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Research Article

Pattern of congenital malformations in newborn: a hospital-based study

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ABSTRACT

Background: Data on congenital malformations from developing countries like India are very few. However, it is important to have comprehensive and reliable data on the description and frequency of congenital malformations to allow surveillance and the implementation of appropriate public health strategies for prevention and management. In this study, we describe the pattern of congenital malformations seen in newborns delivered in tertiary care hospital of western Maharashtra. The objective was to study various newborn characteristics and to determine the frequency and pattern of congenital malformations at birth. Cross-sectional study conducted in Govt Medical College and Hospital Miraj, a tertiary care hospital in district of Maharashtra from June 2014 to November 2014 targeting all newborns delivered in hospital during study period.
Methods: Data was collected by administering a semi structured questionnaire and a devised newborn screening clinical examination protocol.
Results: Out of all 892 newborns (live births and still births), 24(2.69%) were having congenital malformations at birth and out of that, malformations involving circulatory system was highest i.e. 29.6% compared to other system.
Conclusions: As compared to other studies circulatory disorders appear to be more common and by improvement in antenatal, postnatal diagnosis, early referral to tertiary hospital and early intervention most of these newborns can be saved.

Keywords: Congenital malformations, Anomalies, Newborns, Birth defects

INTRODUCTION

Birth defects are a diverse group of disorders of prenatal origin that can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. Maternal infections such as rubella, maternal illnesses like diabetes mellitus (DM), iodine and folic acid deficiency, exposure to medicinal and recreational drugs including alcohol and tobacco, certain environmental chemicals, and doses of radiation are all other factors that cause birth defects.[1] Birth defects, Congenital malformations (CMs) and congenital anomalies (CAs) are interchangeable terms used to describe developmental defects that are present at birth.[2] Congenital anomalies represent defective morphogenesis during early fetal life. A broader definition includes metabolic or microscopic defects at a cellular level. Major anomalies have serious medical, surgical and cosmetic consequences.[3]

Congenital anomalies affect approximately 1 in 33 infants leading to 6.6% deaths in infants and causing significant morbidity in children.[4] According to WHO Fact sheet, out of all causes of 2.761 million deaths worldwide during the neonatal period in 2013, congenital malformations contributed to 276000 deaths, preterm birth complications 965000, intra partum related complications (birth asphyxia) 662000, neonatal sepsis 421000 and other important causes 437000.[5] Community based study by Indian council of Medical Research

(ICMR) reported that congenital malformations accounted for 6.6% of neonatal deaths in the rural as well as urban slum communities.[6]

Congenital malformations are emerging as important perinatal problem contributing sizeably to the perinatal mortality and morbidity with considerable repercussion on the mothers and the families affected. The life-threatening congenital malformations must be identified by thorough clinical examination because early diagnosis and surgical correction or palliation of these infants offer the best chance for survival.[7] Most of the studies in the past focus on the infections and their effects during infancy and childhood. Very few studies have been conducted to know about the morbidity rates and outcomes due to congenital malformations.[4] The purpose of the present study is to present the spectrum of congenital malformations in the newborns, at birth, diagnosed clinically at a referral hospital in western maharashtra.

METHODS

This hospital based descriptive Cross-sectional study was conducted in a tertiary care hospital in a district of Maharashtra from June 2014 to November 2014. It was a part of project conducted under dept. of community medicine GMC Miraj. Total consecutive sampling method was used and from the deliveries conducted during the study period, 890 deliveries were available for study after considering inclusion and exclusion criteria. Inclusion Criteria:a. All the live births and their mothers during the study period.

b. All the still births and their mothers during the study period. c. Mothers of newborns who had given consent to carry out study. Exclusion Criteria: Mothers not willing to participate in the study. All newborns delivered in tertiary care hospital constituted the target population for the study. There were 892 newborns (including twins), of which 865 were live births and 27 were still births. Data was collected in Labour room and/or postnatal care ward by devised newborn screening clinical examination protocol. Devised newborn screening method and the predesigned, semi-structured interview schedule was pretested in a pilot study in the Labour room & postnatal care ward of Obstetrics and Gynecology Department of study hospital before the final data collection to check for acceptability, clarity of language, accuracy of translation. Validity, reliability, and quality of raw data and necessary corrections were made accordingly.

