Evaluation of Fetal Congenital Anomalies After 23 Weeks of Gestation: Clinical Analysis of 444 Cases

Kazım GEZGİNÇ[1], Rengin KARATAYLI[1], Fatma YAZICI[1], Mehmet Sait İÇEN[1], Ali ACAR[1]

Konya, Turkey

OBJECTIVE: In this study it is objected to identify the frequency and distribution of congenital anomalies in babies born at Selçuk University Meram Medical School, Department of Obstetrics and Gynecology.

STUDY DESIGN: Fetal congenital anomalies detected after 23 weeks' of gestation from January 2008 to December 2010 were retrospectively reviewed. Fetal congenital anomalies were diagnosed prenatally or after birth.

RESULTS: A total of 444 infants were diagnosed to have congenital anomalies among 7032 live and deceased newborn infants delivered between January 2008-December 2010. The mean maternal age was 27.84±6.48 years (range 15-47 years). The average gestational age at delivery was 35.32±4.5 weeks (range 22-43 weeks). Among all births, 106 infants (23.9%) had nervous system anomalies, 77 (17.3%) had multiple congenital anomalies, 68 (15.3%) had cardiovascular system anomalies, 66 (14.9%) had digestive system anomalies, 45 had (10.1%) urogenital system anomalies, 33 (7.4%) had hydrops fetalis, 17 (3.8%) had chromosomal anomalies, 13 (2.9%) had respiratory system anomalies, 12 (2.7%) had musculoskeletal system anomalies.

CONCLUSION: Most of our patients preferred to continue pregnancies with congenital anomalies because of sociocultural and religious beliefs. So, reducing the incidence of congenital anomalies by controlling environmental risk factors and preconceptional care are of great importance.

Key Words: Congenital anomalies, Antenatal care, Prenatal diagnosis

Gynecol Obstet Reprod Med 2011;17:142-145

Introduction

A congenital anomaly is an abnormality of structure, function or metabolism present at birth that results in physical or mental disability, or is fatal. Several thousand different birth defects have been identified. Congenital anomalies may be inherited or sporadic, isolated or multiple, apparent or hidden, gross or microscopic.[1]

Some risk factors for congenital anomalies include fetal factors, such as plurality and sex, and parental factors, such as ethnicity, socioeconomic status, lifestyle (including tobacco and alcohol consumption, drug use, and medication during pregnancy), age, body weight, congenital diseases, and environmental exposure (including air and drinking water pollutants, organic solvents, electromagnetic radiation, and biological agents).[2]

Early diagnosis of fetal abnormalities, usually performed at 14-16 weeks of gestation generally by a transvaginal scan, has been the subject of intensive investigation in recent years.[3] Congenital heart defects (CHD) were the most common non-chromosomal subgroup, followed by limb defects, anomalies of urinary system and nervous system defects.[4]

The subject of legal termination of pregnancy (TOP) covers many aspects of life (religious, philosophic, social, medical and ethical).[5] Early scanning and screening along with a wide use of rapid molecular genetic tests for fetal karyotyping and an earlier cardiac scan may contribute to achieving late TOP.

In this study it was objected to identify the frequency and distribution of congenital anomalies in babies born at Selçuk University Meram Medical School, Department of Obstetrics ond Gynecology.

Material and Method

Cases of fetal congenital anomalies after 23 weeks' gestation in our clinic from January 2008 to December 2010 were

[1]Selcuk Unıversıty Meram Medıcal School Department Of Obstetrıcs And Gynecology, Konya

Address of Correspondence: *Rengin Karatayl*
Selcuk University Meram Medical School Department of Obstet. And Gynecol. Akyokuş, Konya
renginkarataylı@hotmail.com

Submitted for Publication: *17. 09 2011*
Accepted for Publication: *12. 12. 2011*

retrospectively reviewed. Fetal congenital anomalies were diagnosed prenatally or after birth. Genetic counseling was given to all parents with the diagnosis of fetal anomaly. The study group consisted of pregnant women who did not accept termination of pregnancy or who had anomalies detected late in gestation. Our medical center serves as a tertiary referral center for pregnant women whose fetuses are suspected to have severe abnormalities. Further information was obtained from the women's medical and demographic records, fetal sex, birth weight, gestational age at diagnosis and delivery, type of anomaly, origin of anomaly were all recorded.

Descriptive parameters are expressed as mean ±SD. Frequencies are given as percentages. Calculations were performed using SPSS software Version 16.0 (SPSS, Inc., Turkey). All anomalies are coded using the International Classification of Disease version 10 (ICD 10).

