The Retrospective Analysis of Major Fetal Abnormality at Deliveries

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Abstract

Objective: The aim of this study is to evaluate the distribution of the major fetal abnormalities with respect to systems and some related clinical properties during five years period.

Methods: A total number of 55493 deliveries and 56030 neonates were examined retrospectively between 2000-2004. Major fetal defects observed at delivery room were recorded and classified for the distribution of year, system, delivery route, fetal sex, and of prognosis.

Results: Single or multiple fetal abnormalities were identified in 247 cases. The incidence of major abnormality was 0.44 % at the time of delivery. The most common abnormality was that of the central nervous system (54.66%). Two third of these cases had delivered vaginally, one third of them had cesarean section. We revealed that 32% of major anomaly cases dead at the antenatal and very early postnatal period.

Conclusions: The prevalence of major fetal abnormalities was 0.44% and central nervous system abnormalities were the most frequent abnormalities at the deliveries in our clinic. We found that 32% of the cases with major abnormality dead at the antenatal or very early postnatal period. Most deliveries of the babies with major abnormality were done by vaginal route. We suppose that in order to determine realistic major abnormality rates, routine ultrasonographic scanning should be done and nationally oriented detailed records should be documented.

Keywords: Major fetal abnormality, birth records.

Doğumlarda majör konjenital anomalilerin retrospektif analizi

Amaç: Beş yıllık süre içinde saptanmış olan majör konjenital anomalilerin yıllara, sistemlere göre dağılımlarının ve bazı klinik özelliklerinin değerlendirilmesi.

Yöntem: 2000-2004 yılları içinde toplam 55493 doğum ve 56030 yenidoğan retrospektif olarak incelendi. Majör konjenital anomalilerin yıllara göre sayısı, sistemlere göre dağılımları, doğum şekli, fetus cinsiyeti ve prognoz değerlendirildi.

Bulgular: Toplam 247 olguda tek veya multipl konjenital anomali saptandı. Kliniğimizde doğum yapan gebelerde majör anomali oranı binde 4.4 olarak belirlendi. Tüm anomaliler içinde en sık görülen anomali tipi merkezi sinir sistemi anomalileri idi (%54.66). Olguların üçte ikisi vaginal, üçte biri ise sezaryen ile doğurtulmuştu. Majör anomali olgularının %32'sinin antenatal ve çok erken postnatal dönemde kaybedildiği belirlendi.

Sonuç: Kliniğimizde doğum yapan gebelerde majör anomali oranı binde 4.4 olup, merkezi sinir sistem anomalilerine daha sık rastlanmıştır. Genelde vaginal yoldan doğumun gerçekleştiği ve mortalitenin %32 olduğu saptanmıştır. Major anomalilerin gerçek oranının belirlenebilmesi için ultrasonografi taramalarının etkin olarak yapılması ve kayıtların ulusal standart bir forma işlenmesinin gerektiği düşünülmüştür.

Anahtar kelimeler: Majör fetal anomali, doğum kayıtları.

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Introduction

Congenital defects are the second causes of perinatal morbidity and mortality all over the world following the leading cause, which is premature delivery. They usually occur at a mean rate of 3-5%.[1-3] General tendency is to diagnose early, and treat if possible, or terminate those defects in an ethical/legal way, which may occur depending on several etiological factors and differ based on age, family characteristic, race and environment.[1,2] Failure to meet those criteria may lead to various social, economical and medicolegal problems. 1 It has been reported that congenital defects can only be diagnosed during the last period of gestation or during the delivery in the areas and hospitals lacking basic facilities.[4]

The aim of this study was to evaluate the distribution of the major fetal abnormalities with respect to systems and some related clinical properties during a five-years period and analyse the delivery method, fetal sex and mortality in those anomalies.

Methods

A total number of 55493 deliveries and 56030 neonates were examined retrospectively between 01.01.2000 and 31.12.2004 at the Gynecology and Obstetrics Clinic of the Göztepe Education and Research Hospital. All neonates at gestational week 22 or over 500 grams were included in the study based on their delivery records. The major congenital anomalies were separately evaluated for the following; number by years, distribution by systems, delivery method, fetal sex and perinatal prognosis. Statistical analysis was performed through quisquare test, and a p value <0.05 was considered significant.

Results

We have found that the number of single, twin, triplet, and quadruplet births was 54914, 559, 20 and 2, respectively, out of all deliveries registered in our clinic for a five-years period. Three of multiple gestations had major fetal anomaly while both babies had the same anomaly only in one case. In our series with a total number of 247 cases (0.44%) with anomaly, the distribution of major congenital anomalies by years is shown in Table 1. The anomaly incidence ranging from 0.4 to 0.5 % in the first four years decreased to the border of 0.26 % in 2004 (p <0.05).

An analysis on the distribution of congenital anomalies by system and region showed that the most frequent anomalies were of the central nervous system (Table 2), which were followed by non-immune hydrops fetalis and multiple anomalies.

