

Pattern of Congenital Anomalies at Birth: A Hospital-based Study

Anuja Bhalerao¹, Krutika Bhalerao²

ABSTRACT

Background: Congenital anomalies form the major cause of adverse neonatal outcome as stillbirths and neonatal mortality. The distribution and prevalence of congenital anomalies may be different with time or with geographical location.

Aims and objectives: The aim of this study is to determine the pattern of congenital anomalies in obstetrics and gynecology department of a rural tertiary medical college and hospital during the period of May 2013 to December 2015. All the babies delivered in this tertiary hospital during this period were included. The newborns were examined by obstetricians and pediatricians for the presence of congenital anomalies and mothers were interviewed using a case record form for sociodemographic variables.

Results: During the study period, 6,076 babies were born; of which, 84 babies had congenital malformations, giving the prevalence of 1.38%. Majority of the women (55.7%) belonged to the age group between 21 years and 30 years. Congenital anomalies were seen more commonly (2.57%) in the multiparae in comparison with the primiparae (0.42%). The predominant system involved was the musculoskeletal system (36.90%) followed by the central nervous system (CNS) (25%) and the gastrointestinal (GI) system (16.6%). Talipes (17.1%) was the most common anomaly in the musculoskeletal group followed by cleft lip and cleft palate in the GI system. It was seen that majority of congenital anomalies were associated with low birth weight (LBW), prematurity, multiparity, and consanguinity.

Conclusion: Health education and awareness for preventable risk factors is to be emphasized in general population, and early prenatal diagnosis and management of common anomalies is strongly recommended for better outcome.

Keywords: Congenital anomaly, Prematurity, Prevalence, Risk factors.

Journal of South Asian Federation of Obstetrics and Gynaecology (2019): 10.5005/jp-journals-10006-1705

INTRODUCTION

Congenital malformations are defects in morphogenesis during early fetal life. As per the World Health Organization (WHO) document of 1972, the term “congenital malformations” should be limited to structural defects at birth.¹ However, in the recent WHO fact sheet of October 2012, congenital anomalies are defined as structural or functional anomalies, including metabolic disorders, present at the time of birth.² Congenital anomalies represent an important cause of neonatal mortality both in developed and developing countries. They account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India.^{3,4} They are an important cause of fetal loss and are responsible for significant increase in number of preterm birth and childhood and adult morbidity affecting mothers and their families. Surveys all over the world have shown that birth prevalence of congenital anomalies is affected due to social, racial, economical, and ecological influences. So to decrease the incidence of various congenital anomalies, it is important to identify prevalence of various anomalies in the society and the risk factors for them.

MATERIALS AND METHODS

This hospital-based cross-sectional study was carried out in the obstetrics and gynecology department of a rural tertiary medical college and hospital during the period of May 2013 to December 2015. All the babies delivered in this hospital during this period were included. The newborn babies were examined by obstetricians and pediatricians for the presence of various congenital anomalies and detailed history was taken from mothers for sociodemographic variables. The newborns were examined

^{1,2}Department of Obstetrics and Gynecology, NKP Salve Institute of Medical Sciences and Research Center and Lata Mangeshkar Hospital, Nagpur, Maharashtra, India

Corresponding Author: Krutika Bhalerao, Department of Obstetrics and Gynecology, NKP Salve Institute of Medical Sciences and Research Center and Lata Mangeshkar Hospital, Nagpur, Maharashtra, India, e-mail: krutika.bhalerao@yahoo.co.in

How to cite this article: Bhalerao A, Bhalerao K. Pattern of Congenital Anomalies at Birth: A Hospital-based Study. *J South Asian Feder Obst Gynaecol* 2019;11(4):252–254.

Source of support: Nil

Conflict of interest: None

methodically and meticulously and were assessed systemwise for the presence of congenital anomalies. Diagnosis of congenital anomalies was done on the basis of clinical evaluation of newborn babies by the pediatrician and other appropriate investigations such as radiography, ultrasonography, echocardiography, chromosomal analysis, etc. Analysis of systemwise distribution of the anomalies was performed. For every 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 of <2.5 kg were considered as low birth weight (LBW). Data were entered into a case record form and then in the Excel sheet and were analyzed statistically.

