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Retrospective analysis of deliveries with congenital anomalies at a tertiary center

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Abstract

Objective: The purpose of this study is to evaluate the incidence, anomaly type and distribution of pregnancies which were found to have congenital anomaly during prenatal period in our regions.

Methods: In this study, congenital fetal anomalies detected via antenatal ultrasonograpgy and/or postnatal examination between January 2009 and December 2012. Congenital anomaly diagnoses were established by hospital ultasonography, decision records of ethic board for delivery and fetal termination, and postnatal examination. It was found that totally 8286 deliveries occurred during the study. Age, gravida, parity, abortus, living child number, weeks of gestation during diagnosis, system anomalies and sub-types seen in fetus were evaluated as maternal demographic data. Minor anomalies (such as choroid plexus cyst, hyperechogenic bowel, short femur, hyperechogenic cardiac focus, moderate pyelectasis etc.) were excluded from the study.

Results: During the study, major congenital anomaly was found in 485 cases where the incidence rate was 5.85%. While single system anomaly was found in 460 cases, multiple system anomalies were seen in 25 cases. The most common single system anomaly detected was central nervous system anomaly (62.1%).

Conclusion: We believe that the extensive use of advanced ultrasonography devices, pregnant women becoming more aware about routine antenatal care and the increase of the number of experienced gynecologists have been improving the detection rate of congenital anomalies.

Key words: Congenital anomaly, prenatal diagnosis, ultrasonography.

Tersiyer bir merkezde konjenital anomalili doğumların retrospektif analizi

Amaç: Bu çalışmanın amacı bölgemizde prenatal dönemde saptanan konjenital anomalili gebeliklerin insidans, anomali tipi ve dağılımlarını değerlendirmektir.

Yöntem: Bu çalışmada Ocak 2009 - Aralık 2012 tarihleri arasında kliniğimizde antenatal ultarasonografi ve/veya doğum sonrası muayene ile saptanan konjenital fetal anomaliler değerlendirildi. Konjenital anomali tanıları hastane ultrasonografi, doğum ve fetal terminasyon etik kurul kararı kayıtları ve postnatal inceleme sonucu konuldu. Çalışma süresinde 8.286 doğum gerçekleştiği saptandı. Maternal demografik veriler olarak yaş, gravida, parite, abortus, yaşayan çocuk sayısı, tanı esnasındaki gestasyonel hafta, fetusta görülen sistem anomalileri ve alt tipleri değerlendirildi. Minör anomaliler (koroid pleksus kisti, hiperekojen bağırsak, kısa femur, hiperekojenik kardiak odak, orta düzeyde renal piyelektazi gibi) çalışmaya dahil edilmedi.

Bulgular: Çalışma süresince 485 olguda majör konjenital anomali olduğu tespit edildi ve insidansı %5.85 olarak bulundu. Tek sistem anomalisi 460 olguda saptanırken, 25 olguda multipl sistem anomalisi izlendi. Tek sistem anomalilerinde en sık tespit edilen merkezi sinir sistem anomalisi idi (%62.1).

Sonuç: Gelişmiş ultrasonografi cihazlarının yaygın olarak kullanımı, gebelerin rutin antenatal bakım konusunda giderek daha bilinçli olması ve deneyimli kadın doğum hekim sayısının artması konjenital anomalilerin tespit oranını arttırdığını düşünmekteyiz.

Anahtar sözcükler: Konjenital anomali, prenatal tanı, ultrasonografi.

