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Original Article

Prevalence and Pattern of Congenital Malformations among Neonates in a Medical College Hospital - A Retrospective Study

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Abstract

Background: Congenital malformations (CMs) represent a major cause of admission in most of the NICU all over the world. They represent a defect in the morphogenesis during early fetal life. With the advances in delivery and newborn care, CMs have emerged as one of the most common causes of perinatal mortality.

Objective: The objective of this study was to determine the prevalence and pattern of CMs among neonates in a teaching hospital.

Materials and Methods: The retrospective study of live neonates from newborn to 28 days of age both inborn and outborn admitted to the unit irrespective of their general condition with CMs comprised the study population. Details of investigations like ultrasonography, radiology, echocardiography, laboratory studies have done were noted from the case record. Their outcome in the form of morbidity, hospital stay, and mortality was analyzed.

Results: In 2132 babies, with malformations were 87 (4.08%). Of which inborn babies were 3.9% and outborn babies were 4.8%. Of the malformed babies were 54% of male and 45% of female, 1% was DSD. Cesarean delivery was 63.2%, other modes were 36.8%. The cardiovascular system was involved in 35.6% of babies, followed by the musculoskeletal system (26.4%), then the genitourinary system 13.8%, gastrointestinal (9.2%), and central nervous system (10.3%). Maternal risk factors associated with malformations were maternal diabetes in 2.3%, age between 21 and 30 in 87.4%, and consanguinity in 8%. Maximum mortality occurred in babies with cardiovascular system malformations (76.5%). Majority of babies with malformations discharged (65.5%) only 19.5% of babies expired and 15% of babies were referred for intervention at a higher center.

Conclusions: CMs represent one of the causes of neonatal mortality. Health-care managers must stress on primary prevention in the form of good antenatal care, nutrition, and drugs to decrease the preventable share of CMs. Early detection and timely management are required to decrease mortality.

Key words: Congenital anomaly, Prematurity, Prevalence, Risk factors

INTRODUCTION

The World Health Organization defines the term congenital malformation (CM) as structural defects present at birth. CM may be minor or major. The minor malformation is defined as structural abnormality present at birth which has minimal effect on clinical function but may have a cosmetic effect, for example, preauricular tag. Major malformation has a significant effect on function or on social acceptability, for example, ventricular septal defect and cleft lip.[1] Dysmorphology is the study of abnormalities of the human form and mechanism that causes these abnormalities. About 20–30% of infant deaths and 30–50% post-neonatal deaths are due to CM. The first trimester, especially between the 3rd and 8th weeks of gestation, is the crucial period for morphogenesis of organs. Any insult in any form during this period can cause congenital abnormality. This is the period where preventive intervention strategy will reduce the incidence of developing CMs.[2] Other risk factors for CM are

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maternal age, drug intake, teratogens, radiation exposure, maternal illnesses, smoking, and alcohol consumption.[3] Different antenatal screening methods such as maternal serum markers, chorionic villus sampling, amniocentesis, cordocentesis, and ultrasonography can be used to detect anomalies. *In utero* intervention for some CMs such as hydrocephalus, posterior urethral valves, cleft lip, and hydronephrosis is gaining popularity.[3] As other causes of infant mortality such as infections and nutritional deficiencies are being brought under control, CMs are rapidly emerging as one of the major worldwide problems.[4,5] The prevalence of CM ranges between 3% and 7% and varies in different geographical, racial, and ethnic parts of the world.[6,7] As far as the involvement of different systems of the body is concerned, the brain has the highest incidence of CM, i.e., 10/1000 followed by heart 8/1000, kidney 4/1000, limb 1/1000, and miscellaneous 6/1000 live births.[8] The prevalence rate of congenital anomalies is increasing due to exposure to teratogens of various kinds.[9] In India, CMs have emerged as the third most common cause of perinatal mortality.[10]

Aim

This study aims to determine the prevalence and pattern of CMs among neonates in a teaching hospital.

MATERIALS AND METHODS

This study was conducted at a tertiary care hospital TKMCH by retrospectively analyzing the case sheets for a period of 1 year from January 2017 to December 2017. All the live neonates from newborn to 28 days of age both inborn and outborn admitted to the unit irrespective of their general condition with CMs comprised the study population. The neonatal examination was done and other information regarding gender, weight, gestational age, mode of delivery, consanguinity, maternal age, antenatal visit record, and family history collected from the case sheets were recorded on a predesigned pro forma. Details of investigations like ultrasonography, radiology, echocardiography, laboratory studies have done were noted from the case record. Marriage was considered consanguineous when it has occurred between a male and a female who are blood related, for example, between brother and sister, between the 1st cousins, etc. Birth weights >2.5 kg, <2.5 kg, and <1.5 kg were categorized and babies with malformations in these groups were analyzed. Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of the past menstrual period, were considered as premature. The outcome in the form of morbidity and mortality was taken up to their hospital stay. Finally, their outcome in the form of morbidity, hospital stay, and mortality was analyzed.

