INVITED COMMENTARY:
CURRENT ISSUES IN OBSTETRICS AND GENETICS

Gastrochisis—an overview

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Gastrochisis is an increasingly common malformation with unique fetal complications. Although it is usually an isolated defect, gastrochisis has a significant mortality rate of 5–10%. Complications include bowel damage, but growth retardation, oligohydramnios and fetal distress in labour are also common. There have been recent data that may help explain some of these observations, which will be reviewed in this article.

DEFINITION AND EPIDEMIOLOGY

Gastrochisis is a para-umbilical defect of the abdominal wall (almost always right sided) resulting in the eversion of abdominal contents into the amniotic cavity. The defect is usually small (less than 4 cm in length) through which various segments of bowel may be extruded and other abdominal organs, such as the fallopian tubes, may also be herniated (Pinzon and Barr, 1955). Unlike exomphalos, the exposed bowel loops are not protected from the amniotic fluid by a covering of peritoneum and the blood supply to the herniated organs is more likely to be interrupted. This can produce intrauterine and neonatal complications, including bowel torsion, atresia, necrosis and short bowel syndrome.

The prevalence of gastrochisis is usually quoted as 0.5–1 cases per 10 000 births, but the incidence has increased in many parts of the world over the last 2–3 decades (Penman et al., 1998; Rankin et al., 1999; Suita et al., 2000). The most striking epidemiological association with gastrochisis is young maternal age, especially less than 20 years of age (Tan et al., 1996; Penman et al., 1998). In England the incidence of gastrochisis doubled in a decade to 1.35 per 10 000 births and in the South West the incidence has been as high as 4.4 per 10 000 births (Figure 1). The rising teenage pregnancy rate in one fetal medicine centre 235 fetuses with abdominal wall defects were diagnosed. The karyotype was abnormal in 36% (n = 116) of the fetuses with exomphalos but in none of the 26 fetuses diagnosed with gastrochisis (Nicolaides et al., 1992). Since gastrochisis is almost always right sided it has been suggested that it may be caused by disruption of the right terminal branch of the superior mesenteric artery (which is located in the extraembryonic coelom of the body stalk) resulting in ischaemia and necrosis and so a secondary paraumbilical defect.

AETIOLOGY

Animal models suggest that radiation damage at the pre-implantation stage may increase the incidence of gastrochisis (Hillebrandt et al., 1998). Very rare familial clusters have been reported suggesting an autosomal inheritance pattern with variable expression (Torfs et al., 1998; Rankin et al., 1999; Suita et al., 2000) but these are exceptional. The incidence of chromosomal abnormalities is probably not increased with a diagnosis of gastrochisis. Over an eight-year period in one fetal medicine centre 235 fetuses with abdominal wall defects were diagnosed. The karyotype was abnormal in 36% (n = 116) of the fetuses with exomphalos but in none of the 26 fetuses diagnosed with gastrochisis (Nicolaides et al., 1992). Since gastrochisis is almost always right sided it has been suggested that it may be caused by disruption of the right terminal branch of the superior mesenteric artery (which is located in the extraembryonic coelom of the body stalk) resulting in ischaemia and necrosis and so a secondary paraumbilical defect.

DIAGNOSIS

In cases of gastrochisis the malformation should usually be suspected on routine second trimester ultrasound. Also, as there is an increased passage of alpha-fetoprotein (AFP) from the fetus into the amniotic
fluid, and hence the maternal circulation, an elevated maternal serum AFP often occurs in these pregnancies. The prenatal detection rate of gastroschisis has been estimated at between 76–90% but in regions with a good routine ultrasound programme it now approaches 100% (Axt et al., 1999; Forrester and Merz 1999; Walkinshaw et al., 1992). Detection is made after careful inspection of the anterior abdominal wall and the amniotic cavity because the loops of bowel are not always obvious next to the defect especially in later pregnancy or with reduced amniotic fluid (Langer et al., 1993). Gastroschisis may be distinguished from exomphalos by the herniation being next to a normal cord insertion and the absence of a covering of the defect (Fogata et al., 1999) (Figure 2). Polyhydramnios may be associated, probably in cases with reduced bowel motility or bowel obstruction, but oligohydramnios is a much more frequent finding and this is associated with a high incidence of intrauterine growth retardation which occurs in up to 60% of affected fetuses (Axt et al., 1999). There is no significant association with chromosomal abnormalities and so invasive testing is not usually offered because of this finding alone. Amyoplasia and arthropyhrosis may occasionally occur with gastroschisis: mobius anomaly and hypoplasia of the pectoral, deltoid and biceps muscles should be excluded on scan (Robertson et al., 1992).