Permission and clearance was obtained from institutional ethical committee before start of the study. Participation was voluntary, informed verbal consent was obtained from the participants, assuring safe nature of method of newborn screening and they were ensured that confidentiality and anonymity would be maintained throughout the study after explaining the purpose of study Malformations detected by the thorough clinical examination, supported when necessary by referral to specialists like paediatrician and other appropriate laboratory investigations for biochemical, hematological and histo-pathological examinations, x-ray, CT scan, MRI scan, ultrasound, echocardiography etc according to the case. Chromosomal analysis was not done and diagnoses such as Down's syndrome were based on clinical findings. The final diagnosis was noted at the time of the discharge. Relevant data was also recorded from previous and current hospital records wherever available. The data was recorded for the newborns that have visible/palpable external malformations. Infants who had congenital malformations involving more than one system were recorded once as having a multiple congenital malformations. The malformations were classified by organ system according to the 10th version of the World Health Organization International Classification of Diseases (ICD-10).[8] Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as preterm. The collected data was entered in Microsoft excel sheet, numerically coded, and then transferred to the SPSS (version 21). All the qualitative data was presented as frequency and percentage.

RESULTS

Table 1 shows Newborn characteristics of study group. Out of 892 newborns, 24 (2.69%) were having congenital malformations at birth. Table 2 shows frequency of single or multiple malformations in newborns. In the present study 22 (91.7%) newborns were born with single congenital malformation and 2 (8.3%) were born with multiple congenital malformations.

Table 1: Newborn characteristics of study group.

Variable		Frequency (N)	Percentage (%)
1.Gestational age	Term	838	93.9
	Preterm	54	6.1
	Total	892	100
2. Mode of delivery	Vaginal	585	65.6
	caesarean	307	34.4
	Total	892	100
3.Deliveries with number of newborns	Single	888	99.6
	Twin	2	0.4
	Total	890	100
4. Type of birth	Live birth	865	97
	Still birth	27	3
	Total	892	100
5. Gender	Female	432	48.4
	Male	460	51.6
	Total	892	100
6. Birth weight	≥2500 grams	673	75.4
	<2500 grams	219	24.6
	Total	892	100

Table 2: Frequency of single & multiple congenital malformations in newborns.

Congenital malformations	Frequency in newborns (No. of malformations)	Percentage (%)
Single	22 (22)	91.7
Multiple	2 (5)	8.3
Total	24 (27)	100

Table 3: Comparison of frequency of congenital malformations in newborns between present study and other studies.

Congenital malformations	Percentage (%)		
	Present study	Singh R et al (2000)[9]	Madi S. A et al (2005)[10]
Single	91.7	43.7	49.4
Multiple	8.3	56.3	50.6
Total	100	100	100

Table 4: Distribution of congenital malformations according to ICD-10.

Organ System	Congenital malformation	ICD-10 code[8] (Chapter XVII)	Frequency (N)	Total	Percentage (%)
1. Nervous system	Anencephaly	Q00.0	2	5	18.5
	Hydrocephalus	Q03.0	1		
	Vermian Agenesis	Q04.3	1		
	Closed Sacral spina bifida	Q05.8	1		
2. Eye	Anophthalmos of Right Eye	Q11.1	1	1	3.7
3. Ear	Ear tags	Q17.0	1	1	3.7
4. Circulatory system	Atrial septal defect	Q21.1	1	8	29.6
	Tetralogy of Fallot	Q21.3	2		
	Ectopia cordis	Q24.8	1		
	Congenital malformation of heart, unspecified	Q24.9	2		
	Patent ductus arteriosus	Q25.0	1		
	Single Umbilical Artery	Q27.0	1		
5. Cleft lip and cleft palate	Cleft palate	Q35.9	1	3	11.1
	Cleft hard palate with unilateral cleft lip	Q37.1	2		
6. Digestive	Imperforate anus	Q42.2	1	1	3.7
7. Genital system	Hypospadias	Q54.9	1	1	3.7
8. Musculoskeletal system	Congenital Dislocation of hip, unilateral	Q65.0	1	5	18.5
	Talipes equinovarus	Q66.0	1		
	Osteogenesis imperfecta	Q78.0	1		
	Omphalocele	Q79.2	2		
9. Other	Cystic hygroma	Q87.8	1	1	3.7
10. Chromosomal abnormalities	Down syndrome	Q90.9	1	1	3.7
		Total	27	27	100

