

Distribution and Incidence of Congenital Malformations in a University Hospital

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Abstract

Objective: It is aimed to determine the incidence and types of congenital anomalies among all neonates in the Department of Obstetrics and Gynecology, Gazi University Faculty of Medicine, between 1988-2005.

Methods: Registries of 17.259 neonates were studied retrospectively. Total malformation incidence, types of these malformations, percentages of isolated and multiple anomalies and their distribution according to gender and maternal age were determined.

Results: 203 anomalies and a 1.18% incidence of congenital malformation was detected. Risk of any anomaly among male fetuses was 1.21% and it was 1.15% among females; and no difference was detected ($p>0.05$). Most common anomaly was meningocele. It was followed by other central nervous system anomalies such as anencephaly and hydrocephaly. 70% of anomalies were isolated and rest were multiple. Risk of having a fetus with congenital malformation varies with age and is most common under 20 and over 40.

Conclusion: Overall congenital anomaly incidence in newborns of our population is 1.18%. Most common anomaly is meningocele, followed by other central nervous system anomalies such as anencephaly and hydrocephaly.

Keywords: Congenital malformation, anomaly.

Bir üniversite hastanesinde konjenital malformasyonların görülme sıklığı ve dağılımı

Amaç: Gazi Üniversitesi Kadın Hastalıkları ve Doğum Anabilim Dalı'nda, 1988-2005 yılları arasında gerçekleşen doğumlarda konjenital anomali tiplerinin ve insidansının belirlenmesi amaçlanmıştır.

Yöntem: Retrospektif olarak incelenen 17.259 doğumda toplam malformasyon sıklığı, bu malformasyonların tipleri, izole ve kombine olarak görülme oranları, anne yaşına göre ve cinsiyete göre dağılımları belirlenmiştir.

Bulgular: Tespit edilen toplam 203 anomali, incelediğimiz popülasyonda, konjenital malformasyonlu fetüs doğma oranının %1.18 olduğunu göstermiştir. Erkek çocuklarda herhangi bir konjenital malformasyon bulunma riski %1.21 ve kızlarda %1.15 olup Ki-kare testi kullanılmasıyla iki grup arasında istatistiksel olarak anlamlı fark bulunmamıştır ($p>0.05$). En sık görülen anomali meningocele olup bunu anensefali ve hidrosefali gibi diğer santral sinir sistemi malformasyonları izlemiştir. Görülen tüm malformasyonların %70'i izole iken geri kalanı multipl konjenital malformasyon olarak görülmüştür. Anomalili bir fetus doğurma riskinin yaşa bağlı olarak değiştiği ve 20 yaşından önce, 40 yaşından sonra sık görüldüğü tespit edilmiştir.

Sonuç: İncelediğimiz toplulukta konjenital malformasyon görülme sıklığı %1.18'dir. En sık görülen anomali meningocele olup bunu anensefali ve hidrosefali gibi diğer santral sinir sistemi malformasyonları izlemektedir.

Anahtar Sözcükler: Konjenital malformasyon, anomali.

Introduction

Incidence of congenital anomalies varies all over the world depending on the genetic factors like chromosome anomalies and single mutation, dietary habits leading to folic acid deficiency, smoking, alcohol and other environmental toxic agents. In the United States of America, where majority of the studies on this subject is conducted, incidence of congenital anomalies has been reported as 2-3 in each 100 delivery¹ whereas prevalence of congenital malformation is 2% in England, 1.2% in Japan and 1.49% in South Africa.²⁻³

A study conducted all over our country found a prevalence rate of 2% for congenital malformations.⁴ This figure includes only the anomalies detectable during the delivery, and it increases up to 5% especially when the renal/cardiac system anomalies that can be detected after the delivery are combined.⁴ In another study which reviewed the whole number of infants with congenital malformation who were born in 22 university hospitals in Turkey for a period of one year, the ratio of congenital malformation was found 3.65%.⁵ In the same study, it has been reported that although isolated incidence of all malformations in our population is similar to the results reported for several other countries, neural tube defects and cleft lip-palate are more frequent.⁵ Another study conducted by the Department of Obstetrics and Gynecology of Gazi University in 1996 reviewing the birth records for a total of eight years reported a prevalence rate of 1.11% for congenital malformations.⁶

Due to the significance of congenital malformation in perinatal morbidity and mortality and its various types and diverse incidences in several countries, it is important for each population, even on regional basis, to know the distribution and incidence of congenital malformations. Our objective was to determine the incidence, distribution and type of congenital malformations in our hospital, which is a tertiary healthcare provider so that approaches in screening, diagnosis and treatment can be well defined.

