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Fetal abdominal wall defects

Federico Prefumo, MD, PhD *, Claudia Izzi, MD

Prenatal Diagnosis Unit, Department of Obstetrics and Gynaecology, University of Brescia, Brescia, Italy

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The most common fetal abdominal wall defects are gastroschisis and omphalocele, both with a prevalence of about three in 10,000 births. Prenatal ultrasound has a high sensitivity for these abnormalities already at the time of the first-trimester nuchal scan. Major unrelated defects are associated with gastroschisis in about 10% of cases, whereas omphalocele is associated with chromosomal or genetic abnormalities in a much higher proportion of cases. Challenges in management of gastroschisis are related to the prevention of late intrauterine death, and the prediction and treatment of complex forms. With omphalocele, the main difficulty is the exclusion of associated conditions, not all diagnosed prenatally. An outline of the postnatal treatment of abdominal wall defects is given. Other rarer forms of abdominal wall defects are pentalogy of Cantrell, omphalocele, bladder exstrophy, imperforate anus, spina bifida complex, prune–belly syndrome, body stalk anomaly, and bladder and cloacal exstrophy; they deserve multidisciplinary counselling and management.

Introduction

The most common abdominal wall defects include ectopia cordis, bladder exstrophy gastroschisis, and omphalocele. The first three are classified in the group of ventral body wall defects, share a similar origin, and are likely to be caused by an abnormal closure of the later body wall folds that approach each other in the midline and close by the end of the sixth postmenstrual week. Alternative hypotheses for the cause of gastroschisis, such as amniotic membrane rupture at the insertion of the cord, abnormal apoptotic patterns during regression of the right umbilical vein, or vascular damage to the

* Corresponding author. Tel.: +39 030 399 5340; Fax: +39 030 399 5034.
E-mail address: federico.prefumo@gmail.com (F. Prefumo).

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base of the umbilicus caused by disruption of the omphalomesenteric artery, provide less convincing explanations [1]. Omphalocele is a separate entity, whose origin is attributed to a failure of physiologically herniated bowel loops to return from the umbilical cord to the abdominal cavity by the 12th postmenstrual week [1].

Epidemiology

In this chapter, we mainly concentrate on gastroschisis and omphalocele: they are the two most common anomalies. In 2011, based on the European registries included in the EUROCAT network [2], the total prevalence of gastroschisis was 3.09 per 10,000 births, with a live birth prevalence of 2.63 per 10,000 [2]. The corresponding figures for omphalocele were 3.29 and 1.13 per 10,000. The prevalence of gastroschisis has increased from 1980 to 2011, whereas that of omphalocele has remained stable (Fig. 1). A similar trend for an increasing prevalence of gastroschisis has been reported in North American populations, involving particularly pregnancies from younger women of non-Hispanic white maternal race and ethnicity [3]. It is speculated that these different trends are related to the cause of the two conditions. Although omphalocele seems to be mainly genetically determined, some evidence suggests that poor socioeconomic status and prenatal care, as well as teratogens (e.g. recreational drugs, salicylates, paracetamol, and pseudoephedrine) may be important contributors to the development of gastroschisis [4,5].

Another important historical trend has been the anticipation of the prenatal diagnosis of abdominal wall defects. This is because omphalocele and gastroschisis are easily diagnosed at the 11–14 weeks nuchal scan: a recent study based on over 45,000 pregnancies reported a sensitivity of 100% for both abnormalities [6], whereas a systematic review of the literature found a sensitivity near to 90% [7]. The increasing diffusion of first-trimester screening for chromosomal abnormalities has, therefore, increased the proportion of cases of these two abnormalities diagnosed at the nuchal scan. In the EUROCAT network, during the period 2007–2011, 22% of the chromosomally normal cases of gastroschisis were diagnosed before 14 weeks, and 50% between 14 and 23 weeks. The corresponding figures for euploid omphalocele were 35% and 30%. The overall prenatal detection rate was 91.6% for gastroschisis and 83.3% for omphalocele [2].

Gastroschisis

Gastroschisis is observed on ultrasound as a full-thickness defect in the abdominal wall, in most cases to the right of the insertion of the umbilical cord. It is only rarely located to the left of cord

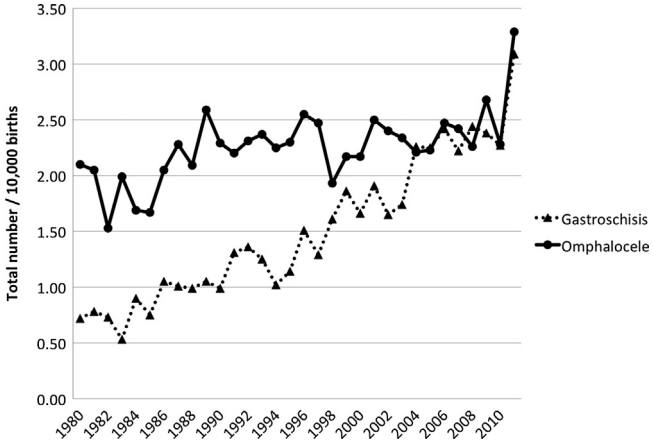

Fig. 1. Total prevalence of gastroschisis and omphalocele (cases per 10,000 births) in the EUROCAT network, 1980 to 2011.