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Fetal intra-abdominal tumors: assessment of spectrum, accuracy of prenatal diagnosis, perinatal outcome and therapy at a tertiary referral center

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ABSTRACT

Objective: To describe the varieties and ultrasound characteristics of prenatally diagnosed fetal abdominal tumors and to scrutinize the accuracy of prenatal diagnosis as well as the postnatal outcome and therapy of affected pregnancies.

Study design: Retrospective study of 354 fetuses found to have abdominal tumors on prenatal sonogram, identified from 1993 to 2009 at a tertiary referral center for prenatal medicine. The cohort was classified into subgroups according to the sonographic appearance of the fetal tumor and the affected anatomic structure (urinary, gastrointestinal and genital tracts and other locations). Sensitivity, specificity, positive predictive value and false-positive rate of ultrasonography in identifying the system of origin were calculated. Relationships between relevant outcome domains and the different subgroups were assessed using the chi-square test and Fisher's exact test.

Results: Our cohort comprised 222 urinary tract lesions, 37 genital tract lesions, 80 gastrointestinal lesions and 15 tumors of other origins. The mean gestational age at diagnosis was 26 + 0 wks. The prenatally established diagnosis was exactly concordant with postnatal findings in 88.9%. Sensitivity, specificity, positive predictive value and false-positive rate of ultrasonography in identifying the system of origin (urinary, gastrointestinal, genital tracts and other locations) were 98.3%, 97.6%, 92.6% and 2.4%, respectively. The favorable postnatal outcome rate was highest among fetuses with genital tract lesions (95%) and lowest among those with tumors of the urinary tract (62%, $p = < 0.001$). Twenty per cent of tumors regressed spontaneously, mostly gastrointestinal tumors (36%, $p = < 0.001$). In 75/354 cases (21%) the parents opted to terminate the pregnancy: intra-uterine fetal demise and neonatal death were each noted in 4%. Prenatal therapy was performed in 24 of 354 cases (7%) and postnatal surgery in 64 cases (18%).

Conclusion: The majority of fetal abdominal anomalies were accurately diagnosed and the vast majority of affected fetuses had a favorable outcome, some tumors even resolved with advancing pregnancy. Pre- and post-natal invasive surgical interventions were mandatory in only a small number of cases.

1. Introduction

Fetal tumors are rarely diagnosed at prenatal ultrasound (US) examinations [1]. The fetal abdomen is one of the most common sites for fetal tumors, besides the heart or the face and neck regions [2,3]. The diagnosis of a fetal intra-abdominal mass might present a diagnostic and management dilemma because of the variety of possible differential diagnoses and the variable course during ongoing pregnancy and neonatal period [4,5]. The most challenging problem is to decide which cases will be amenable to US monitoring, postnatal surgery or even to prenatal invasive therapy. The prenatal diagnosis of an intra-abdominal anomaly is often imprecise, however, and the prediction for the need of prenatal intervention is poor [4–7]. In clinical practice, many tumors are not seen until relatively late during pregnancy and often as incidental findings in the late second or third trimester [6,8]. In recent years, however, the increasing implementation of first trimester screening and the widespread use of modern US techniques allow an early detection of fetal anomalies more often in the first and early second trimesters [9,10].

The literature about this issue consists mainly of relatively small case series [6,8]. We therefore aimed to analyze the data of a large number of fetal intra-abdominal anomalies detected prenatally at a tertiary referral center, focussing on the diagnostic

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Table 1a
Details about the fetal outcome in case of prenatal diagnosis of a fetal abdominal tumor, rate of associated anomalies and results of karyotyping.

Location	Total (% of total)	Outcome				Associated anomalies (%)	Karyo-typing (%)	Abnormal karyotype (%)
		Alive and well (%)	TOP (%)	IUFD (%)	Post partum or neonatal death (%)			
Urinary tract	222(62.7)	137(61.7)	67(30.2)	9(4.0)	9(4.0)	102(45.9)	124(55.9)	17/124(13.7)
Genital tract	37(10.5)	35(94.5)	0	0	2(5.4)	4(10.8)	8(21.6)	1/8(12.5)
Gastro-intestinal tract	80(22.6)	67(83.8)	6(7.5)	4(5.0)	3(3.8)	33(41.3)	38(47.5)	7/38(18.4)
Others	15(4.2)	12(80.0)	2(13.3)	0	1(6.7)	2(13.3)	3(20.0)	0/3
Total	354	252(71.2)	75(21.2)	13(3.7)	15(4.2)	141(39.8)	173(48.9)	25/173(14.5)

TOP: Termination of pregnancy.
IUFD: Intrauterine fetal demise.

accuracy, as well as the outcome and therapy of affected pregnancies. This may help to improve the parental multidisciplinary counseling and management.

