Pattern of Congenital Anomalies at Birth

Anuja Bhalerao , Richa Garg
Department of Obstetrics and Gynaecology
NKP Salve institute of medical sciences, Nagpur,India
Email: anuja_bhalerao@yahoo.com , rich006@gmail.com

Abstract

Congenital anomalies are a major cause of stillbirths and neonatal mortality. The pattern and preva-lence of congenital anomalies may vary over time or with geographical location. The aim of this pa-per is to determine the pattern of congenital anomalies in newborns and to study maternal and peri-natal risk factors. This prospective case study was carried out in the Obstetrics and Gynaecology department of a rural tertiary Medical College and Hospital over 2 and half years. All the babies born in this hospital during this study period were included. The newborns were examined for the presence of congenital anomalies and mothers were interviewed for socio-demographic varia-bles. During the study period, 6076 babies were born, of which 84 had congenital malformations, making the prevalence 1.38%. Congenital anomalies were seen more commonly in the multipara (2.57%) in comparison with primipara (0.42%). The predominant system involved was musculo-skeletal system (36.90%) followed by central nervous system (25%), gastro-intestinal (GI) system (16.6%). Congenital anomalies were more likely to be associated with low birth weight, multiparity, maternal age (between 20 to 30 years) and consanguinity. The congenital anomalies affected signifi-cantly higher proportion of male babies (2.59%) than their female counterparts (0.75%).

Keywords

Congenital anomaly, prematurity, prevalence, risk factors

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I.   Introduction

Congenital malformations represent defects in morphogenesis during early fatal life. According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth [1] However, as per the more recent WHO fact-sheet of October 2012, congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.[2] Congenital anomalies are an important cause of neonatal mortality both in developed and developing countries. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.[3,4] It is not only a leading cause of foetal loss, but also contributes significantly to preterm birth, childhood and adult morbidi-ty along with considerable repercussion on the mothers and their families. Worldwide surveys have shown the birth prevalence of congenital anomalies varies due to social, racial, economical and eco-logical influences. In order to decrease the incidence of various congenital anomalies, it is important to identify their prevalence in the society and the risk factors involved.

II.   Patients and Methods

This hospital based prospective case study was carried out in the Obstetrics and Gynaecology de-partment of a rural tertiary Medical College and Hospital during the period of May 2013 to Decem-ber 2015. All the babies born in this hospital during this period were included. The newborns were examined for the presence of congenital anomalies and mothers were interviewed for socio-demographic variables. The newborns were examined meticulously and assessed systematically for the presence of congeni-tal anomalies. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the pediatrician and other appropriate investigations such as radiography, ultrasonography, echo-cardiography and chromosomal analysis etc., Analysis of system wise distribution of the anomalies was performed. For each case, a detailed antenatal and maternal history such as age, parity, history of consanguinity, including the familial and gestational factors, was obtained by interviewing the parents. Antenatal ultrasonography findings were also noted.

Birth weights >2.5 kg were considered to be normal; whereas, birth weights <2.5 kg were termed as low birth weight (LBW).

Data was entered into excel data sheet and appropriate statistical analysis was performed. Propor-tion was calculated and the association was tested with Chi-square test and Fisher′s exact test. P < 0.05 was considered to be statistically significant.

III.   Results

During the study period, 6076 newborns were born in our institution; of which 84 had congenital malformations, making the prevalence 1.38%. Among all the newborns, 18 babies were born of twin delivery, one of triplet delivery and 2 of these 21 babies, that were products of multiple gestations, had one or more congenital anomalies.

The predominant system involved was musculo-skeletal system (36.90%) followed by central nerv-ous system (CNS) (25%) and gastro-intestinal (GI) system (16.6%). Talipes (17.1%) was the most common anomaly seen in the musculoskeletal group and likewise cleft lip (6.6%), meningomyelocele (6.3%) in CNS and cleft palate (3.5%) in GI system.

Regarding the parity of the mothers, 4429 were primipara and rest 1647 were multipara. Cases of congenital anomaly were found in 0.95% of multiparas, whereas in primiparas, the proportion was only 0.42%. It has been seen that more than half of the mothers were aged between 20 and 30 years (55.7%) with only 11.11% of the mothers were over the age of 30 years. The prevalence of congeni-tally anomalous babies born was 1.24% for mothers <20 years, 1.03% for 20-30 years and 3.78% for >30 years. This difference was statistically significant. In the present study, there were 3 consan-guineous couples and one couple showed some congenital anomaly (33.3%) in their babies whereas, the prevalence of anomalies was only1.36% in non-consanguineous couples. This difference in per-centage was highly significant. LBW was found to have a higher risk of congenital anomalies. The occurrence of congenital anomalies was about 15.18 % in case of babies delivered with low birth weight (Table 2). The congenital anomalies affected significantly higher proportion of male babies (2.59%) than their female counterparts (0.75%).

