely that the number of anomaly cases terminated by chance differ between cases of gastroschisis and exomphalos.

Table 2 - Abdominal wall defects: other anomaly registers 1995-96

	Exomphalos		Gastroschisis		Total	
Register	n	rate	n	rate	n	rate
England & Wales ONS						
1995	50	0.77	88	1.35	138	2.12
1996	41	0.63	94	1.44	135	2.07
1995-1996	91	0.70	182	1.40	273	2.09
West Midlands ONS						
1995	7	1.04	6	0.89	13	1.93
1996	3	0.44	7	1.03	10	1.47
1995-1996	10	0.74	13	0.96	23	1.70
West Midlands CAR						
1995	26	3.82	16	2.35	42	6.18
1996	21	3.07	24	3.51	45	6.58
1995-96	47	3.45	40	2.93	87	6.38
Northern Region NorCAS						
1995	6	1.74	17	4.93	23	6.67
1996	15	4.41	19	5.58	34	9.99
1995-1996	21	3.07	36	5.25	57	8.32

Rate: rate per 10,000 births

The CAR incidence rates for the individual diagnoses of exomphalos and gastroschisis and the combined abdominal wall defect rate are significantly different to that reported by ONS for both

[6] Tan K H, Kilby M D, Whittle M J, Beattie B R, Booth I W, Botting B J (1996). Congenital anterior abdominal wall defects in England and Wales 1987-93: retrospective analysis of OPCS data. *British Medical Journal* **313**, 903-6.

England & Wales and the West Midlands. Incidence rates reported by the Northern Region Congenital Abnormality Survey (NorCAS) are significantly different from the national rate. These results suggest under-reporting by the national Congenital Malformation System and the reasons for this have been discussed elsewhere[7].

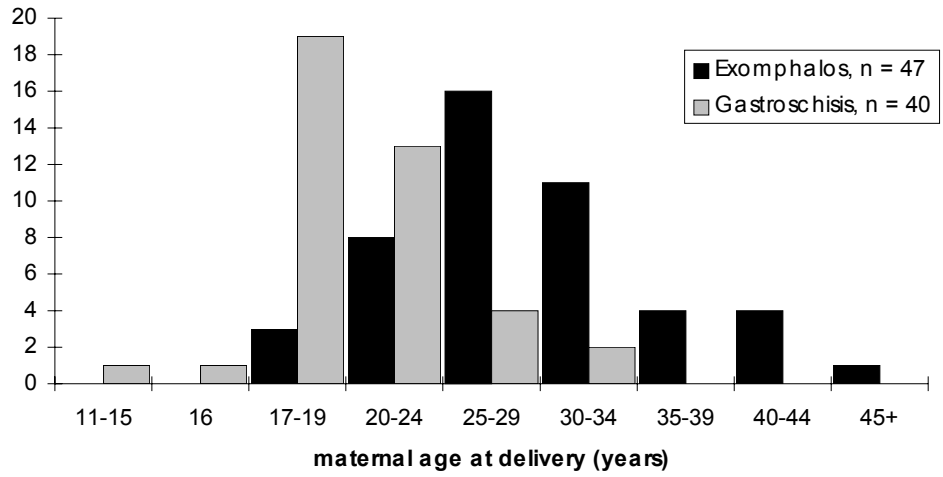
The West Midlands CAR operates in a similar way to NorCAS placing an emphasis on anomalies diagnosed prenatally. Reported incidence rates for exomphalos and all abdominal wall defects from these registers do not differ significantly, with higher exomphalos and lower combined abdominal wall defect incidence rates seen in the West Midlands. The incidence rate for gastroschisis in the West Midlands during 1995-96 was significantly lower than that reported in the Northern region ($p < 0.05$).

