A case of monozygous preterm twins with gastroschisis

Gastroschisis is a structural defect present at birth in which part of the abdominal wall is missing, allowing the intestines and other organs to protrude through the opening. This article highlights a rare case of gastroschisis affecting a set of monozygous preterm twins. The proposed aetiology of gastroschisis is reviewed alongside a discussion of how this case report might contribute to current understanding of the pathogenesis of the condition.

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Key points
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1. The incidence of gastroschisis is increasing and identifying a cause for this condition is therefore vital.
2. The prevailing opinion is that the aetiology of gastroschisis is largely exogenous.
3. This case describes a rare occurrence of monozygous twins with gastroschisis and hence contributes evidence for the interplay of genetics and environmental factors in the pathogenesis of the condition.

Gastroschisis is a congenital defect, characterised by protrusion of the intestine through a paraumbilical defect of the abdominal wall (FIGURE 1). It is not normally associated with other congenital anomalies and most babies born with this condition are expected to survive normally. This is a relatively rare problem but the incidence is increasing. The calculated incidence ranges from 1-5 in 10,000 worldwide. In the UK, the incidence has increased from 2.5 in 10,000 (1994) to 4.4 in 10,000 (2004). Most cases appear in the first pregnancy of younger mothers. Geographically there appears to be a north-south divide in rates of gastroschisis across the UK: southern England has a lower prevalence than the Midlands, northern England, Wales and Glasgow.

There is currently no known definitive cause of gastroschisis but most consider the aetiology likely to be exogenous in nature. There has been much speculation about the contribution of maternal drug and alcohol consumption in pregnancy but the causative relationship remains inconclusive. The most widely accepted explanation is disruption to the blood supply in early pregnancy, in particular early interruption of the fetal omphalo-mesenteric arterial blood supply.

Given that gastroschisis generally occurs sporadically, hereditary factors have tended to be understated. There are only a few isolated case reports of recurrence in families. Gastroschisis has been previously noted in both monozygotic and dizygotic twins, but the occurrence of this appears to be extremely rare. This observation...
combined with case reports describing cases of twins in which only one twin is affected is further support for an exogenous aetiology of this condition’.  

The case  

The mother was a 19-year-old, previously fit and well, primiparous woman of low socioeconomic status and slight build. She was rubella immune, had negative antenatal serology and was not documented to have smoked, drank alcohol or used drugs during the pregnancy. Antenatal scanning detected monochorionic diamniotic twins with intra-uterine growth restriction, too little amniotic fluid (oligohydramnios) and gastroschisis. They were delivered at a local district general hospital (DGH) by emergency caesarean section for worsening maternal pre-eclampsia at 29 weeks’ and four days gestation. The babies were delivered in good condition, with Apgar scores of 9 at one and five minutes. Cord gases were satisfactory and neither baby required any resuscitation at birth. The birthweights were 1000g and 1080g for Twin A and Twin B respectively, which were both below the tenth centile for gestation. Both babies were intubated and ventilated electively for transfer to the regional neonatal surgical unit on day 1 of life. Once there, both underwent full corrective surgery and had broviac catheters sited to aid administration of parenteral nutrition while feeds were established. They progressed well postoperatively, with minimal ventilatory requirements. Twin A was extubated on day 7 post-operatively, and Twin B was extubated on day 3. They both required a period on continuous positive airways pressure (CPAP) for presumed surfactant-deficient lung disease of prematurity (SDL) and feeds were slowly established. Twin B did considerably better than Twin A: CPAP was discontinued by day 12; by day 20, Twin B was fully enteral fed. Twin A remained on CPAP with a slow increase in enteral feeds up to discharge from the tertiary unit on day 37. She was also noted to have a moderate sized patent ductus arteriosus (PDA) on echocardiogram. An update from the DGH on day 144 revealed Twin A to have required CPAP until day 42 and subsequently high-flow humidified oxygen delivery (Optiflow™) until day 58. At the time of publication, she remained on 0.03-0.06L/min of nasal cannula oxygen and had established full enteral feeds, although still requiring some feeds via the nasogastric tube. 

Discussion  

Gastroschisis remains a reasonably rare problem, but the incidence has increased sharply from the early 1960s when it was approximately 1 in 150,000, to the current UK incidence of around 4.4 in 10,000’. Although this discrepancy may be partially explained by improvements in reporting practices, it remains a worrying trend. Studies have demonstrated a relationship between maternal demographics and drug and alcohol use in pregnancy. It has been suggested that the pathogenesis in this context is explained by a vascular accident at the time of involution of the right umbilical vein or of the development of the superior mesenteric artery’. Certainly, the relationship between gastroschisis and the use of vasoactive substances in pregnancy lends credence to the hypothesis of a vascular aetiology. Some studies have been able to demonstrate elevated risks associated with maternal use of aspirin, ibuprofen, and pseudoephedrine’.

Genetic factors are also under investigation. One early study suggested a deletion of the BMP1 gene resulted in a phenotype that resembled a human neonate with gastroschisis. Blood samples were collected from patients with gastroschisis but no mutation of the human BMP1 gene was identified’. This study therefore provided further evidence of a non-genetic aetiology for gastroschisis. Another study¹ suggested a range of genetic polymorphisms that were found to be associated with an increased risk of gastroschisis, particularly in association with maternal smoking; the hypothesis supports a joint interaction between genetic and environmental factors. Epidemiologically, geographic gradients are suggested in Europe and the UK’. Gastroschisis seems more frequent in Caucasians, and in northern compared to southern Europeans. The increasing prevalence of the condition suggests that environmental factors that have also increased in prevalence are likely to be involved. This is supported by the observation of a relationship between maternal factors such as young maternal age, low socioeconomic status, low maternal body mass index (BMI), poor maternal diet and discordant family life’. The understanding of these factors is important, as it will help in the development of preventative strategies in the future. These observations indicate the need for further studies investigating possible interactions between genetics and the environment as an explanation for gastroschisis. Current thinking is that the aetiology of gastroschisis is likely to be found in the interplay of multiple genes, and the addition of environmental exposures. This would be supported by the current case of twins with identical genetic and environmental exposures. This case fits the demographics of a young, first time mother of low socioeconomic status and appears to be one of the few case reports of gastroschisis documented in twins.

References  