Table 4 shows that malformations involving circulatory system (Atrial septal defect, tetralogy of Fallot, ectopia cordis, unspecified congenital malformation of heart, patent ductus arteriosus, single umbilical artery) was 29.6% followed by nervous system (anencephaly, hydrocephalus, vermian agenesis, closed sacral spina bifida) which was 18.5% and musculoskeletal system (congenital dislocation of hip, talipes equinovarus, osteogenesis imperfecta, omphalocele) which was 18.5% and cleft lip, cleft palate was 11.1% of total malformations. The congenital malformations for eye (anophthalmos of right eye), ear (ear tags), digestive system (imperforate anus), genital system (hypospadias), chromosomal abnormalities (Down syndrome), and others (cystic hygroma) were 3.7% each. The most common system involved was circulatory followed by nervous and musculoskeletal system.

DISCUSSION

Out of 892 newborns, 24 (2.69%) were having congenital malformations at birth in our study. This is similar to the cross- sectional study conducted by Khan Z et al (2.8%) Desai NA et al (3.61%), Singh A et al (1.5%), Taksande A et al (1.91%) Shah K et al (2.38%).[3,11-14] Table 3 shows comparison of frequency of congenital malformations in newborns between present study and other studies.

In our study the most common system involved was circulatory followed by nervous and musculoskeletal system. Similarly Kumar V et al in rural based study reported that malformations involving cardiovascular system were commonest (37%), followed by musculoskeletal (30%), gastrointestinal system (23%), central nervous system (13%) and genitourinary system (6.6%) and Takshande et al found that Cardiovascular malformations were most common in live births, followed by musculoskeletal malformations.[14,15] The CNS defects were most commonly seen in still born. While Desai NA et al in their study observed that the most common system involved was musculoskeletal system (31.65%), followed by gastrointestinal (17.2%) and cardiac anomalies (16.46%), Ali A et al in a prospective study found that 94 of 460 live births (20.2 /1000) had at least a congenital malformation, the predominant systems involved were musculoskeletal system (7.9/1000), followed by genitourinary (7.1/1000), central nervous system (2.4/1000), digestive (1.1/1000) and chromosomal anomalies (0.9/1000).[12,16] Ndibazza J et al in their study revealed that the most commonly affected systems were the musculoskeletal (42.7 per 1000 births) and skin (16.1 per 1000 births).[17] Sarkar S et al found that the predominant system involved was Musculo-skeletal system (33.2%) followed by gastro-intestinal (GI) system (15%) and central nervous system (CNS) (11.2%).[18]

CONCLUSIONS

As compared to other studies circulatory disorders appear to be more common and by improvement in antenatal, postnatal diagnosis, early referral to tertiary hospital and early intervention most of these newborns can be saved. We need to involve and make aware all the health care workers who are providing maternal and child health care working in government or private sector so as to quantify exact pattern of congenital malformations involving any particular system.

Limitations

There were no compulsory blood tests and other investigations for each and every newborns except neonatal screening by physical examination. Those likely to have malformations were subjected for further investigations. As this is a hospital based study in tertiary care hospital, it may or may not be true presentation of community prevalence or pattern of congenital malformations. The subjects borne with congenital malformations could not be followed up due to feasibility and considering resource constrain of time and money.

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