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Introduction

Congenital anomaly is a development defect existing during birth and defining abnormal deviation from standard type during morphogenesis on a part of structure, type and functions of fetus.[1] Major structural anomalies detected in 2-3% of all newborns are seen as the most common second reason of perinatal mortality and morbiditiy after preterm deliveries.[2,3] Fetal structural anomalies may be seen as organogenesis defects where isolated organs are affected as well as blastogenesis defects which affect the large part of the body.[4] No reason can be detected frequently in the etiology of cases with congenital anomaly. Nevertheless, maternal systemic diseases, environmental agents, alcohol, smoking, addictive substances, toxic drugs taken during pregnancy, radiation, maternal infections, deterioration of placental blood flow and perinatal infections are also the factors considered as congenital anomaly as well as the genetic factors blamed in the etiology.[5]

The possibility of terminating pregnancies up to 10 weeks in addition to advanced imaging methods and other prenatal diagnosis tools have increased the expectancy of delivering healthy babies without any malformation.[6] Together with the developments in imaging technology, the increase in the experience of obstetricians for scanning antenatal fetal anomaly has helped to increase the detection rate of congenital anomaly.[7] Detecting fetus with major congenital anomaly is one of the major aims of antenatal care. Antenatal should be evaluated in detail in case of the presence of delivering baby with anomaly in the history, kin marriage, maternal systemic disease, baby with anomaly or habitual abortus history. Our clinic provides tertiary healthcare service where patients in Southeastern Anatolia are frequently referred with the presence/suspicion of fetus with congenital anomaly.

The aim of our study is to present the frequency, types, systems and distributions of cases diagnosed with congenital anomaly in our region.

Methods

In our study, cases with congenital major fetal anomaly detected between January 2009 and December 2012 in the Department of Obstetrics & Gynecology, Faculty of Medicine, Dicle University were evaluated retrospectively. Congenital anomaly diagnoses were established by analyzing hospital ultrasonography records, and the decision records of ethic board for delivery and fetal termination. It was found that totally 8286 deliveries were carried out during the study. Age, gravida, parity, abortus, living child number, weeks of gestation during diagnosis, system anomalies and types seen in fetus were evaluated as maternal demographic data. Ultrasonographic examinations of cases included to the study were carried out by Voluson 730 PRO (General Electric Healthcare, Milwaukee, WI, USA). Minor anomalies (such as choroid plexus cyst, hyperechogenic bowel, short femur, hyperechogenic cardiac focus, moderate pyelectasis etc.) detected were excluded from the study. After obtaining the approval of ethics board and by the consent of family, the termination is carried out to pregnants in our clinic which have severe sequel risk at less than 24 weeks of gestation. The approval of local ethic board of Dicle University was obtained for the study.

Results

It was found that 8286 deliveries and terminations were carried out in our clinic during the four years of period and 485 cases among them had major congenital anomaly. We found the incidence of congenital anomaly as 5.85% in our study. While congenital anomaly was found in a single system in 460 cases, it was found that the anomaly was in more than one system in 25 cases. When the distribution of cases found to have anomaly in a single case was analyzed, it was seen that the most common anomaly was central nervous system (CNS) anomaly in 301 cases (62.1%) (**Table 1**).

In our study, maternal demographic data such as age, gravida, parity, abortus, living child number etc. have been shown in **Table 2**. It was found that there was kin marriage in the history of 133 cases with congenital anomaly (23.3%) and 56 of the cases (11.5%) had delivery history with anomaly in their previous pregnancies. Delivery and termination records showed that 437 of 485 cases (90.1%) delivered vaginally while 48 of them

Table 1. Maternal demographic parameters.

Parameters	Mean±SD	Min.-max.
Age	28.3±6.8	14-50
Gravida	4.0±2.9	1-16
Paritu	2.5±2.7	0-15
Abortus	0.5±0.9	0-6
Living child number	2.2±2.3	0-13
Weeks of gestation	24.7±7.9	10-41

Max.: maximum, **Min.**: minimum, **SD**: standard deviation.

(9.9%) were delivered by cesarean section. The decision for vaginal or cesarean delivery was made according to obstetric indications.

The cases were separated into 3 groups according to their weeks of gestation during referral. The cases in Group 1 were at <24 weeks of gestation, the cases in Group 2 were at 24-48 weeks of gestation, and the cases in Group 3 were at >28 weeks of gestation. The distribution of the cases according to the weeks of gestation has been given in **Table 3**.