Maternal age

Table 3 - Abdominal wall defects: maternal age, West Midlands 1995-96

	Exomphalos		Gastroschisis		All births	
Age (yrs)	**n**	**% total**	**n**	**% total**	**n**	**% total**
11-15		0.0%	1	2.5%	422	0.3%
16		0.0%	1	2.5%	1,088	0.8%
17-19	3	6.4%	19	47.5%	8,877	6.5%
20-24	8	17.0%	13	32.5%	30,575	22.4%
25-29	16	34.0%	4	10.0%	45,757	33.5%
30-34	11	23.4%	2	5.0%	35,050	25.7%
35-39	4	8.5%		0.0%	12,386	9.1%
40-44	4	8.5%		0.0%	2,146	1.6%
45+	1	2.1%		0.0%	109	0.1%
Total	**47**	**100.0%**	**40**	**100.0%**	**136,410**	**100.0%**

Figure 7 - Abdominal wall defects: maternal age, West Midlands 1995-96



As noted earlier in this document there is an association between gastroschisis and low maternal age with a higher proportion of cases seen in women under 20. This correlation is not reported for exomphalos. Table 3 and Figure 7 confirm these observations with differences seen in the mean ages for the two anomaly groups. Over 50% of gastroschisis cases were born to mothers under 20 years of age with a mean maternal age of 20.5 ± 3.9 years compared to a mean maternal age of 29.1 ± 6.8 years for exomphalos ($p < 0.001$).

[7] The OPCS Monitoring Scheme for Congenital Malformations (1995). A review by a working group of the Registrar General's Medical Advisory Committee. *Office of Population Censuses and Surveys*.

Table 4 - Gastroschisis observed and age-standardised expected cases West Midlands and Northern Region 1995-96

	West Midlands			Northern region		
Age (yrs)	**Cases**	**All births**	**rate**	**Cases**	**All births**	**Exp.**
11-15	1	422	23.70	1	289	0.68
16	1	1,088	9.19		718	0.66
17-19	19	8,877	21.40	14	5,375	11.51
20-24	13	30,575	4.25	13	15,719	6.68
25-29	4	45,757	0.87	5	22,704	1.98
30-34	2	35,050	0.57	3	17,269	0.99
35-39		12,386	0.00		5,557	0.00
40-44		2,146	0.00		826	0.00
45+		109	0.00		54	0.00
Total	**40**	**136,410**	**2.93**	**36**	**68,511**	**22.50**

Rate: rate per 10,000 births
Exp.: expected number (age-standardised)

The association between gastroschisis and maternal age allows incidence rates for gastroschisis to be standardised for the maternal age of all deliveries. Table 4 shows the number of cases of gastroschisis expected for the Northern Region population using a West Midlands age-standardised rate (22.50 cases) compared with that observed (36 cases). This result is statistically significant.

Table 4 shows that when compared to NorCAS (another large regional anomaly register with similar notification methods and sources) the West Midlands has a lower crude incidence rate and a lower age-standardised incidence rate for gastroschisis, both results are statistically significant.

Table 5 - Abdominal wall defects: district of residence, West Midlands 1995-96

	Exomphalos			Gastroschisis			Total		
District of residence	**n**	**rate**	**O/E**	**n**	**rate**	**O/E**	**n**	**rate**	**O/E**
Birmingham	16	5.2	1.50	8	2.6	0.88	**24**	**7.8**	**1.22**
Coventry	4	4.9	1.44	6	7.4	†2.53	**10**	**12.4**	**†1.94**
Dudley	5	6.4	1.86	4	5.1	1.75	**9**	**11.5**	**1.81**
Herefordshire	0	0.0	0.00	0	0.0	0.00	**0**	**0.0**	**0.00**
Sandwell	4	4.8	1.40	1	1.2	0.41	**5**	**6.0**	**0.94**
Shropshire	0	0.0	*0.00	7	6.9	†2.34	**7**	**6.9**	**1.08**
Solihull	0	0.0	0.00	0	0.0	0.00	**0**	**0.0**	**0.00**
North Staffordshire	3	2.8	0.80	3	2.8	0.94	**6**	**5.5**	**0.87**
South Staffordshire	3	2.1	0.62	3	2.1	0.73	**6**	**4.3**	**0.67**
Walsall	4	5.7	1.65	0	0.0	0.00	**4**	**5.7**	**0.89**
Warwickshire	2	1.7	0.50	2	1.7	0.59	**4**	**3.4**	**0.54**
Wolverhampton	1	1.5	0.44	2	3.1	1.04	**3**	**4.6**	**0.72**
Worcestershire	5	3.9	1.14	4	3.1	1.07	**9**	**7.1**	**1.11**
Total	**47**	**3.4**	**1.00**	**40**	**2.9**	**1.00**	**87**	**6.4**	**1.00**

Rate: rate per 10,000 births
O/E: observed rate/regional rate
* significantly lower rate than West Midlands expected, $p < 0.05$
† significantly higher rate than West Midlands expected, $p < 0.05$

Variations in incidence rates for residents of District Health Authorities are unaffected by screening and referral policies by the maternity units who serve these populations. Coventry residents had statistically significantly higher incidence rates of gastroschisis and all abdominal wall defects when compared with the rest of the West Midlands during 1995 and 1996.

