

Retrospective analysis of fetal anterior abdominal wall defects

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ABSTRACT

Objective: To analyze fetal abdominal defects diagnosed during the prenatal period in the perinatology department in a tertiary center in Turkey.

Methods: This retrospective study consisted of 27 cases diagnosed with fetal abdominal wall defects between January 2011 and February 2014 in the perinatology outpatient clinic of Celal Bayar University, Manisa, Turkey.

Results: Eighteen (66.7%) cases were diagnosed with omphalocele, 6 (22.2%) had gastroschisis, and 3 (11.1%) had limb body wall defects. Twenty-one (77.7%) patients diagnosed either as omphalocele or limb body wall defect were offered karyotype analysis; 11 (52.4%) of them accepted the intervention, and 2 of the 11 patients (18.2%) had abnormal karyotype. Regarding the omphalocele cases; 12 (66.6%) cases had isolated omphalocele, whereas 6 of the 18 cases (33.3%) had associated anomalies. Expectant management was performed in 8 (66.7%) of 12 isolated omphalocele cases. Two of the isolated omphalocele group (16.7%) had missed abortion, the other 2 (16.7%) had termination of the pregnancy because of the associated chromosomal anomaly (47,XXY and 45,X0). Three of the gastroschisis group (50%) had missed abortion, and the other 3 (50%) had expectant management with cesarean delivery between 38-39 gestational weeks. Cases with limb body wall defect were terminated due to the lethal condition.

Conclusion: The prenatal diagnosis of fetal abdominal wall defects is important, because they differ greatly in terms of perinatal and neonatal morbidity and mortality due to underlying chromosomal abnormalities and associated structural anomalies.

Anterior abdominal wall defects include omphalocele, gastroschisis, ectopia cordis, bladder exstrophy, body stalk anomaly (BSA), OEIS complex (omphalocele, bladder exstrophy, imperforate anus, spina bifida complex) and pentalogy of Cantrell.¹ Among these defects, omphalocele and gastroschisis are the most common ones, with a prevalence of 3 per 10,000 live births for each.¹ The prenatal differential

diagnosis of omphalocele and gastroschisis is important, because they differ greatly in terms of perinatal and neonatal morbidity and mortality due to the underlying chromosomal abnormality and associated structural anomalies.² In this study, we report 27 cases with fetal anterior abdominal wall defects diagnosed during the prenatal period and review the relevant literature.

Methods. This retrospective study consisted of 27 cases diagnosed with fetal abdominal wall defects between 2011 and 2014 in the perinatology outpatient clinic at the Celal Bayar University, Manisa, Turkey. The patients were admitted to our perinatology outpatient clinic for routine detailed sonographic examination during gestational weeks 18-23, or they were referred to our perinatology outpatient clinic due to the suspicion of fetal abdominal wall defect. The data regarding maternal age and gestational age at the time of diagnosis was recorded. Sonographic examinations were performed with a Voluson 730 Pro system with a RAB 3,5-MHz array probe (GE Medical Systems, Milwaukee, WI, USA). All the pregnancies with abdominal wall defects were included in the study. The exclusion criteria were the physiologic herniation of the gut. Omphalocele was diagnosed, if a midline abdominal defect surrounded by a thin membrane was detected into which intraabdominal organs herniated.³ Gastroschisis was diagnosed if bowel loops were seen freely in the amniotic fluid with the umbilical cord in the normal insertion place.³ A body stalk anomaly was suspected if complex midline defects were detected concomitantly such as cranial defects, fascial clefts, thoracic, and abdominal defects.⁴ All cases with additional sonographic findings and omphalocele (both isolated forms and with associated anomalies) cases were offered fetal karyotype analysis. However, it was performed only in 11 cases. Four had transabdominal chorionic villous sampling (CVS) between 11-14 gestational weeks and 7 of them had amniocentesis (AC) between 16-20 gestational weeks. The outcome of the pregnancy was recorded from the data files.

Results. Of the 27 cases with anterior abdominal defects, 18 (66.7%) had omphalocele. Twelve of 18 omphalocele cases (66.6%) had isolated omphalocele, whereas 6 of them (33.3%) had additional structural

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anomalies. Six of the cases (22.2%) had gastroschisis, and 3 cases (11.1%) were diagnosed as limb body wall defects. Gestational age at the time of diagnosis was 16 weeks for isolated omphalocele, 14.33 weeks for gastroschisis, and 15.33 weeks for omphalocele with additional sonographic findings. Two patients had concomitant neural tube defects, one patient had a cardiac anomaly (ventricular septal defect), one had megacystis, and one had cystic hygroma. One of them was a conjoint twin. Out of the 11 karyotyping, 2 (18.2%) had chromosomal abnormalities (Klinefelter Syndrome and Turner Syndrome were diagnosed). No pregnant woman with gastroschisis had karyotyping. Expectant management was performed in 8 (66.7%) of 12 isolated omphalocele cases. Two of the isolated omphalocele group (16.7%) had missed abortion; the other 2 (16.7%) had termination of the pregnancy because of the associated chromosomal anomaly (47,XXY and 45,X0). Four of them (33.4%) had expectant management with cesarean delivery between the 38-39 gestational weeks. The other 4 of the isolated omphalocele cases (33.4%) were followed-up at another center, so we could not obtain the outcomes. Three of the gastroschisis group (50%) had missed abortion, and the other 3 (50%) had expectant management with cesarean delivery at the 38-39th gestational weeks. Cases with limb body wall defects were terminated due to the lethal condition. Cases with concomitant neural tube defects, hydrops fetalis, and conjoint twin were terminated. The VSD and megacystis with normal karyotype were managed expectantly.