COUNSELLING

After diagnosis the patient should be informed of the 5–10% incidence of fetal or neonatal death and long-term digestion problems associated with short gut syndrome. The prognosis for an isolated gastroschisis may be worse than for an isolated exomphalos because of the bowel protection in the latter malformation. Therefore, termination of pregnancy should be mentioned as an option in cases of gastroschisis. However, this is rarely chosen because of the much lower rates of other associated syndromes/abnormalities (Mann et al., 1984; Nicolaides et al., 1992). If the pregnancy is to continue, serial ultrasound examinations are recommended to detect gut complications, assess fetal well-being and plan delivery. The cases that survive to the end of pregnancy and into the neonatal period may be a self-selected group with a good prognosis. For this reason, combined with our inability to predict some fetal complications, the prognosis and counselling given to women at 20 weeks should be guarded. A consultation with a paediatric surgeon should be made to give the parents a better insight into the surgical management of gastroschisis.

COMPLICATIONS

Although 90–95% of infants with gastroschisis survive, the condition may be associated with severe complications for the fetus. As pregnancy progresses in gastroschisis the bowel usually becomes coated in an inflammatory fibrous peel resulting in thickening of the bowel wall, matting of the intestines, decreased bowel motility and obstruction. Morrison et al. (1998) demonstrated that the amniotic fluid in cases of gastroschisis contained inflammatory cells and significantly elevated levels of the pro-inflammatory cytokine, interleukin-8 compared to controls (Morrison et al., 1998). A chick model of gastroschisis suggests that bowel inflammation...
occurs due to the effects of the amniotic fluid on exposed bowel loops and worsens with gestational age due to the increased concentration of the surrounding fluid (Tibboel et al., 1986). Therefore, a vicious circle may occur: the exposed bowel becomes inflamed due to contact with the amniotic fluid and releases an inflammatory exudate into the amniotic fluid which increases in concentration with gestational age and further aggravates the bowel.

The cause of the increased frequency of CTG abnormalities and meconium staining has been intriguing (Crawford et al., 1992). In a study of 115 cases of gastroschisis between 1980 and 1996 in the South West of England, the infants with intestinal atresia had a significantly higher birth weight and fewer CTG abnormalities than those with patent bowel (Dixon et al., 2000). We hypothesized that bowel atresia, and so obstruction above the level of exposed loops of bowel, may significantly protect the fetus from growth retardation and CTG abnormalities in labour, perhaps due to the prevention of protein, salt or fluid loss across exposed bowel loops. Also, we found that staining of the amniotic fluid occurred just as often in fetuses with bowel atresia, so meconium could not have been passed to produce the characteristic green/black colour (Dixon et al., 2000). This suggests that the liquor staining in these pregnancies is the result of bile vomiting. Therefore, staining of the liquor in gastroschisis should not necessarily be taken to indicate meconium passage as a result of fetal hypoxia.

Compared to controls, chick embryos with gastroschisis were smaller and with reduced levels of sodium, chloride, amino acids and glucose (Lopez de Torre et al., 1991). A recent study from our unit showed that there was a significant decrease in fetal cord blood serum protein but a rise in amniotic fluid protein in fetuses affected with gastroschisis compared to normal controls or those with exomphalos (Carroll et al., 2001) (Figure 3). These studies support the hypothesis that in utero protein and fluid loss from the bowel to the amniotic fluid may account for the raised incidence of growth retardation, oligohydramnios and intrauterine death that occur in these pregnancies. The resultant hypovolaemia could also explain the high incidence of CTG abnormalities that occur in labour in gastroschisis.