When compared with the reminder of the West Midlands Shropshire residents had a lower incidence rate of exomphalos and a higher incidence rate of gastroschisis, both these differences are statistically significant. This may have suggested an error in classifying abdominal wall defects but all cases were reviewed and found to be correctly classified.

Associated anomalies

Table 6 - Abdominal wall defects: associated anomalies, West Midlands 1995-96

Anomaly	Exomphalos	Gastroschisis
Chromosomal +/- cardiac	16	
Neural tube	6	
Skeletal	6	
Multiple systems	5	
Cardiac	4	1
Renal	2	
Syndrome (Beckwith)	1	
Total	**40**	**1**
% of cases	**88.9%**	**2.4%**

Table 6 shows that 89% of exomphalos cases had anomalies of other systems compared with 2% of gastroschisis cases. Two cases of gastroschisis had other gastrointestinal malformations, one case had jejunal atresia and another had shortening of the bowel, these were considered to be secondary to the abdominal wall defect.

The most common additional malformations were chromosomal anomalies and these only occurred in cases of exomphalos (40%). These chromosomal anomalies were 5 cases each of trisomy 13 and trisomy 18 (one with a ventricular septal defect), 2 cases of trisomy 21, one case each of Turner's syndrome (45X), triploidy (69XXX) and deletion of chromosome 7 (with an atrial septal defect and a ventricular septal defect) plus one case with an unidentified chromosomal anomaly. The cardiac anomaly reported in a gastroschisis case was an atrial septal defect that was diagnosed postnatally.

The prenatal diagnosis and termination of cases with associated anomalies would result in a reduction in the number of registerable births born with exomphalos.

DETECTION

Prenatal diagnosis

Table 7 - Exomphalos cases, outcome by prenatal diagnosis, West Midlands 1995-96

Outcome	Diagnosed < 24 weeks	Diagnosed > 24 weeks	No prenatal diagnosis	Total
Late fetal loss	2		1	**3**
Late fetal loss - TOP	27			**27**
Stillbirth	2			**2**
Stillbirth - TOP		1		**1**
Neonatal death			2	**2**
Neonatal death - TOP				**0**
Post neonatal death				**0**
Alive	3	3	6	**12**
Total	**34**	**4**	**9**	**47**

Table 8 - Gastroschisis cases, outcome by prenatal diagnosis, West Midlands 1995-96

Outcome	Diagnosed < 24 weeks	Diagnosed > 24 weeks	No prenatal diagnosis	Total
Late fetal loss			1	**1**
Late fetal loss - TOP	4			**4**
Stillbirth				**0**
Stillbirth - TOP				**0**
Neonatal death				**0**
Neonatal death - TOP				**0**
Post neonatal death	1			**1**
Alive	30	4		**34**
Total	**35**	**4**	**1**	**40**

Tables 7 and 8 show the outcome of pregnancies compared with the timing of any prenatal diagnosis. A prenatal diagnosis allows the parents the option of terminating affected pregnancies. This is still possible after 24 weeks but the pregnancy will then end in either a stillbirth or a neonatal death.

The presence of other anomalies in cases of exomphalos has not affected the number of early prenatal diagnoses made, 72% of exomphalos cases (34 of 47) diagnosed before 24 weeks compared with 98% of gastroschisis cases (39 of 40). Ten cases of abdominal wall defects were undiagnosed until delivery, two of these were cases of exomphalos that had ultrasound diagnoses of minor cord anomalies but the presence of major abdominal wall defects was not suspected.

Following a prenatal diagnosis before 24 weeks 79% of cases of exomphalos (27 of 34) and 11% of gastroschisis cases (4 of 35) were terminated. The increased termination rate with exomphalos is likely to be due to the poorer prognosis of the associated anomalies.

The natural history of the cases not terminated was that 37% (7 of 19) of exomphalos cases and 6% (2 of 36) of gastroschisis cases were stillborn or died during the first year of life. These survival rates may be affected by the early identification and termination of the more serious cases. An elevated perinatal mortality rate is therefore reported in pregnancies affected by gastroschisis[8]. There were no perinatal deaths reported in the above data, increased antenatal surveillance and early delivery may be partly responsible.