The distribution of major congenital anomalies according to the systems and the analysis of their subtypes have been given in the **Table 4**. While 25 newborns had more than one system anomaly, 460 newborns had a single system anomaly. In the group found to have anomaly in more than one system, the most common anomaly group observed was hydrocephaly + meningocele seen in 14 cases, followed by encephalocele + polycystic kidney in 8 cases, and the third common anomaly group was encephalocele + omphalocele in 3 cases. In terms of single system anomaly, the most common anomaly was CNS anomalies and the most common sub-type was anencephaly (55/301). Although cardiac anomalies are the most common anomalies seen at postpartum period worldwide, we found it as the fifth most common anomaly in our study.

Discussion

Congenital anomaly incidence is reported as 3-5% in the studies conducted in referred centers.[3,8,9] The congenital anomaly incidence both in the world and Turkey may vary depending on the races, geographical regions, socioeconomic level, environmental factors, and dietary habits. In the studies conducted in different regions in Turkey, congenital anomaly incidence was reported as 0.44% by Göynümer et al., as 0.29% by Tomatır et al., as 2% by Çakmak et al., and as 1.12% by Kurdoğlu et al.[10-13] The incidence of anomaly per pregnancy was found as 2.79% by Bayhan et al. at our center in 2000.[7] In our study that we performed during the last decade, we found anomaly incidence as 5.85%. This rate is consistent with other studies in the literature; however, it was higher than the studies conducted in different regions of Turkey. It is possible to think that the studies conducted in such centers have high rates since scientific publications are made particularly at tertiary referred hospitals and fetuses with anomalies are referred to such centers. When two studies conducted at different periods in our center are compared, we observe that anomaly incidence doubles. We attribute such increase to the common use of modern devices in parallel to developments in screening technology, awareness of patients about antenatal care and the increase of the number of experienced obstetricians compared to previous years.

Table 2. Distribution of congenital anomalies according to the systems.

System	Number of cases (n)	Case percentage (%)
Central nervous system	301	62.1
Muscle-skeletal system	24	4.9
Craniofacial anomalies	8	1.6
Genitourinary system	61	12.6
Gastrointestinal system	35	7.2
Skin-lymphatic system	47	9.7
Cardiovascular system	24	4.9
Respiratory system	3	0.6
Others	8	1.6

Depending on the anomaly types, the rate of detecting at ultrasonography varies between 22% and 55%. While the detection rate of CNS anomalies is almost 100%, this rate has been reported as 25%-65% for cardiovascular system, and more lower for cleft palate and lip.[3] As in other studies carried out in Turkey, CNS anomalies in our study are seen as the most common anomaly in 62.1% of cases.[7,10-13] In the analysis of subtypes of CNS anomalies in our study, we found that the most common anomalies were anencephaly, hydrocephaly, and ventriculomegaly. In the study carried out by Kurdoğlu et al. at Van region, anencephaly group was the most common subtype of CNS, which was similar to our study.[13]

Madi et al. reported kin marriage as 68% in the cases with congenital anomaly.[14] In our study, we found the rate of kin marriage as 23.3%. We attributed the high anomaly rate despite the low rate of kin marriage in our study to the fact that preconceptional care was not disregarded in the region, and the non-awareness that

Table 3. Distribution of the cases with congenital anomaly according to the weeks of gestation.

Weeks of gestation	Number of cases (n)	Case percentage (%)
<24	264	54.4
24-28	62	12.8
>28	159	32.8
Total	485	100

Table 4. Distribution according to systems and subtype analysis of congenital anomalies.