Methods

The registries of a total of 17.259 deliveries carried out in the Obstetrics and Gynecology Clinic of the Medical Faculty of Gazi University between

1988 and 2005 were retrospectively examined. Fetuses with antenatal or postnatal congenital malformation were determined by inspecting the birth records and patient files. Parameters included total incidence of malformations, types of the malformations, isolated and concomitant incidences, and distributions by maternal age and gender.

Throughout the follow-up period of pregnancy, triple screen test was conducted between the gestational weeks 16 and 18 as well as ultrasonographic examination at least once at each trimester in order to detect potential anomalies. Patients with an abnormality detected during the ultrasonographic evaluations or with a high risk in the dual or triple test were consulted to the experts on perinatology for detailed ultrasonic examination. If necessary, advanced examination techniques such as chorionic villus biopsy, cordosynthesis and amniosynthesis were conducted.

Results

A total of 17.259 deliveries between 1988 and 2005 was examined. Of 17,259 neonates, 8720 (50.54%) were female, and 8538 were male (49.46%). Detection of 203 anomalies in total indicated that prevalence of fetal delivery with congenital malformation was 1.18%. The distribution of those anomalies for both sexes is shown at Table 1. Based on this table, one-hundred-three of 203 anomalies were in girls and a hundred in boys. It corresponds to 50.7% of total congenital malformations in boys, and 49.3% in girls. Ratio of any congenital malformation in neonates was 1.21% for boys and 1.15% for girls, and no statistically significant difference was found between the two groups when compared with Chi-square test ($p>0.05$).

The distribution of congenital malformations in the patient groups reviewed is shown at Table 2.

Table 2 provides type and incidence of 203 congenital malformations. The figure of 203 for anomalies is not similar to the number of fetuses born with congenital malformation as a fetus can have multiple malformations. The most common anomaly was meningocele, followed by other central nervous system malformations like anencephaly and hydrocephaly. When organ system anomalies are classified in general, the most fre-

Table 1. Incidence of congenital malformation by gender.

Gender	Number	Percentage %
Male	104	50.7
Female	101	49.3

Table 2. Distribution of congenital malformations.

Anomaly	Number	Percentage %
Meningocele		25
12.19		
Anencephaly	23	11.21
Hydrocephaly	22	10.73
Cardiac anomalies	17	8.29
Renal anomalies	16	7.80
Pes equinovarus	13	6.34
Encephalocele	10	4.87
Omphalocele	7	3.41
Diaphragmatic hernia	7	3.41
Coanal atresia	7	3.41
Genital anomalies	7	3.41
Gastroschisis	6	2.92
Polidactyly	6	2.92
Hypospadias	6	2.92
Bowel dilatation	6	2.92
Cleft lip	5	2.43
Cleft palate	4	1.95
Syndactyly	4	1.95
Epispadias	4	1.95
Urachus Cyst	3	1.46
Trisomy 21	2	0.96
Trisomy 18	1	0.48
Intestinal atresia	1	0.48
Intracranial mass	1	0.48
Holoprosencephaly	1	0.48
Accessory finger	1	0.48
Total	205	100

Table 3. Distribution of congenital malformations.

	Isolated Anomalies		Multiple Anomalies	
	Number	Percentage %	Number	Percentage %
Central Nervous System	60	29.26	15	7.31
Urinary System	20	9.75	9	4.39
Musculoskeletal System	17	8.29	7	3.41
Cardiovascular System	12	5.85	5	2.43
Gastrointestinal System	10	4.87	4	1.95
Abdominal Wall Defects	7	3.41	6	2.92
Facial Defects	6	2.92	3	1.46
Genital System	5	2.43	2	0.96
Others	7	3.41	3	1.46

Table 4. Comparison of maternal age with incidence of malformation

Maternal Age	Number of anomaly	Percentage %	n (Number of patients)
< = 20	23	2.13	1,077
21 - 30	134	1.00	13,361
31 - 40	31	1.24	2,498
> 40	11	3.39	324

quent ones were the central nervous system anomalies with 82 cases, which were followed by cardiac malformations with 17 cases, renal anomalies and genital system anomalies with 16 cases respectively.

The prevalence and percentage of isolated and multiple malformations in our population are shown at Table 3.