Results

A total of 444 infants were diagnosed with congenital anomalies among 7032 live and deceased newborn infants delivered between January 2008-December 2010. Out of these, 231 infants (46.6%) were male, 207 (52%) were female and 6 infants (1.4%) had indeterminate sex. 53 fetuses (11.9%) with fetal anomalies deceased on follow-up or were in utero exitus at admission. 429 cases (96.6%) were singleton pregnancies, 10 cases (2.3%) were twin pregnancies, and 5 cases (1.1%) had triplet pregnancies. The mean birth weight was 2508.08±886.70 grams (500-5000 grams). The mean maternal age and parity were 27.84±6.48 years (range 15-47 years) and 2.02±1.28 (range 0-7) respectively. Most of the patients aged between 18-35 (359/444, 80.9%), there were 16 patients (3.6%) aged below 18, and 39 pregnant women (15.6%) aged over 35 years. The average gestational age at delivery was 35.32±4.5 weeks (range 22-43 weeks). Most of the gestations reached to term and birth at term consisted 43.9% of cases. Among all births, 106 (23.9%) had nervous system anomalies, 77 (17.3%) had multiple congenital anomalies, 68 (15.3%) had cardiovascular system anomalies, 66 (14.9%) had digestive system anomalies, 45 (10.1%) had urogenital system anomalies, 33 (7.4%) had hydrops fetalis, 17 (3.8%) had chromosomal anomalies, 13 (2.9%) had respiratory system anomalies, 12 (2.7%) had musculoskeletal system anomalies. The distribution of individual congenital anomalies is demonstrated at table 1. Hydrocephalus (35/106, 33%) was the single most common CNS anomaly. Combined CNS anomalies composed the second most frequent anomalies (26/106, 24.5%). Multiple anomalies were the second most common congenital anomalies, and the third frequent anomalies consisted of cardiovascular system anomalies and the most common cardiac anomaly was isolated ventricular septal defect (VSD) (22/68, 32.4%). Chromosomal disorders were detected in 33 patients, and out of these 17 (51.5%) did not have any structural anomalies. Chromosomal disorders are summarized at table 2. 16 patients with chromosomal disorders (48.5%) had multiple congenital anomalies. 13 patients (39.4%) with chromosomal disorders aged above 35. The most common chromosomal anomaly detected was Down Syndrome (26/33, 78.8%). 14 cases with Down Syndrome (42%) had cardiovascular system anomalies and 13 cases (50%) had multiple anomalies.

Discussion

Congenital anomalies account for approximately one-third of infant deaths and one-quarter of neonatal deaths.[1]

Most studies show that the detection rate of such scans is approximately 60-87% (Yagel el at. 1995) compared with the routine second trimester scan .[3]

Mendelian factors are known to account for about 20% of human congenital diseases, and multifactorial polygenic influences are blamed for another 15%. Ten percent are believed to result from environmental causes (toxins, radiation, diet, drugs, infection and metabolic disorders), and 5% are associated with aneuploidy. The remaining 50% result from unknown causes.[1,6]

In Vaknin et al[5] study, assessment of the affected fetal organs in which there were no underlying chromosomal/genetic abnormalities or fetal infection revealed that over 50% were CNS and cardiac abnormalities. Hydrocephalus was the single most common CNS anomaly in their series. In our study, among all births, 106 cases (23.9%) had nervous system anomalies, 77 (17.3%) had multiple congenital anomalies, 68 (15.3%) had cardiovascular system anomalies and hydrocephalus was the single most common CNS anomaly. The reason why isolated hydrocephaly is the single most common anomaly in our study may be due to the decision of the families in order not to terminate pregnancies since this anomaly itself is not accepted as a significant anomaly and reason for termination.

In our study, we evaluated cases that were diagnosed after 23 weeks' of gestation, those who did not accept termination of the pregnancy or those that were diagnosed after birth. As a result of our study, we detected that sociocultural and religious beliefs play an important role in the management of pregnancies with anomalies, so in populations arguing termination of pregnancies with anomalies, prevention of fetal anomalies is more important than prenatal diagnosis. According to our study results, 43.9% of fetuses with fetal anomalies were born at term, on the other hand 37.7% of multiple fetal anomalies that were incompatible with life were born between 34-37 weeks of gestation, and 36.4% were born at term.

When a fetal structural anomaly is identified, the pregnant woman should be offered a timely consultation with a trained genetic counselor and with a maternal-fetal medicine special-

ist and/or a medical geneticist. The counseling should be unbiased and respectful of the patient's choice, culture, religion, and beliefs.[7] Parents should be informed that major or minor fetal structural anomalies, whether isolated or multiple, may be part of a genetic syndrome, sequence, or association, despite a normal fetal karyotype. At our clinic, all patients with fetal anomalies were administered genetic counseling, and later on are asked for their decision on termination or the continuation of the gestation. In our country, most of our patients prefer to continue pregnancies with congenital anomalies because of sociocultural and religious reasons. So reducing the incidence of congenital anomalies by controlling environmental risk factors and preconceptional care is of great importance. Further investigation on more detailed risk factors is warranted to elucidate causal factors, and to prevent the occurrence of congenital anomalies.[2]

If it is important to conduct surveillance of congenital anomalies to look for associations with potential environmental teratogens, to support health service planning, and to monitor prenatal diagnosis and screening programme then ascertainment of defects at national level must be improved.[8]