**FETAL THERAPY**

Due to the possible secondary effects of severe oligohydramnios (pulmonary hypoplasia, limb compression and cord compression) two cases of gastroschisis with associated oligohydramnios were treated with serial trans-abdominal amnioinfusion of saline (Dommergues et al., 1996). The authors felt that the treatment may have reduced the complications of severe oligohydramnios and prematurity and it was observed that bowel peel and matting were minimised. In a retrospective study of 20 cases of gastroschisis the most significant poor prognostic factors were prematurity and the degree of inflammation of exposed bowel. Prematurity was most likely in the infants with severe bowel inflammation and heavily stained fluid. The amniotic fluid of the most severely affected fetuses contained higher concentrations of interleukins and TNF-α than gestation-matched controls (Luton et al., 1999). These cytokines may mediate bowel injury and possibly stimulate premature delivery (Morrison et al., 1998) Hence, the investigators proposed that amniotic fluid exchange would improve the prognosis in gastroschisis, and cases treated by amnioinfusion had a significant improvement in the success of surgical repair compared to those not treated in this way (Luton et al., 1999).

**MANAGEMENT**

Growth scans are difficult to interpret as the abdominal circumference is usually decreased in gastroschisis as a result of some bowel being outside the abdomen. However, careful evaluation of the amniotic fluid volume can detect if oligohydramnios develops and Doppler assessment can be used to assess placental function. With such monitoring, delivery is often delayed until 37–38 weeks (Crawford et al., 1992).

Premature onset of labour is common and there is a high incidence of CTG abnormalities during labour (Crawford et al., 1992). These CTG effects may be due to oligohydramnios producing cord compression. However, abnormal CTG tracings are often associated with normal Apgar scores so in pregnancies with this malformation they may not indicate ‘asphyxia’ during labour (Dixon et al., 2000). These may be due to the haemodynamic effects of fluid and protein loss across the fetal bowel to the amniotic fluid during pregnancy.

It is now rare for a case to be delivered without the abnormality having been prenatally diagnosed. To enable the best outcome for the fetus with gastroschisis it is vital that there is a team of obstetricians, neonatologists and paediatric surgeons with sufficient experience. Although controversy still exists over the obstetrical management of the fetus with gastroschisis, there is no clear evidence of any advantage of elective Caesarean section over vaginal delivery (Rinehart et al., 1999).
NEONATAL MANAGEMENT

A senior neonatologist and neonatal nurse should be present at the delivery and the paediatric surgeons promptly informed of the delivery of a baby with gastroschisis. To limit fluid and protein loss and prevent hypothermia, a silastic bag is immediately placed over the exposed abdominal contents. Handling of the bowel is minimised to prevent vascular compromise and nasogastric decompression, intravenous fluids (albumin/saline infusion) and broad-spectrum antibiotics are commenced. Any metabolic acidosis is corrected and, if possible, surgery is performed within 4 h of delivery.

The type of repair performed is relative to the degree of bowel inflammation, bowel matting and size of the abdomen relative to the extruded bowel that is present at delivery. Bowel in good condition may be replaced into the abdomen and primary fascial closure performed (Bianchi and Dickson, 1998). Delayed closure involves temporary coverage of exposed bowel with a silastic/dracon abdominal pouch and early parenteral nutrition.

After repair, bowel motility may be a problem particularly in the first week and parenteral feeding in the short term is required. Although long-term prognosis after successful surgery is excellent, morbidity is more likely when bowel obstruction, sepsis, delayed nutrition and closure complications occur. Mortality is associated with gut injury, late repair and the development of necrotizing enterocolitis (Snyder, 1999).

Long-term follow up studies suggest that those surviving initial surgery have an excellent prognosis. In one follow-up study of 23 subjects with neonatal repair for gastroschisis less than 10% had problems with bowel adhesions requiring further surgery with the remainder in excellent health (Davies and Stringer, 1997).

SUMMARY

The rising incidence of gastroschisis is of concern and further epidemiological studies are needed to understand the environmental factors that may be responsible. Greater understanding of the pathophysiological consequences of gastroschisis on the fetus may allow better in utero treatment options, reduction of IUGR and prematurity and improved post-operative outcome. Multicentre trials of amnioinfusion are currently being considered.

REFERENCES


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