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Fetal abdominal wall defects: six years experience at a tertiary center

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Summary

The authors' aim was to detect the associated anomalies and their effect on the management of the fetuses with omphalocele and gastroschisis. Between the period of 2007-2013, the data of fetuses with abdominal wall defects were analyzed. Chromosomal abnormalities and associated morphologic anomalies diagnosed by ultrasonography and autopsy were evaluated. Of the 61 fetuses, ten (20.4%) omphalocele cases and nine (75%) gastroschisis cases were isolated. Chromosomal abnormalities were found in seven fetuses with omphalocele cases. All fetuses with abnormal karyotypes had multiple additional anomalies. Termination rate was 65.3% for omphalocele group versus none in the gastroschisis group. To give better counseling about the prognosis and outcome of the fetuses with abdominal wall defects, detection of additional anomalies as well as type of the defect are essential tools even if the karyotype is normal.

Key words: Abdominal wall defects; Chromosomal abnormality; Fetal anomaly; Gastroschisis; Omphalocele.

Introduction

Congenital anterior abdominal wall defects comprise a wide range of anomalies, with omphalocele and gastroschisis being the most common types. Although these defects are categorized under the same heading, their incidences, clinical properties, evaluation, and management differ from each other. The diagnosis of abdominal wall defects can be made sonographically as early as 12 weeks. The early detection of an abdominal wall defect however, is alone not enough to make a proper decision about the prognosis of the fetus.

In the absence of legal termination of pregnancy, antenatal diagnosis seems to have no significant impact on the outcome of neonates with anterior abdominal wall defects [1]. On the other hand, in countries where the termination of pregnancy is allowed, counseling the parents is possible through a detailed morphologic and genetic evaluation of the fetuses. If the karyotype is normal, management of pregnancy mostly depends on the presence of additional structural anomalies [2].

The aim of this study was to determine the role of associated chromosomal and structural anomalies to the management in fetuses with abdominal wall defects.

Materials and Methods

The present study was performed retrospectively from the recordings of the fetuses with abdominal wall defects from 2007 to 2013 in Izmir Tepecik Training and Research Hospital, which is the most widely circulating tertiary center in Aegean region of Turkey. Institutional Review Board approval was taken from the center. The fetuses diagnosed with abdominal wall defects were reevaluated again in the authors' perinatology department to identify the type of the defect and additional anomalies.

Women whose fetuses had abdominal wall defects were counseled with a genetician for chromosomal analysis. After obtaining informed consents of the parents, chorionic villus sampling, amniocentesis or cordocentesis were performed according to the gestational ages. Termination of pregnancy and autopsy were offered to the women when additional anomalies with bad prognostic outcomes or abnormal karyotypes were detected. All of the ultrasonographic findings of the terminated fetuses were confirmed with postmortem examination. Findings of autopsy and prenatal ultrasonography were evaluated together to identify the all associated anomalies.

The fetuses with the defects apart from omphalocele and gastroschisis were excluded from the study. The authors collected information regarding the maternal ages, gestational ages at diagnosis, fetal genders, fetal karyotypes, prenatal ultrasound, and postmortem findings. Fetuses were grouped as isolated, which had no additional anomalies other than omphalocele and gastroschisis. The anomalies secondary to the primary defects such as pulmonary hypoplasia and intestinal malformations were not considered as additional anomalies. Ectopia cordis and cloacal extrophy malformations were thought as parts of the Pentalogy of Cantrell (supraumbilical omphalocele - congenital heart anomalies - lower sternal, anterior diaphragmatic and pericardial defects) and omphalocele - cloacal extrophy - imperforate anus - spinal defects (OEIS) syndromes, respectively. The women who did not terminate their pregnancies were examined with ultrasonography until end of their gestations. Additional anomalies detected at later gestational weeks during follow up were also recorded.

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Table 1. — *Clinical properties of fetuses with omphalocele and gastroschisis.*

	Omphalocele n (%)	Gastroschisis n (%)	Total n (%)
Number of cases	49 (80.3%)	12 (19.7%)	61 (100%)
Maternal age*	27.2 ± 6.3	24.6 ± 6.7	26.9 ± 6.3
Gestational age at diagnosis*	16.1 ± 3.5	17.4 ± 3.4	16.3 ± 3.5
Female-male ratio	33/16 : 2.1	7/5 : 1.4	40/21 : 1.9
Isolated	10 (20.4%)	9 (75%)	19 (31.2%)
Non chromosomal syndromes	3 (6.1%)	-	3 (4.9%)
Chromosomal syndromes	7 (14.3%)	-	7 (11.5%)
Associated anomalies	39 (79.6%)	3 (25%)	42 (68.8%)
Termination	32 (65.3%)	-	32 (52.5%)