Fetal system	Anomaly type	Detected at <24 weeks of gestation	Detected between 24 and 28 weeks of gestation	Detected at >28 weeks of gestation
Central nervous system (n=301)	Anencephaly	34	14	7
	Hydrocephaly	32 (Meningocele was found in 14 cases)	14	6
	Ventriculomegaly	24	12	11
	Meningomyelocele	38	4	2
	Bifid spine	19	12	7
	Encephalocele	18 (Polycystic kidney was found in 8 cases + omphalocele was found in 3 cases)	4	3
	Acrania	24	0	0
	Arnold-Chiari type-2	6	2	0
	Dandy-Walker syndrome	2	2	1
	Holoprosencephaly	3	0	0
Muscle-skeletal system (n=24)	Extremity anomalies	1	4	2
	Kyphoscoliosis	1	4	1
	Syndactylia, polydactylia	0	2	3
	Fokomelia	0	1	2
	Tanatophoric dysplasia	0	1	2
Craniofacial anomalies (n=8)	Cleft palate-lip	0	1	3
	Cleft lip	0	1	1
	Cleft palate	0	0	2
Genitourinary system (n=61)	Polycystic kidney	11	4	7
	Multicystic kidney	6	4	3
	Megacystitis	7	2	4
	Posterior urethral valve (PUV)	4	1	0
	Hypospadias-epispadias	0	1	2
	Renal agenesia	2	0	0
	Extrophia vesica	2	0	0
	Inguinal hernia	0	0	1
Gastrointestinal system (n=35)	Gastroschisis	14	4	6
	Omphalocele	4	2	2
	Imperforate anus	0	1	1
	Tracheoesophageal fistula	0	1	0
Skin-lymphatic system (n=47)	Cystic hygroma	24	1	1
	Non-immune hydrops	12	6	3
Cardiovascular system (n=24)	Ventricular septal defect	3	2	1
	Cardiomegaly	2	1	1
	Dextrocardia	0	1	2
	Fallot's tetralogy	1	1	1
	Hypoplastic left heart	0	0	2
	Transposition of the great arteries (TGA)	0	1	1
	Exstrophy cordis	2	0	0
	Tricuspid atresia	1	0	1
Thoracic-respiratory system anomalies (n=3)	Adenoid cystic malformation	0	1	2
Other (n=8)	Diaphragmatic hernia	2	1	2
	Amniotic band syndrome	1	1	0
	Conjoined twins	1	0	0

CNS anomalies could be prevented by folic acid supplementation.

Although cardiac anomalies are the most common anomaly type worldwide, they are also the most overlooked anomaly group during ultrasonographic examination. While most commonly observed congenital anomalies are caused by cardiac reasons, we observed them as the fifth most common anomalies in our study. In terms of their subtypes, the most common anomalies were ventricular septal defect, cardiomegaly, dextrocardia and fallot's tetralogy, respectively. We believe that the rate of cardiac anomaly has a low incidence due to the difficulty of antenatal detection, narrow range of termination indication and low number of experienced pediatric cardiologist. While cardiovascular system anomaly was found as 3.4% (10/294) by Bayhan et al. at our center, it was found as 4.9 (24/485) in our study. Although the rates are still found low as there is an increase by years, we believe that such anomalies are overlooked during screening programs, it is difficult to detect minor cardiac defects and such cases do not apply to our clinic during their pregnancies as our clinic is a reference center.

In terms of the relationship between maternal age and congenital anomaly in the studies performed in different regions of Turkey, it is seen that the cases with anomaly are mostly between 21 and 30 years old.[12,13,15] Mean age of the cases in our study was 28.34±6.76 and it was found similar with other studies. The regular and cesarean delivery rates in fetuses with our study was found as 90.1% and 9.9%, respectively in our study, and these rates are consistent with other studies carried out in Turkey.[10-13,15-17]

Conclusion

In conclusion, we believe that the common use of advanced ultrasonography devices, awareness of pregnants about routine antenatal care, and the increased number of experienced gynecologists increase the detection rate of congenital anomalies. It is considered to be more appropriate to carry out society- and nation-based studies instead of hospital-based studies in order to detect congenital anomaly incidence.

Conflicts of Interest: No conflicts declared.

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