8 Adair C D, Rosnes J, Frye A H, Burrus D R, Nelson L H, Veille J C (1996). The role of antepartum surveillance in the management of gastroschisis. *International Journal of Gynaecology & Obstetrics* **52(2)**, 141-4.

Ultrasound diagnosis

Figure 8 - Abdominal wall defects: gestation at ultrasound diagnosis, West Midlands 1995-96

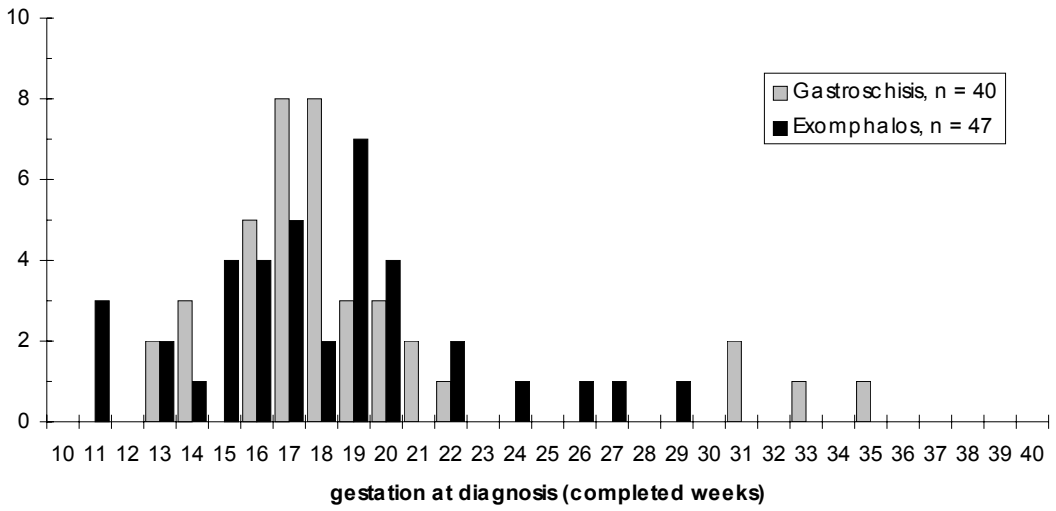


Figure 8 shows the gestation at first diagnosis of cases of gastroschisis and exomphalos. The majority of cases of both abdominal wall defects are diagnosed before 20 weeks. Raised maternal serum AFP and routine ultrasound have high sensitivity in identifying this group of anomalies and it is of interest that the median gestational age of diagnosis is close to when the test was performed. There is paucity of information as to whether data reported in Figure 8 indicates the gestational age of an initial ultrasound or as to whether other ultrasounds were performed before a definitive diagnosis.

Three cases of exomphalos were diagnosed at 11 weeks gestation.

A cohort of patients was scanned at Birmingham Women's Hospital and 44 (85%) had their final ultrasound report reviewed. Of these 13 (30%) had reduced liquor before delivery and 11 (25%) had elevated umbilical artery resistance index.

DELIVERY & POSTNATAL DATA

Place of delivery

Table 9 - Abdominal wall defects: place of booking and termination, West Midlands terminations of pregnancy (all gestations) 1995-96

	born at		
booked at	BWH	Other	Total
Birmingham Women's	2		**2**
Other hospitals	2	28	**30**
Total	**4**	**28**	**32**

Table 9 illustrates any changes between the maternity unit of booking and unit of birth for abdominal wall cases that were terminated, including any stillbirth or neonatal deaths. All of these cases will have a prenatal diagnosis. Of those cases booked at maternity units other than the regional tertiary centre 94% were managed at the hospital of booking.

Table 10 - Abdominal wall defects: place of booking and birth, West Midlands excluding terminations of pregnancy 1995-96

	born at		
booked at	BWH	Other	Total
Birmingham Women's	10		**10**
Other hospitals	24	21	**45**
Total	**34**	**21**	**55**

Table 10 shows the increased number of referrals made to the Birmingham Women's Hospital for delivery in cases that were not terminated. Nine of the 21 cases who were not born at the Women's Hospital had no prenatal diagnosis.