Table 1. Congenital anomalies and their distribution by years.

Year	Single (n)	Multiple (n)	Baby (n)	Anomaly (n) (%)
2000	11545	131	11814	55 (0.46)
2001	11424	112	11542	58 (0.50)
2002	10427	107	10643	42 (0.39)
2003	10616	91	11802	62 (0.53)
2004	10090	140	10235	30 (0.26)
Total	**54912**	**581**	**56030**	**247 (0.44)**

Table 2. The distribution of congenital anomalies by systems.

	n	%
Central nervous system	135	54.66
Hydrops fetalis	40	16.19
Multiple anomaly	14	5.67
Craniofacial defects	10	4.05
Gastrointestinal system	10	4.05
Skeletal system	7	2.83
Urinary system	4	1.62
Cardiovascular system	3	1.21
Down syndrome	4	1.62
Genital system	2	0.80
Other (tumors, abdominal wall defects, etc)	18	7.30

Table 3. The fetal prognosis in congenital anomalies.

		Antenatal mortality		Early postnatal mortality		total mortality	
Year	Anomaly (n)	(n)	(%)	(n)	(%)	(n)	(%)
2000	55	18	(32.73)	1	(1.81)	19	(34.55)
2001	58	15	(25.86)	7	(12.07)	22	(37.93)
2002	42	12	(28.57)	3	(7.14)	15	(35.71)
2003	62	9	(14.52)	8	(12.90)	17	(27.42)
2004	30	5	(16.67)	1	(3.33)	6	(20.00)
Total	**247**	**59**	**(23.89)**	**20**	**(8.10)**	**79**	**(31.99)**

The prognosis of congenital anomalies is shown in Table 3. Fiftynine (74.68%) of the anomaly cases were lost during the antenatal period while 20 of them (25.32%) died during the early postnatal period. The mortality rate was found 31.99% in anomaly cases.

Table 4. The distribution of congenital anomalies by gender.

	Female	Male	Total
Alive	89	82	171
Dead	40	36	76
Total	**129 (%52.23)**	**118 (%47.67)**	**247**

Discussion

It has been reported that the mean prevalence of congenital anomalies is 3 to 5% in developed countries.[1,5,6] The rates reported before 1990s are usually lower in our counrty.[3,7-9] The rates in the referenced clinics are similar to the samples in the west.[4]

In cases where conditions for prenatal diagnosis are difficult, an incidence rate of congenital anomaly up to 14% has been reported in series with a high risk of anomaly.[6] An anomaly incidence between 6 and 16% was reported in the delivery of dead fetus.[8,10-12] The lower anomaly rates in our series may result from determination of anomaly rate only, but not prevalence; restricted possibilities in the prenatal diagnosis and insufficiencies in the registering system were also responsible for such lower rate.

We found that anomalies didn't significantly differ in the five-years period for which we examined, and they changed in a range from 0.50% to 0.26% in the average. We believe that prospestive studies are required to understand this tendency to decrease observed in recent years. We found that mortality was mostly seen during the antenatal period. Similar results were reported in our country.[3,4]

As seen in our series, the most frequent type of anomaly in our country is of the central nervous system.[3,4,13] Aquiar et al. reported a neural tube defect rate of 0.47%. 14 Since such type of defects do not go overlooked and they receive particular attention, they are recorded almost hundred percent, and superior to other defects. However, in series with prenatal diagnosis, multiple anomalies and anomalies of the cardiovascular system (16%) are preceeded by the anomalies of the urinary system (20%).

As seen in our series, the cardiac anomalies which are rare in our country 3,4,8 (3-10/ 10000), are recognized easier in the countries where possibilities for prenatal and postnatal diagnosis are better (60/10000), and they rate in the foreground among congenital anomalies.[6,15] It has been reported that the diagnosis is made rather through direct observation in series lacking a broad use of ultrasonography, and thus types of anomaly would be heavily morphologic in general.[4] However, it has been proposed that it is not possible to detect anomalies particularly related with internal organs in centers where the possibilities for prenatal diagnosis are restricted, as also seen with our series.[3,4,8]

Higher incidence of congenital anomalies in girls is parallel to the results of some previous studies.[3,4,7] Also, the rate of delivery methods is similar to the ones reported in previous studies.[4,16]

Compared to the developed countries, being unable to get a post-mortem examination even in the researh centers[17,18] leads to insufficiency in etiological factors and final diagnosis. Most of the cases in our series lack the findings of a post-mortem examination.

In conclusion, the rate of congenital anomaly was 0.44% for the deliveries in our clinic during a five-years period. Unfortunately some detectable malformations went unnoticed due to failure of early diagnosis particularly during the prenatal period and insufficient postnatal analysis. Lack of guidelines defining the weeks and by whom the prenatal diagnosis can be made as well as lack of a standard form for registering the anomalies and also influence of personal and organizational structures on the existing registers all over the country held us to obtain to the exact figures. Necessary steps should be taken nationwise and organizationwise in order to eliminate such insufficiencies.

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