RESULTS

During the study period, 6,076 babies were delivered in our institution; of which, 84 babies had congenital malformations, giving the prevalence of 1.38%. Among the newborns, 18 babies were born of twin delivery, one of triplet delivery, and two of these 21 babies, born of multiple gestations, had one or more congenital anomalies. The system involved predominantly was the musculoskeletal system (36.90%) followed by the central nervous system (CNS) (25%) and the gastrointestinal (GI) system (16.6%). The most common anomalies were talipes (17.1%) in the musculoskeletal group, cleft lip (6.6%) and meningomyelocele (6.3%) in CNS (Table 1), and cleft palate (3.5%) in the GI system. As far as the parity of the mothers is concerned, 4,429 mothers were primiparas and rest 1,647 mothers were multiparas. Congenital anomalies were found in 0.95% of multiparas, whereas in primiparas, they were only 0.42%. More than half of the mothers who gave birth to anomalous babies were aged between 20 years and 30 years (55.7%) with only 11.11% of the mothers were over the age of 30 years. The prevalence of congenitally 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 proving that increasing age has association with anomalies. In the present study, there were three consanguineous couples and one couple had a congenitally anomalous baby (33.3%), whereas the prevalence of anomalies was only 1.36% in nonconsanguineous couples. This difference in percentage was highly significant. Low birth weight was found to be associated with higher risk of congenital anomalies. The occurrence of congenital anomalies was about 15.18% in case of babies delivered with LBW (Table 2). The congenital anomalies affected significantly higher proportion of male babies (2.59%) than their female counterparts (0.75%).

DISCUSSION

The pattern and prevalence of various congenital anomalies varies with time and geographical locations, showing an intricate

correlation between various known and unknown genetic and environmental factors including sociocultural, racial, and ethnic variables.⁵ Timely detection and treatment of infections by antibiotics, and nutritional deficiencies which are the causes of congenital malformations have become important causes of perinatal mortality in India.⁶ In the present study, the prevalence of congenital malformations in the newborns was 1.38%, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9%.^{7,8} Studies from different parts of the world have shown different frequency of congenital malformations.^{9,10} Although our results are same as reported in other studies,⁷⁻¹¹ the prevalence of congenital anomalies would have been more than the present rate, if abortions and stillbirths would have been included. The tertiary care hospital usually has a large nonspecific catchment area and encounters complicated cases as referred for treatment. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Large community-based study should be ideal for true estimation of incidence of congenital anomalies in a population. With regard to the pattern of congenital anomalies in the study, the most common system involved was the musculoskeletal system (36.9%), followed by CNS (25%), the gastrointestinal tract (GIT) (16.6%), the genitourinary (10.7%), and the cardiovascular system (3.5%). This was in accordance with studies conducted by others.¹²⁻¹⁷ Some studies had higher incidence of CNS malformations followed by GIT and the musculoskeletal system,^{9,18} whereas study by Suguna Bai et al.¹⁹ reported GI malformations as the most common one. Male babies were more commonly associated with congenital anomalies than females in the present study. Male preponderance was similar to the other studies.^{6,7} It may be due to the fact that the female babies were affected with more lethal congenital malformations and so could not survive to be born with signs of life. The association between LBW with increased risk of congenital malformations was appreciable.⁶ Our finding is in accordance with that congenital anomalies were significantly higher in low-birth-weight babies as compared to babies weighing more than 2.5 kg, which is similar with the previous studies reported from this country.¹⁷ Suguna Bai et al.¹⁹ showed 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 babies were born of mothers aged 20–30 years, and it was statistically significant. Previous studies have reported significantly higher incidence of malformations among the multiparas.⁶ Our result is consistent with this finding,

Table 1: Systemwise 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
Gastrointestinal system	14	16.66

Table 2: Association between congenital anomalies and maternal and perinatal risk factors

Variable	Groups	Congenital anomaly		Congenital anomaly		Total	p value, df value, χ^2
		Yes	Percentage	No	Percentage		
Maternal age	<20 years	26	1.24	2,058	98.76	2,084	p = 0.00001, df = 2, $\chi^2 = 27.36$
	20–30 years	37	1.09	3,347	98.91		
	>30 years	23	3.78	585	96.22		
Parity	Primiparas	19	0.42	4,410	99.58	4,429	p ≤ 0.0001, $\chi^2 = 62.81$
	Multiparas	68	2.57	2,569	97.43		
Consanguinity	Present	1	33.3	2	66.66	3	p ≤ 0.0001, $\chi^2 = 624.1$
	Absent	2	0.03	6,074	99.97		
Birth weight	Very low	12	15.18	67	74.82	79	

which indicates a positive correlation between the birth order and the incidence of congenital anomalies. Consanguineous marriages play a major role in the occurrence of congenital malformations.²⁰ In the present study also, the 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, formulated guidelines for various preventive measures in developing countries like India. It indicates that health education, antenatal care, prenatal tests, and strong preventive measures are needed to decrease the incidence of various anomalies. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations, and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies.