Discussion. Omphalocele and gastroschisis are different entities: omphalocele is mainly genetically determined and occurs if the physiologically herniated bowel loops fail to return to the abdominal cavity at around the twelfth gestational week.⁵ Gastroschisis occurs if the body wall folds fail to come up together in the midline and close at around sixth gestational week.¹ The incidence of gastroschisis is also influenced by environmental factors, such as teratogens, and poor socioeconomic status.⁶ Recent studies show an increasing prevalence of gastroschisis compared with the prevalence in the 1990's.¹ Heydanus et al² analyzed 44 cases with abdominal wall defects. They report the ratio of omphalocele to gastroschisis as 3:1.² Similarly, we found the omphalocele to gastroschisis ratio as 3:1 in our series. However, the recent data showed that the prevalence of gastroschisis has increased in the last decade, whereas the prevalence of omphalocele has been stable resulting in nearly the same prevalence for both

conditions.¹ We think that this increase in gastroschisis may be attributed to both toxic environmental factors, and also increased prenatal detection rate. In our series, gastroschisis remained less common regarding the current data from Europe. It could be that these cases with isolated gastroschisis might not be referred to our center. It might be also attributed to the steady environmental conditions.

The prenatal detection rate is more than 90% for gastroschisis, and more than 80% for omphalocele.¹ Gastroschisis is an approximately 2-4 cm full-thickness defect in the abdominal wall through which the bowel loops herniate freely. It is generally found in the right side of the umbilicus. There is no peritoneal sac over the herniated organs. Concomitant anomalies are uncommon with gastroschisis. Up to 10% of the cases may be complicated with intestinal atresia.⁷ Intra uterine death of the fetus can occur in 5% of the cases.¹ Intrauterine growth restriction is common in infants with gastroschisis. They are typically 2000-2500 g at birth. Overall survival rate is more than 90%.

Omphalocele occurs with the midline defect through which intraabdominal organs herniate within a sac. The defect is generally greater than that of gastroschisis.⁷ Bowel loops and other intraabdominal organs such as the liver can herniate into the sac.¹ The umbilical cord inserts onto the sac membrane, not into the intact abdominal wall.¹ Rarely, the sac of the omphalocele can be ruptured and bowel loops can be seen freely in the amniotic cavity as in gastroschisis. In order to make a differential diagnosis between these 2 separate entities, the insertion of the umbilical cord will be guiding. Another pitfall in prenatal diagnosis of omphalocele is the need for differential diagnosis with an umbilical hernia. An umbilical hernia is a physiologic condition completed by the end of the eleventh gestational week.⁸ The accurate diagnosis of omphalocele should be made after the twelfth week. However, recent studies reported that the detection could be made as early as tenth week.⁹ The diagnosis is easier if the liver is also eviscerated.¹⁰

A body stalk anomaly is rare with an estimated incidence of 1 in 14000 to 42000 pregnancies.¹¹ A major anterior abdominal defect is accompanied by limb deformities, kyphoscoliosis, craniofacial defects, and absent or short umbilical cord. The herniated organs can be seen in extraembryonic coelom.¹² The fetus has usually a normal karyotype.¹¹ Pentalogy of Cantrell occurs above the umbilical cord insertion, and is defined by the anterior diaphragmatic hernia, sternal clefting, ectopia cordis, and intracardiac defect.^{13,14} Bladder exstrophy and cloacal exstrophy occur below

the umbilical cord level.¹⁵ Renal anomalies, neural tube defects, vertebral anomalies, and distention of bladder are helpful findings in the diagnosis of cloacal extrophy.⁵

The main limitation was the difficulty in obtaining the follow-up results of the newborns. Some pregnancies were terminated elsewhere. Therefore, we could not evaluate the postnatal period. The second limitation was that we could not obtain the autopsy findings of the terminated fetuses, because the parents did not accept the pathologic evaluation of the fetus due to religious reasons.

As a result, the accurate definition of the fetal anterior abdominal wall defects during the prenatal period is important to maintain the correct prenatal management and prepare the patient for the proper postnatal intervention. Regarding the omphalocele, amniocentesis should be performed. A detailed ultrasonography and fetal echocardiography should be also performed to exclude associated anomalies. Cesarean delivery is reasonable with an omphalocele mass greater than 5 cm in diameter. Regarding the gastroschisis, fetal karyotype analysis is not recommended. A detailed ultrasonography should be performed to exclude any other malformations. The mode of delivery depends on the obstetrical indications.

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