

A case of recurrence of gastroschisis

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Objective

Our objective was to describe a case report of recurrence of gastroschisis and review the published literature on the subject.

Methods

Description of a case report and review of the published literature.

Results

We report a case of a 20 year old healthy pregnant woman with recurrence of gastroschisis in two consecutive pregnancies. Medical history was unremarkable except for history of cigarette smoking. Family medical history was unexceptional. At 20+5 weeks of pregnancy gastroschisis and bilateral mild pyelectasis was diagnosed by routine ultrasound. Amniocentesis was performed that showed a normal karyotype (46, XX) and fetal echocardiogram was normal. At 27+6 weeks, a dilated intra-abdominal intestinal loop was detected, with 17mm and no thickening of the wall. At 31+6 weeks the intra-abdominal intestinal dilatation was accompanied by an extra-abdominal one measuring 13mm without accompanying oedema. At 35+6 weeks the ultrasound showed fetal growth to be in the 8th percentile and a round anechogenic structure with 40x30mm was found herniated. Since bladder was not visualized during the entire exam, the hypothesis of bladder evisceration was assumed. The last ultrasound performed at 37 weeks showed the same findings. An elective caesarean section was performed at 37+5 weeks of gestation and a 2290g female newborn was born with an APGAR score of 9/10. An abdominal wall defect with evisceration of intestine, bladder, upper genital tract (uterus, tubal and ovaries) was diagnosed. After stabilization surgery was performed and the abdominal defect was corrected.

Conclusion

Gastroschisis is an abdominal wall defect found in about 1 per 4000 births with an increasing incidence described worldwide. It is majorly classified as a sporadic abnormality although familial cases have been reported, the risk of recurrence calculated to be 1%– 3. 5%. The only consistently reported risk factor is young maternal age, the incidence among teenage mothers being 6-10 times greater compared to those older than 25 years. Smoking, alcohol abuse, recreational drugs use and lower socioeconomic status are other possible risk factors for the development of the congenital malformation. More recently there is growing enthusiasm on the possible role of selected genetic polymorphisms involved in angiogenesis, inflammation and blood vessel integrity (nitric oxide synthase 3-NOS3; intercellular adhesion molecule 1-ICAM1; atrial natriuretic peptide NPPA; alfa adducing 1-ADD1) and their interaction with “traditional” risk factors for the development of the disease. These data support the role of a vascular defect as part of a multi factorial etiology involving both genetic and environmental factors.