CONCLUSION

This study highlights the prevalence of musculoskeletal and CNS anomalies in this region. Congenital anomalies were more likely to be associated with LBW, multiparity, maternal age (between 20 years and 30 years), and consanguinity. The congenital anomalies affected significantly higher proportion of male babies than their female counterparts. Prepregnancy high-dose folic acid supplementation, regular antenatal visits per WHO schedule, and prenatal diagnosis are recommended for their prevention and early detection. In spite of high risk of recurrence, there are no guidelines and preventive measures in India. Drastic preventive measures need to be taken by increasing awareness through social media, rallies, education camps, prenatal diagnosis, and timely termination to decrease incidence of congenital anomalies.

REFERENCES

1. Patel ZM, Adhia RA. Birth defects surveillance study. *Indian J Pediatr* 2005;72(6):489–491. DOI: 10.1007/BF02724426.
2. World Health Organization. Section on congenital anomalies. [Cited on 2012 Oct]. Available from: <http://www.who.int/mediacentre/factsheets/fs370/en/>.
3. Bhat BV, Ravikumara M. Perinatal mortality in India-need for introspection. *Indian J Matern Child Health* 1996;7:31–33.
4. Agarwal SS, Singh U, Singh PS, et al. Prevalence and spectrum of congenital malformations in a prospective study at a teaching hospital. *Indian J Med Res* 1991;94:413–419.
5. Birch MR, Grayson N, Sullivan EA, AIHW Cat. No. PER 23. Birth Anomalies Series No. 1. Sydney: AIHW National Perinatal Statistics Unit; 2004. Recommendations for development of a new Australian birth anomalies system: A review of the congenital malformations and birth defects data collection.
6. Mohanty C, Mishra OP, Das BK, et al. Congenital malformations in newborns: a study of 10,874 consecutive births. *J Anat Soc India* 1989;38:101–111.
7. Chaturvedi P, Banerjee KS. Spectrum of congenital malformations in the newborns from rural Maharashtra. *Indian J Pediatr* 1989;56(4):501–507. DOI: 10.1007/BF02722424.
8. Taksande A, Vilhekar K, Chaturvedi P, et al. Congenital malformations at birth in Central India: a rural medical college hospital based data. *Indian J Hum Genet* 2010;16(3):159–163. DOI: 10.4103/0971-6866.73412.
9. Khatemi F, Mamoori GA. Survey of congenital major malformations in 10/000 newborns. *Iran J Pediatr* 2005;15:315–320.
10. Tomatir AG, Demirhan H, Sorkun HC, et al. Major congenital anomalies: a five-year retrospective regional study in Turkey. *Genet Mol Res* 2009;8(1):19–27. DOI: 10.4238/vol8-1gmr506.
11. Mir NA, Galczek WC, Soni A. Easily identifiable congenital malformations in children: survey of incidence and pattern in 32,332 live born neonates. *Ann Saudi Med* 1992;12(4):366–371. DOI: 10.5144/0256-4947.1992.366.
12. Gupta RK, Singh A, Gupta R. Pattern of congenital anomalies in newborn at birth: a hospital based prospective study. Proceedings of the 42nd National Conference of Indian Academy of Pediatrics (Pedicon); Jan 6–9; Kolkata, India. 2005.
13. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. *Indian Pediatr* 1994;31(10):1187–1191.
14. Tibrewala NS, Pai PM. Congenital malformations in the newborn period. *Indian Pediatr* 1974;11(6):403–407.
15. Mishra PC, Baweja R. Congenital malformations in the newborn—a prospective study. *Indian Pediatr* 1989;26(1):32–35.
16. Verma M, Chhatwal J, Singh D. Congenital malformations – a retrospective study of 10,000 cases. *Indian J Pediatr* 1991;58(2):245–252. DOI: 10.1007/BF02751129.
17. Mathur BC, Karan S, Vijaya Devi KK. Congenital malformations in the newborn. *Indian Pediatr* 1975;12(2):179–183.
18. Dutta V, Chaturvedi P. Congenital malformations in rural Maharashtra. *Indian Pediatr* 2000;37(9):998–1001.
19. Suguna Bai NS, Mascarene M, Syamalan K, et al. An etiological study of congenital malformation in the newborn. *Indian Pediatr* 1982;19(12):1003–1007.
20. Hudgins L, Cassidy SB. Congenital anomalies. In: Martin RJ, Fanaroff AA, Walsh MC, ed. *Neonatal-Perinatal Medicine*, 8th ed. Philadelphia: Mosby-Elsevier; 2006. pp. 561–581.
21. Madi SA, Al-Naggar RL, Al-Awadi SA, et al. Profile of major congenital malformations in neonates in Al-Jahra region of Kuwait. *East Mediterr Health J* 2005;11(4):700–706.
22. Al-Gazali LI, Dawodu AH, Sabarinathan K, et al. The profile of major congenital abnormalities in the United Arab Emirates (UAE) population. *J Med Genet* 1995;32(1):7–13. DOI: 10.1136/jmg.32.1.7.