RESULTS

In this study, 2132 babies were screened and found that the incidence of CM in live births was 87 babies (4.08%). In the present study, 20.7% of outborn babies with malformations were referred to us so this may be the reason for a higher incidence [Figure 1]. There are no significant gender variations observed in the study. In the present study, 23% of malformed babies were preterm and 77% of babies were full term. In the present study, 2.3% of malformed babies had birth weight ≤1500 g. In this study, 42.2% of babies with malformations were low birth weight while 59.8% of babies with weight >2500 g. In this study, male babies were more affected with malformations. 54% of total malformed babies were male and 45% of female babies. The incidence of malformation was higher (87.4%) in mother aged 21–30 years and 9.2% in mother >31 years [Figure 2]. 8% incidence of CM was found in consanguinity marriage. No risk factor was noted in 95.4% of high-risk mothers, 2.3% of GDM and 2.3% of thyroid disorders were noted. There is no significant difference observed

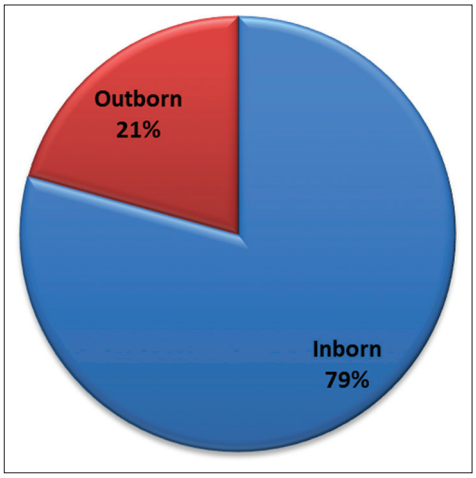

Figure 1: Type of born

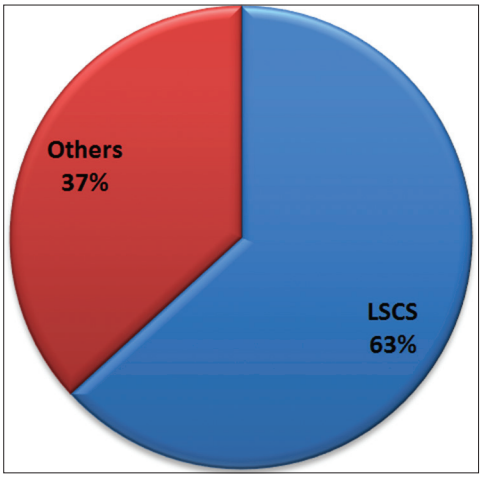

Figure 2: Type of delivery

in the birth order of the baby. LSCS were had a higher incidence of 63.2% CM [Figure 2]. The most common systems involved in this study were cardiovascular system (35.6%) and musculoskeletal system (22.3%), followed by gastrointestinal tract (15.9%), genitourinary system (264%), and genitourinary system (13.8%) [Table 1]. 19.5% mortality were noted in this study; the higher number was in cardiovascular system 76.5% [Figure 3].

DISCUSSION

Many studies in India have addressed the prevalence of birth defects in the country four. Their frequency varies from 1.94% to 2.03% of birth on an average.[5] In the present study, the incidence of CM in live births was 4.08%, this was marginally higher when compared with the study by Taksande *et al.*,[11] which shows an incidence of 1.9% in live births. Singh and Gupta[12] show an incidence of 1.5% in live births and 8.7% in stillbirths. Malla[13] shows an incidence of 0.36% in live births and 2.0% in stillbirths. In the present study, 20.7% of outborn babies with malformations were referred to us so this may be the reason for a higher incidence. In the present study, 23% of malformed babies were preterm and 77% of babies were full term. A study by Malla[13] and Dutta *et al.*[14] showing similar results (36% preterm and 64% full-term, and 40.6% preterm and 59.4% full-term babies, respectively).