*Data are presented as mean ± SD;

Results

During the six-year period, 63 cases of abdominal wall defects were identified. Among these defects 49 were omphalocele, 12 were gastroschisis, and two were body stalk anomaly. Clinical properties of fetuses with omphalocele and gastroschisis are shown in Table 1. Two omphalocele cases were from dichorionic twin pregnancies. Mean maternal age of the study population was 26.9 ± 6.3 years. For omphalocele and gastroschisis groups, mean maternal age was 27.2 ± 6.3 and 24.6 ± 6.7 years, respectively. The mean gestational age at diagnosis was 16.1 ± 3.5 weeks for omphalocele and 17.4 ± 3.4 weeks for gastroschisis group. Female to male ratio of the fetuses was 2.1 in omphalocele group and 1.4 in gastroschisis group.

Table 2. — *Distribution of associated anomalies in fetuses with omphalocele and gastroschisis.*

Anomalies	Omphalocele n	Gastroschisis n	Total n
Central nervous system	20	1	21
Neural tube defect	15		15
Choroid plexus cyst	2	1	3
Corpus callosum agenesis	2		2
Dandy walker malformation	1		1
Skeletal system	20		20
Vertebral deformity	7		7
Club foot	6		6
Limb*	5		5
Sacral agenesis	2		2
Cardiac	12		12
Ventricular septal defect	3		3
Hypoplastic left heart	3		3
DORV	3		3
Other	3		3
Face and neck	7		7
Urinary	6		6
Multicystic kidney	4		4
Duplex ureter	1		1
PUV	1		1
Hydrops	6		6
Gastrointestinal	5		5
Hyperechogen bowel	3		3
Imperforate anus	2		2
Genital	4		4
Ambiguous genitalia	3		3
Bifid uterus	1		1
Diaphragmatic hernia	2		2
Cloacal extrophy	2		2
Ectopia cordis	1		1
Other	12	1	13

*Excluding club foot; DORV: double outlet right ventricle; PUV: posterior urethral valve.

Table 3. — *Associated anomalies of fetuses with chromosomal abnormalities.*

Case	Maternal age	Gestational age at diagnosis	Ultrasound findings	Autopsy findings	Karyotype	Karyotype method
1	33	21	DORV, meningomyelocele, single umbilical artery	Bilateral radius and thumb agenesis, horseshoe kidney	47,XX+18	AC
2	23	17	Diaphragmatic hernia, single umbilical artery, bilateral club foot, micrognathia, clinodactyly, absent nasal bone	Hypoplastic left heart, duplex kidney	47,XX+18	AC
3	38	19	Bilateral choroid plexus cysts, meningomyelocele, ascites, hyperechogen bowel, bilateral club foot, bilateral clenched hands, AVSD	Ambiguous genitalia, polydactyly in right foot, left heterotaxy	47,XY+18	AC
4	35	16	Increased nuchal fold, bilateral choroid plexus cysts, AVSD	Micrognathia, bilateral clenched hands	47,XX+18	AC
5	29	18	PUV, hypoplastic left heart	Corpus callosum agenesis, microphtalmia	47,XY+18	AC
6	29	16	Increased nuchal fold, bilateral choroid plexus cysts, micrognathia	Bilateral club foot, umbilical cord cyst	47,XX+18	AC
7	24	16	Cystic hygroma, hydrops, VSD	Horseshoe kidney	45,X	AC

AC: amniocentesis; AVSD: atrioventricular septal defect; CCA: corpus callosum agenesis; DORV: double outlet right ventricle; PUV: posterior urethral valve; VSD: ventricular septal defect.

Of all the abdominal wall defects, 19 fetuses (ten omphalocele and nine gastroschisis cases) were found to have no additional anomalies. The most common associated anomalies were central nervous system, cardiac and skeletal defects in omphalocele group, whereas minor anomalies (single umbilical artery, choroid plexus cyst) were found in gastroschisis cases. There were three syndromes other than aneuploidies associated with omphalocele, two of them were OEIS cases, and one of them was a Pentalogy of Cantrell case. Distribution of the associated anomalies according to their sites are shown in Table 2.

Seven chromosomal abnormalities, which included six Trisomy 18 and one Turner syndromes were found. All of the chromosomal abnormalities were detected from the fetuses with omphaloceles and multiple additional anomalies (Table 3). In the present study, 52.5% of the pregnancies with abdominal wall defects and 65.3% of pregnancies with omphaloceles were terminated. No fetus was terminated in gastroschisis group.