Gestation

Table 11 - Abdominal wall defect cases live births only, gestation at delivery (completed weeks), West Midlands 1995-96

Gestation	Exomphalos	Gastroschisis	Total
32 wks	2	1	3
33 wks	1	1	2
34 wks		4	4
35 wks	1	7	8
36 wks	1	8	9
37 wks	1	4	5
38 wks	6	8	14
39 wks	1	2	3
40 wks	1		1
Total	**14**	**35**	**49**

Seventy-five percent of gastroschisis cases were delivered (either spontaneously or iatrogenically) by 37 completed weeks of pregnancy. The median gestational age at delivery for gastroschisis was 36 weeks compared to 38 weeks cases of exomphalos.

Mode of delivery

Table 12 - Abdominal wall defect cases live births only, mode of delivery, West Midlands 1995-96

Mode of delivery	Exomphalos	Gastroschisis	Total
Vaginal - spontaneous onset	7	16	**23**
Vaginal - induced	1	4	**5**
Emergency caesarean section	2	9	**11**
Elective caesarean section	4	6	**10**
Total	**14**	**35**	**49**

The optimal mode of delivery for babies with abdominal wall defects is debated. Elective caesarean section delivery has been advocated to reduce the incidence of trauma to the exteriorised abdominal contents. There is however no evidence that vaginal delivery exposes the fetus to any increased risk of trauma. Table 11 shows that many cases of gastroschisis will end before 37 weeks so the potential for imposing a predictable time of delivery is unachievable in most cases.

A policy of elective caesarean section delivery would leave a large number of cases open to the increased risk of emergency caesarean section at the time of presentation in premature labour, with no proven benefit. Because of an increased perinatal mortality rate at term in fetuses with gastroschisis, a randomised-controlled trial comparing the elective induction of pregnancies at 36 weeks and expectant management is planned (RRAP).

Surgery

Table 13 - Abdominal wall defect cases surgical interventions for live births, West Midlands 1995-96

Surgery	Exomphalos	Gastroschisis	Total
Birmingham Children's Hospital	11	30	**41**
Leicester Royal Infirmary		3	**3**
Nottingham		2	**2**
none	3		**3**
Total	**14**	**35**	**49**

Of those babies with abdominal wall defects who were liveborn 84% were referred to the Birmingham Children's Hospital (BCH) for surgery. The cases who had surgery at Nottingham and Leicester were referred pre or postnatally from North Staffordshire Maternity and Walsgrave hospitals respectively.

The 3 exomphalos cases with no surgical intervention included 2 babies with trisomy 13 who both died in the early neonatal period. The third child had Beckwith's syndrome, with a very minor exomphalos and was discharged without any surgical intervention.

Transfers

Table 14 - BCH gastroschisis cases time from delivery to the start of surgery, West Midlands 1995-96

Interval	n
< 2 hrs	
2 - 3 hrs	8
3 - 4 hrs	9
4 - 5 hrs	7
5 - 6 hrs	4
6 - 7 hrs	1
7 - 8 hrs	1
>= 8 hrs	
Total	**30**

Table 14 shows the time interval between birth and the start of surgery. This time interval ranged from 2 hrs 1 min to 7 hrs 42 mins with a median value of 3 hrs 51 mins. Twenty-seven of the 30 cases of gastroschisis referred to Birmingham Children's Hospital were born at Birmingham Women's Hospital. The remaining 3 cases delivered outside the regional centre and had time intervals to surgery of 2 hrs 56 mins, 3 hrs 25 mins and 5 hrs 40 mins. All 30 cases had a prenatal diagnosis, which allowed some degree of planning regarding the place of delivery.

Paediatric outcomes

Table 15 - BCH gastroschisis cases paediatric outcomes, West Midlands 1995-96

Period	Started enteral nutrition	Time to full feeds	Hospital length of stay
< 7 days			
7 - 13 days	5	1	8
14 - 20 days	4	6	13
21 - 27 days	9	7	3
28 - 34 days	3	2	1
35 - 41 days	5	3	1
42 - 48 days			
49 - 55 days	2	1	3
>= 56 days	2	2	1
unknown		8	
Total	**30**	**30**	**30**

Babies spent a median time on enteral nutrition of 24 days (range 11 to 94 days) and took 25 days (range 13 to 95 days) to reach full feeds spending a median of 32 days in hospital (range 18 to 127 days). Two infants were discharged to local paediatric units before going home.