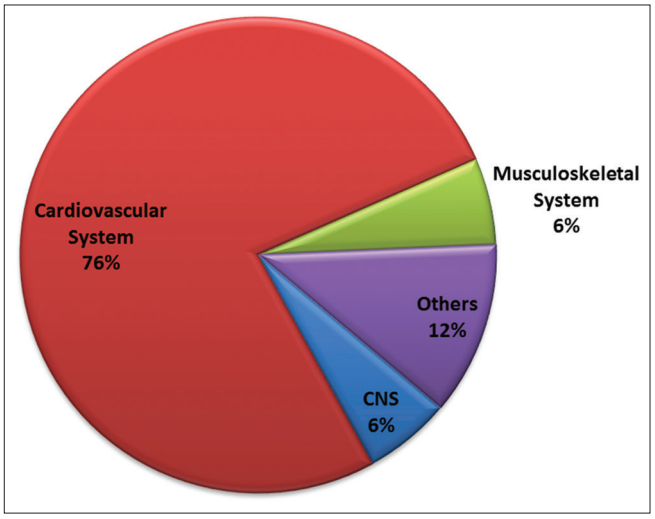

Figure 3: Mortality distribution

Table 1: Type of malformation

System	Malformation type	Frequency (%)	Percentage of total
CNS - 9	Meningomyelocele	4 (4.6)	10.30
	Colpocephaly	1 (1.1)	
	Hydrocephalus	3 (3.5)	
	Arachnoid cyst	1 (1.1)	
Cardiovascular system - 31	ASD	10 (11.5)	35.60
	PDA	7 (8.1)	
	Single atrium	1 (1.1)	
	VSD	10 (11.5)	
	Congenital heart block	1 (1.1)	
	HLHS	1 (1.1)	
	Dextrocardia	1 (1.1)	
Musculoskeletal system - 23	Congenital dislocation of hip	1 (1.1)	26.40
	CTEV	10 (11.5)	
	Skeletal dysplasia	1 (1.1)	
	Bifid thumb	1 (1.1)	
	Syndactyly	1 (1.1)	
	Cleft lip	1 (1.1)	
	Cleft palate	1 (1.1)	
	Cleft lip and palate	4 (4.6)	
	Preauricular tag	3 (3.5)	
Genitourinary system - 12	Hydronephrosis	5 (5.7)	13.80
	Hypospadias	2 (2.3)	
	Hydrocele	4 (4.6)	
	Ambiguous genitalia	1 (1.1)	
Digestive system - 8	Tracheoesophageal fistula	2 (2.3)	9.20
	Diaphragmatic hernia	1 (1.1)	
	Ileal atresia	1 (1.1)	
	Mesenteric cyst	1 (1.1)	
	Anorectal malformations	1 (1.1)	
	Imperforate anus	2 (2.3)	
Others - 4	Multiple congenital anomalies	1 (1.1)	4.60
	Right Lung hypoplasia	1 (1.1)	
	Single umbilical artery	1 (1.1)	
	Epulis	1 (1.1)	

In the present study, 2.3% of malformed babies had birth weight ≤1500 g that were similar to a study by Patel and Adhia[15] (9.8% of malformed babies). In this study, 42.2% of babies with malformations were low birth weight while 59.8% of babies with weight >2500 g. A study by Patel and Adhia showing results of 59.8% of babies with weight ≤2500 g and 40.2% of babies with weight >2500 g. In this study, male babies were more affected with malformations. 54% of total malformed babies were male and 45% of female babies. A study by Taksande *et al.* showing similar results (61% of male babies and 37.4% of female babies, and 64.7% of male babies and 34% of female babies, respectively). The incidence of malformation was higher (87.4%) in mother aged of 21–30 years, and 9.2% in mother >31 that is high on comparing with a study by Taksande *et al*.[11] and Saiyad and Jadav[16] (incidence of malformation 36% and 20% live births, respectively). Taksande *et al.* reported a higher incidence of malformations among the multiparas (19.5%). In the present study, incidence was 19.6%. Our result was primipara having 41.3%.[6] The most common systems involved in this study were cardiovascular system (35.6%) and musculoskeletal system (22.3%), followed by gastrointestinal tract (15.9%), genitourinary system (264%), and genitourinary system (13.8%). This was comparable with a study conducted by Taksande *et al.* which shows cardiovascular system (23%), musculoskeletal system (21.9%), gastrointestinal tract (14%), genitourinary (18.9%), and central nervous system (9.1%). Central nervous system malformations were predominantly seen in the study by Sugunabai[17] and Malla[13] (44% and 40%, respectively);[8] gastrointestinal system malformations are predominantly seen in the study by Desai and Desai.[18]

CONCLUSIONS

Differences between studies might be the effect of different racial, ethnic, and social factors in various parts of the world. Congenital anomalies are an important cause of infant and childhood deaths, chronic illness, and disability. We have to develop strategies to diagnose, treat, rehabilitate, and prevent birth defects. In preparation of this and effective planning, crucial measures include obtaining data on prevalence, nature of birth defects, genetic contributions, morbidity, and mortality. The community-based study should be ideal for true estimation of the prevalence of congenital anomalies in a population. Increasing awareness about maternal risk factors during pregnancy and educational programs on CMs needs to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

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