Discussion

In this study, of the 61 fetuses with abdominal wall defects, 49 were associated with omphalocele, 12 were with gastroschisis. The high ratio of omphalocele to gastroschisis cases (4:1) in the present study compared to other studies was related to the characteristic of the study population [3]. It is thought that gastroschisis is caused by environmental factors rather than genetic. Vasoconstrictive agents, smoking, alcohol, and young maternal age were the most blamed causative factors [4, 5].

Termination of pregnancy is considered as an option in counseling women about their fetuses with abdominal wall defects. Determining type of the defect is helpful in some extent to give information about outcomes of the fetuses but more prognostic factors are needed. Various factors such as size of the defect, presence of liver herniation, and degree of pulmonary hypoplasia were reported [6-8]. However, among these factors associated structural and chromosomal abnormalities were the most essential factors affecting outcomes of the fetuses.

In the present study, chromosomal abnormalities were found in 14.3% of omphalocele cases. Trisomy 18 was the most common chromosomal abnormality seen in fetuses with omphaloceles as stated in previous studies but other trisomies, monosomies, deletions, and triploidies were also reported [9, 10]. The rate of structural anomalies of fetuses with omphalocele was 76.7%, similar to most studies, which varied from 27% to 91% [2, 6, 11-17]. Central nervous system defects were the most common associated anomalies followed by skeletal and cardiac defects. Neural tube defects were detected in more than half of the central nervous system anomalies. There was no chromosomal abnormality in fetuses with isolated omphaloceles in the present study. In the previous studies, the chromosomal abnormalities ranged between 10.3% and 31.9% for omphalocele and were all associated with structural anomalies [3, 6, 13-16]. This finding does not mean that fetal karyotyping is not needed in isolated omphaloceles. The high rate of aneuplodies among omphalocele cases and the possibility of missing additional anomalies by ultrasonography mandate fetal karyotyping in fetuses with omphaloceles.

The necessity of prenatal karyotyping in gastroschisis cases is subject to some debate. The present authors did not detect any chromosomal abnormality in fetuses with gastroschisis as reported in some studies [6, 13-17]. On the contrary, there are studies reporting low rates of aneuploidies including trisomy 13, 18, 21, and sex chromosome abnormalities [2, 13, 18-21]. Among these studies except that performed by Barisic *et al*. [21], abnormal karyotypes were found in multiple additional anomalies. Although the present number of cases of gastroschisis were not sufficient to interpret, it seems reasonable not to offer prenatal karyotyping in isolated cases. Haddock *et al*. [22] reported that gastroschisis is not associated with major anomalies. Similarly, the anomalies of gastroschisis cases in the present study were minor (choroid plexus cyst, single umbilical artery). However, recent many studies reported the association of major anomalies with gastroschisis with a range of 5.3% - 53.2% [2, 6, 13-16, 18, 20]. The present authors believe that actual frequencies should be lower than reported due to the inclusion of minor anomalies or the ones that were consequences of gastroschisis such as intestinal atresia and malrotation.

Some congenital syndromes other than aneuploidies such as OEIS, Pentalogy of Cantrell, and Beckwith Wiedemann syndrome are more common in fetuses with omphaloceles. In the present study, of the 49 omphalocele cases, two fetuses with OEIS and one fetus with Pentalogy of Cantrell syndromes were diagnosed via autopsy findings. It is difficult to detect some syndromes in utero and certain characteristic findings such as macroglossia in Beckwith Wiedemann syndrome appear later in gestational weeks. The wide variation in expression may be the reason that Beckwith Wiedemann syndrome was not diagnosed among terminated fetuses. Gastroschisis is associated rarely with syndromes compared to omphalocele [2, 13]. Likewise, the present authors did not detect any finding suggesting part of a syndrome.

Of the fetuses with omphaloceles, 65.3% were terminated. When the present authors excluded the fetuses with abnormal karyotypes, termination rate for pregnancies with omphaloceles were still high (59.5%) due to the lethal or serious anomalies. Further, even if no prenatal karyotype had been performed, termination would have still offered to these aneuploid fetuses. The present authors found that associated structural malformations played more important role than aneuploidies in determining the pregnancy termination. For this reason, prenatal diagnosis of abdominal wall

defects should lead to a comprehensive search for associated structural anomalies. No fetuses with gastroschisis were terminated owing to the abnormal karyotype or additional anomalies. Since the prognosis was better in gastroschisis than any other abdominal wall defects, the present authors do not offer karyotyping in isolated gastroschisis. However, correct prenatal ultrasonographic detection of isolated gastroschisis cases should be more accurate than reported [13, 18].

Conclusion

When a fetus with abdominal wall defect is encountered, distinguishing the type should be the first step of the evaluation. It is also important to follow these fetuses at certain intervals in order to detect all associated anomalies. In conditions where the defect is suspicious or additional anomalies are associated, prenatal karyotype analysis should be done.

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