Table 1 showing System wise distribution of congenital anomalies (N=84)

System	Number	Percentage
Musculoskeletal System	31	36.9%
Central nervous system	21	25%
Cardiovascular system	3	3.57%
Respiratory system	6	7.14%
Genitourinary system	9	10.7%
Gastro-intestinal system	14	16.66%

Table 2 showing association between congenital anomalies and maternal and perinatal risk factors

Variable	Groups	Congenital anomaly	Congenital anomaly	Congenital anomaly	Congenital anomaly	Total	P Value df value X2
		Yes	Yes	No	No		
		Number	Percentage	Number	Percentage		
Maternal age	<20 years	26	1.24%	2058	98.76%	2084	X2=27.36
	20-30 years	35	1.03%	3349	98.97%	3384	df=2
	>30 years	23	3.78%	585	96.22%	608	p=0.00001
Parity	Primipara	16	0.36%	4413	99.64%	4429	X2=124.98
	Multipara	68	4.12%	1579	95.88%	1647	p=<0.00001
Parental Consanguinity	Consanguinity Present	1	33.3%	2	66.66%	3	X2=22.47
	Consanguinity Absent	83	1.36%	5990	98.63%	6073	p=0.000002
Birth Weight	low Birth weight <2.5 kg	12	15.18%	67	84.82%	79	x2=111.92
	Birth Weight > 2.5 kg	72	1.20%	5925	98.80%	5997	p=<0.0001
Gender	Male	54	2.59%	2024	97.41%	2078	p=<0.0001
	Female	30	0.75%	3968	99.25%	3998	x2=34.26

IV. Discussion

The pattern and prevalence of congenital anomalies may vary over time or with geographical loca-tion, reflecting a complex interaction of known and unknown genetic and environmental factors in-cluding socio-cultural, racial and ethnic variables [5]. With improved control of infections and nutri-tional deficiency diseases, congenital malformations have become important causes of perinatal mor-tality in developing countries like India [6].

In the present study, the prevalence of congenital malformations in the newborns were 1.38%, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9%.[7,8] There are other reports from different parts of the world representing different frequency of congenital malformations.[9,10] Although we got nearly the same result as reported in other stud-ies, [7,8,9,10,11] the prevalence of congenital anomaly would have been more than the present rate, if we could have included the abortions and stillbirths. Tertiary care hospital usually do not have specific catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Commu-nity based study should be ideal for true estimation of incidence of congenital anomalies in a popu-lation.

With regard to pattern of congenital anomalies in the study, the most common system involved was musculoskeletal system (36.9%), followed by CNS (25%), gastro-intestinal tract (GIT) (16.6%), genitourinary (10.7%), and cardiovascular system (3.5%). This was comparable with studies con-ducted by others.[12,13,14,15,16,17] Some studies however recorded higher incidence of CNS malformations followed by GIT and musculoskeletal system, [9,18] whereas Suguna Bai et al.[19] reported GI malformations as the most common one.

More male babies with congenital anomalies than females were noted in the present study. Male preponderance was similar to the other studies.[6,7] It may be because of the fact that the females were afflicted with more lethal congenital malformations and could not survive to be born with signs of life.

Association of LBW with increased risk of congenital malformations is very well-documented [6]. Our finding is in accordance with that. The incidence of congenital anomalies was significantly higher in low birth weight babies as compared with the babies weighing more than 2.5 kgs, which is in conformity with the previous studies reported from this country [17].

Suguna Bai et al [19] reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta et al. [18] documented statistically insignificant association of increased maternal age and congenital anomalies. Regarding the relationship between maternal age and babies born with congenital malformations our study found, that the majority of malformed ba-bies were born of mothers aged 20-30 years, and it was statistically significant.

Previous studies have reported significantly higher incidence of malformations among the multipa-ras [6]. Our result is consistent with this finding, which indicates a positive correlation between the birth order and the incidence of congenital anomalies.

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Consanguineous marriages are reported to play a major role in the occurrence of congenital malfor-mations [20]. In the present study also, prevalence of malformed babies was more when born out of consanguineous marriages as seen in studies from Kuwait, Arab [21,22] and also India [17].

Despite the high risk of recurrence of congenital malformations, there are no well-accepted preven-tive measures in developing countries like India. It indicates that strong preventive measures for congenital anomalies in this region are needed. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguin-eous marriages need to be highlighted to decrease the incidence of congenital anomalies.

V. Conclusion

This study has highlighted the prevalence of musculoskeletal and central nervous system anomalies in this region. Congenital anomalies were more likely to be associated with low birth weight, multi-parity, maternal age (between 20 to 30 years) and consanguinity. The congenital anomalies affected significantly higher proportion of male babies than their female counterparts. Pre-pregnancy folic acid supplementation, regular antenatal visits and prenatal diagnosis are recommended for its pre-vention and early detection.

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