One patient died in this series with gastroschisis and an atresia he had only 5-6 cm of small intestine.

RECOMMENDATIONS

1.PERICONCEPTIONAL CARE

1.1 Recurrence of gastroschisis is less than 1%. For exomphalos the overall recurrence is less than 3%, but different aetiologies may carry different risks.

2.ANTENATAL CARE

2.1 Ultrasound examination of the umbilical cord insertion is mandatory as part of a 20 week mid trimester scan. If such an examination is undertaken, the prospective diagnosis of congenital anterior abdominal wall defects can be made with high sensitivity and specificity.

2.2 If the ultrasound diagnosis of a congenital anterior abdominal wall defect is made then consideration should be given to obtaining a second opinion at a tertiary referral fetal medicine centre. This is particularly prudent for exomphalos where a detailed ultrasound examination of the fetus is required to exclude co-existent structural anomalies (especially cardiac defects) and for consideration for karyotyping. Consideration should be given to methods of rapid karyotyping, especially when other structural anomalies are present.

2.3 Pregnancy monitoring should involve monthly ultrasound surveillance including biometry, Doppler studies of the umbilical artery and careful observation of the bowel to identify dilatation.

3.INTRAPARTUM CARE

3.1 Congenital anterior abdominal wall defects usually require surgical management in the immediate postnatal period. It may be beneficial for the mother to be transferred to a centre close to a paediatric surgical unit for delivery. Antenatal counselling with both neonatologists and paediatric surgeons before delivery is considered useful.

3.2 Gastroschisis may be associated with abnormal CTG changes. Extra vigilance is required as bilous amniotic fluid staining and meconium staining are difficult to differentiate.

4.POSTPARTUM CARE

4.1 A neonatologist should be present at the birth and a large bore nasogastric tube (10 Fr) should be passed to empty the stomach.

4.2 In gastroschisis intravenous colloid 10 mls/kg should be given. The baby is kept warm with cling film or a bowel bag to reduce heat and fluid loss and put in the right side decubitus position with the bowel on the right. The colour of the bowel should be observed. Venous congestion is the first sign of vascular compromise. Check to see if the bowel is twisted. Very rarely a small gastroschisis defect needs to be enlarged as a matter of urgency. Emergency transfer must take place to a paediatric surgical unit in a transport incubator with medical and nursing support. Vitamin K and antibiotics should be administered.

4.3 In exomphalos there is not the same urgency for closure. The baby should be fully assessed and then discussed with the paediatric surgical unit.

5.GENERAL

5.1 If parents choose to opt for termination of pregnancy then it is preferable for a postmortem to be performed on the fetus to make an accurate diagnosis and, therefore, to give the couple accurate information regarding recurrence risk.

5.2 Continuing prospective audit of outcome for congenital anterior abdominal wall defects is mandatory and data should be held on the West Midlands Congenital Anomaly Register. There is an urgent need for prospective early and long term follow up of babies surviving into infancy and childhood.

DEFINITIONS

Denominators
The population at risk in the calculation of rate or ratio.

Early neonatal death
Death during the first week of life, 0-7 completed days (on or before the 7th day of life, 0-6 days 23 hours 59 minutes).

Late fetal losses
For CESDI a late fetal loss is defined as a spontaneous abortion (miscarriage) occurring from 20 weeks 0 days (140 days) up to the end of 23 weeks 6 days (167 days). If gestation is unknown or uncertain, birthweights of 300 grams or above are reported.

Neonatal death
Death during the first 28 days of life, 0-28 completed days (on or before the 28th day of life, 0-27 days 23 hours 59 minutes).

Perinatal mortality rate
The number of stillbirths and early neonatal deaths (i.e. those occurring in the first week) during a stated year per 1,000 live and stillbirths occurring in the same year.

Post neonatal death
Death between 1 month and 1 year of age (28 days and over, up to just before 1st birthday).

Registerable births/deaths
Births or deaths that must be legally notified to the Registrar for Births and Deaths include all those delivered after 24 completed weeks of pregnancy, and all live births.

Stillbirth
Legal definition England & Wales.
"A child which has issued forth from its mother after the 24th week of pregnancy and which did not at any time after being completely expelled from its mother breathe or show any other signs of life".

Appendix A - Congenital Anomaly Register Notification Card

Appendix B - Congenital Anomaly Register Notification Form