Table 1: The distribution of individual congenital anomalies

System Involved	Anomaly	Cases	
		No	(%)
Central Nervous System	*Hydrocephalus	35	
	*Anencephalus	9	
	*Ventriculomegaly	8	
	*Microcephaly	7	
	*Meningomyelocele	6	
	*Combined	26	
	*Other[1]	15	
	Total	**106**	**(23.9)**
Cardiovascular System	*VSD	22	
	*Hypoplastic left heart	5	
	*Transposition of Great Vessels	5	
	*Aortic coarctation	4	
	*Tetralogy of Fallot	3	
	*Ebstein's anomaly	3	
	*Combined cardiac anomalies	12	
	*Other[2]	10	
	Total	**68**	**(15.3)**
Gastrointestinal System	*Gastroschisis	14	
	*İntestine Atresia	10	
	*Cleft Lip With/Without Palate	10	
	*Diaphragmatic Hernia	6	
	*Omphalocele	5	
	*Tracheo-Oesophageal Fistula, Oesophageal Atresia And Stenosis	5	
	*Cleft Palate	3	
	Total	**66**	**(14.9)**
Genitourinary System	*Hydronephrosis	20	
	*Posterior Urethral Valve	7	
	*Bilateral Renal Agenesis	4	
	*Multicystic Dysplastic Kidney	4	
	*Polycystic Kidney Disease	4	
	*Other	6	
	Total	**45**	**(10.1)**
Respiratory System	*Choanal Atresia	7	
	*Hydrothrax	3	
	*Congenital Cystic Adenomatoid Malformation	2	
	*Lung Agenesis	1	
	Total	**13**	**(2.9)**
Skeletal System	*Bilateral Clubfoot	3	
	*Osteogenesis İmperfecta	2	
	*Achondroplasia	2	
	*Combined Anomalies	3	
	*Other[3]	3	
	Total	**12**	**(2.7)**
Hydrops Fetalis	**Total**	**33**	**(7.4)**
Multiple Malformations	**Total**	**77**	**(17.3)**
Chromosomal-genetic abnormalities[4]	**Total**	**17**	**(3.8)**
Others[5]	**Total**	**7**	**(1.6)**

[1] Other CNS stands for: 7 cases of cystic higroma, 2 cases of meningocele, 2 cases of encephalocele, a case of acrania, a case of lissencephaly. [2] Other cardiovascular system stands for: 2 cases of truncus arteriosus, 2 cases of total pulmonary venous return anomaly, 2 cases of ectopia cordis, a case of rhabdomyoma of the heart. [3] Other a case of arthrogryposis. [4]Cases without any structural abnormality [5]Others : Endocrine and Metabolic Disorders.

Table 2: Distribution of Chromosomal-genetic abnormalities

Chromosomal-genetic abnormalities[1]	No	%
Trisomy 21	26	
Trisomy 18	2	
Trisomy 13	2	
Triple X Syndrome	1	
Other	2	
Total	**33**	**(7.4%)**

[1] Chromosomal-genetic abnormalities with and without structural anomalies

Yirmiüçüncü Gebelik Haftasından Sonraki Fetal Konjenital Anomalilerin Değerlendirilmesi: 444 Vakanın Klinik Analizi

AMAÇ: Bu çalışmada amaç, Selçuk Üniversitesi Meram Tıp Fakültesi Obstetri kliniğinde konjenital anomali ile doğan bebeklerde anomali sıklıklarını ve dağılımını tanımlamaktır.

GEREÇ VE YÖNTEM: Ocak 2008 ve Aralık 2010 tarihleri arasında 23. Gebelik haftasından sonra tespit edilen fetal konjenital anomaliler retrospektif olarak tarandı. Konjenital anomaliler prenatal olarak veya doğum sonunda tespit edildi.

BULGULAR: Ocak 2008 ve Aralık 2010 tarihleri arasında gerçekleşen toplam 7032 canlı ve ölü doğum arasında toplam 444 bebek konjenital anomalili olarak tespit edildi. Ortalama anne yaşı 27,84±6,48 (15-47 yıl) idi. Doğumdaki ortalama gestasyonel yaş 35.32±4.5 hafta idi (22-43 hafta). Tüm doğumlar arasında , 106 bebekte (%23,9) santral sinir sitemine ait anomaliler, 77 bebekte (%17,3) multipl konjenital anomaliler, 68 bebekte (%15,3) kardiyovasküler system anomalileri, 66 bebekte (%14,9) sindirim sitemine ait anomalier, 45 hastada (%10,1) ürogenital sisteme ait anomaliler, 33 bebekte (%7,4) hidrops fetalis, 17 bebekte (%3,8) kromozomal anomaliler, 13 bebekte (%2,9) respiratuar sisteme ait anomaliler, 12 bebekte de (%2,7) kas-iskelet sistemine ait anomaliler tespit edildi.

SONUÇ: Konjenital anomalili bebeğe sahip ailelerin çoğu sosyokültürel faktörlerden ve dini inanışlarından dolayı gebeliklerini devam ettirdiler. Bu nedenle, çevresel faktörlerin kontrol edilmesi ve prekonsepsiyonel bakım konjenital anomalilerin insidansını azaltmada büyük önem taşımaktadır.

Anahtar Kelimeler: Konjenital anomali, Antenatal bakım, Prenatal tanı

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