Seventy percent of all malformations were isolated while the rest was multiple congenital malformation. It is notable that 80% of the central nervous system anomalies was isolated, where the other ratios were as follows; 69% for the urinary system, 70.8% for the musculoskeletal system, 70.5% for the cardiovascular system, 71.4% for the gastrointestinal system, 54% for the abdominal wall defects, 66.6% for the facial defect and 71.4% for the genital system anomalies.

We also evaluated the relation between maternal age and incidence of congenital malformation, and found out that fetal deliveries with congenital malformations occurred mostly between 21 and 30 years of age. In percentages, risk for delivering a fetus with anomaly varies with age, and it is more frequent before 20 years of age and after 40 years of age. The results are given at Table 4.

Discussion

Incidence of congenital malformation shows variation among populations depending on the socioeconomic status, dietary habits, geographical regions, races and environmental factors. Its ratio ranges from 1.49% to 3.2% for several countries. In a 13-year-study carried out in Australia (1983-1995), the ratio of congenital malformation was found 3.2% and the most common malformation was the hip dislocation.⁷ In Saudi Arabia, ratio of congenital malformation was found 1.7% in an analysis of 14,762 births.⁸ Seventy-five percent of these anomalies was major anomalies like anencephaly, meningomyelocele, and 25% included minor anomalies like polydactyly, urachus cyst, and the central nervous system was the most involved system. In a study conducted in our university in 1996, incidence of congenital malformation had been reported 1.11% while we found it 1.18%.

It has been reported by studies conducted so far investigating the relation between gender and congenital malformations that the distribution of anomalies had no gender difference. We also found out that gender of fetus, either boy or girl, had no impact on the incidence of congenital malformation, and distribution for both genders is similar.

References

1. Marion RW, Fleischman AR. The assessment and management of neonates with congenital anomalies. In: Evans MI, editor. Reproductive Risks and Prenatal Diagnosis. Norwalk: *Appleton and Lange* 1992;341-357
2. Robertson NRC. A manual of neonatal intensive care, 3rd edition. London: *Edward Arnold* 1993;336-340
3. Venter PA, Christianson AL, Hutamo CM, Makhura MP, Gericke GS. Congenital anomalies in rural black South African neonates-a silent epidemic? *S Afr Med J* 1995;85:15-20
4. Apak MY. Approach to genetic diseases and genetic counseling. In: Aydinli K, editor. Prenatal Diagnosis and Management. Istanbul: Perspektif Yayın, 1992: 1-18
5. Tuncbilek E, Boduroglu K, Alikasifoglu M. Results of the Turkish congenital malformation survey. *Turk J Pediatr* 1999; 41(3): 287-297.
6. Ö.Himmetoğlu, M.B.Tıraş. The incidence of congenital malformations in a Turkish population. *International Journal of Gynecology and Obstetrics* 1996; 55: 117-121
7. Riley MM, Halliday JL, Lumley JM. Congenital malformations in Victoria, Australia, 1983-95; an overview of infant characteristics. *J Paediatr Child Health* 1998;34(3): 233-40.
8. Al-Jama F. Congenital malformations in newborns in a teaching hospital in eastern Saudi Arabia. *J Obstet Gynaecol* 2001;21(6):595-598
9. Hollier LM, Leveno KJ, Kelly MA, McIntire DD, Cunningham FG. Maternal age and malformations in singleton births. *Obstet Gynecol* 2000;96(5 Pt 1):701-706.
10. Cunningham FG, MacDonald PC, Gant NF, Leveno KJ, Gilstrap III LC. Williams Obstetrics, 19th edition. Connecticut: Appleton and Lange, 1993:928-931.
11. Stevenson RE, Allen WP, Pai GS, Best R, Seaver LH, Dean J, Thompson S. Decline in prevalence of neural tube defects in a high-risk region of the United States. *Pediatrics* 2000;106(4):825-827.
12. Chung CS, Nemechek RW, Larsen IJ, Ching GH. Genetic and epidemiological studies of clubfoot in Hawaii. General and medical considerations. *Hum Hered* 1969; 19(4): 321-42.
13. Hoffmann JIE. Congenital heart disease: incidence and inheritance. *Pediatr Clin NA* 1990; 37: 25-43.
14. Carpenter WM, Cura MR, Dibbins AW. Perinatal management of ventral wall defects. *Obstet Gynecol* 1984; 64: 646-651.
15. Lowry RB, Baird PA. Familial gastroschisis and omphalocele. *Am J Hum Gen* 1982; 34: 517-518. .
16. Gorlin RJ, Cervenka J, Pruzansky S. Facial clefting and its syndromes. *Birth Defects* 1971; 7: 3-49.