2. Methods

This was a retrospective observational study including 354 fetuses that were prenatally identified with an abdominal tumor out of a total of 44,560 pregnancies seen at our tertiary referral center of obstetrics and prenatal medicine for targeted US examination from 1993 to 2009. Approval from our institutional review board was obtained before data collection. We retrospectively analyzed the gestational age at diagnosis, the accuracy of the prenatal sonogram, the antenatal course of affected individuals and the outcome. For this purpose, the prenatal US findings, autopsy reports, histological diagnosis, and the neonatal and pediatric charts were reviewed for a medium follow-up period of 10 months (ranging from 1 week after birth to 4 years of life).

The cohort was classified into subgroups according to the sonographic appearance of the fetal tumor and the affected anatomic structure (urinary, gastrointestinal, genital tracts and other locations). Since a widely accepted classification concerning the US features of fetal abdominal tumors does not exist, we classified our results as proposed by Hyett and Thilaganathan et al. [6,7].

We categorized the primary outcome parameters as: (1) the pregnancy outcome (live-birth with neonate well-being, pregnancy termination (TOP), intrauterine fetal death (IUFD) and neonatal death), (2) the natural course of the fetal abdominal tumor (regression, persistence/progress), (3) the need for invasive therapy in the prenatal or neonatal period, and (4) the concordance of the prenatal diagnosis with the postnatal pathological or pediatric findings.

Secondary outcome parameters were the presence of associated structural and chromosomal anomalies and the distribution of fetal gender.

All US examinations were performed by experienced sonographers who had expertise in prenatal US using an Acuson 128 XP (Siemens, Erlangen, Germany), an ATL HDI 5000 (Philips, Solingen, Germany) or a GE Voluson 730 Expert (GE Healthcare, Milwaukee, USA). The autopsies and histological examinations were performed by experienced pathologists.

The data were analyzed using SPSS 17.0 for Windows (SPSS INC. 2008, Chicago, IL, USA). Sensitivity, specificity, positive predictive value and false-positive rate of ultrasonography in identifying the system of origin were calculated. Relationships between relevant outcome domains and the named subgroups were assessed using the chi-square test and Fisher's exact test. The alpha level was set at 0.05. The evaluation of different subgroups concerning the gestational age at diagnosis was determined by using the ANOVA group comparison technique and the Duncan's multiple-range procedure.

Cases which were examined by prenatal US only once during pregnancy were excluded from the calculation of the tumor remission and persistence rate because of lack of information. The concordance rate of the prenatal diagnosis with postnatal findings could not be evaluated for cases with spontaneous prenatal tumor remissions (20.1%) and loss of follow-up (13.8%).

3. Results

A total of 354 fetuses with prenatally detected abdominal tumors were enrolled in this study. Our cohort comprised 222 lesions of the urinary tract, 37 genital tract lesions, 80 gastrointestinal lesions and 15 tumors of other origin (Tables 1a, 1b and 2). The vast majority of tumors were of cystic nature (332/354, 93.8%), and only 6.2% showed a solid structure (22/354). Figs. 1–3 show the variety of prenatal findings and their corresponding US features.

The mean gestational age at diagnosis was 26 + 0 wks, ranging from 11 + 3 to 40 + 5 wks (Table 3). With regard to the affected anatomic structure the analysis with one-way ANOVA indicated a statistically significant difference between the subgroups ($F_2 = 16.15$; $p < 0.001$). Further analysis using Duncan's multiple-range procedure revealed that the prenatal diagnosis of tumors of the urinary tract was made significantly earlier than those in the

Table 1b
Details about the fetal outcome in case of prenatal diagnosis of a fetal abdominal tumor, prenatal course and therapy dependent on the affected anatomic structure.

Location	Total (% of total)	Persistence during pregnancy (%[a])	Regression during pregnancy (%[a])	Single prenatal examination (=cases excluded)[a]	Antenatal therapy (%)	Postnatal surgery (%)
Urinary tract	222(62.7)	164(86.3)	26(13.7)	32	9(4.0)	24(10.8)
Genital tract	37(10.5)	21(72.4)	8(27.5)	8	3(8.1)	2(5.4)
Gastro-intestinal tract	80(22.6)	47(64.4)	26(35.6)	7	2(2.5)	36(45.0)
Others	15(4.2)	10(90.9)	1(9.0)	4	0	2(13.3)
Total	354	242(79.9)	61(20.1)	51	24(6.8)	64(18.0)

[a] When an affected fetus was examined only once during pregnancy it was excluded from the analysis of the natural course during pregnancy. Perinatal data were reported so that these cases could be included in the outcome analysis.