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Original Research Article

Study of congenital malformations in newborns: a hospital based prospective study

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ABSTRACT

Background: Congenital malformations represent a defect in the morphogenesis during fetal life. Since the introduction of primary health care and immunization programme, congenital malformations have emerged as one of the commonest cause of perinatal mortality. The objective was to study the incidence, systemic distribution, various maternal risk factors and immediate outcome of congenital malformations in hospital delivered neonates.
Methods: This study was conducted at a tertiary care hospital for a period of 2 years. All the hospital delivered live neonates and stillbirth babies with congenital malformations are included in this study. Detailed history, examinations and investigations are carried out to identify etiological factors. Their outcome in form of morbidity and mortality are taken up to their hospital stay.
Results: Out of 9600 babies with malformations 171(1.88%) having single malformation and 23(0.25%) having multiple malformations. Incidence of malformations was higher in stillbirths (24.25/1000 livebirths) against than in live births (19.96/1000 livebirths). The cardiovascular system was involved in 23.4% of babies, followed by musculoskeletal system (22.3%) then gastrointestinal (15.9%) and genitourinary system (15.4%). Multiple malformations were seen in 11.8% cases. Maternal risk factors associated with malformations were oligohydramnios in 4.12%, previous abortion in 2.5%, eclampsia in 2.5%, polyhydramnios in 1.54%, maternal diabetes in 1.54% and consanguinity in 1.03%. Maximum mortality occurred in babies with gastrointestinal system malformations (56.5%) followed by cardiovascular system malformations (21.7%). Majority of babies with malformations discharged (78.9%) only 11.8% of babies expired and 2.6% of babies left against medical advice (LAMA).
Conclusions: Congenital malformations represent one of the causes of neonatal mortality. Stillborn babies have higher incidence of malformations. Antenatal ultrasonography and maternal risk factors has important role to identify malformations. Early detection and timely management required to decrease the mortality.

Keywords: Congenital malformations, Maternal risk factors, Neonatal mortality, Stillbirth

INTRODUCTION

According to the World Health Organization (WHO) in 2010, an estimated 270 000 deaths during the first 28 days of life were reported due to congenital anomalies globally. According to March of Dimes (MOD) global report on birth defects 7.9 million births (6% of total births) occur annually worldwide with serious birth defects. According to joint WHO and MOD meeting report, birth defects account for 7% of all neonatal mortality and 3.3 million under five deaths.[1]

The prevalence of birth defects in India is 6-7% which translates to around 1.7 million birth defects annually,

they account for 20-25% of perinatal deaths and 10-15% of neonatal deaths in India.[1,2] Early intrauterine period during 3rd to 8th week of gestation is the vital period of life for normal development of organs. Birth defects are structural, functional and metabolic disorders at birth. Various factors were identified like heredity factors, viruses and drugs but cause unidentified in 40-60% of cases.[3] Antenatal ultrasonography, fetal echocardiography and hematological investigations in mother are important tool for the diagnosis. Antenatal maternal risk factors also give clue for some malformations.

This study was done to determine the distribution and demographic characteristic of birth defects that will help to plan future strategies for prevention, early diagnosis and timely management.

METHODS

This is a prospective observational study done in a civil hospital Ahmedabad from 1st November 2010 to 31st October 2012. Prior permission from institutional Human Research Ethical Committee was taken. All the newborns were examined and assessed systematically for the presence of congenital anomalies at the time of birth. All the live neonates and stillbirth babies with birth defects were included in this study. Only hospital delivered babies were included. Outdoor patient and children with malformations were excluded. Written consent was taken from parents after explaining the purpose of the study.

For each case, a detailed antenatal and maternal history including the age of the mothers, parity, history of consanguinity was obtained by reviewing the maternal and labor ward records and by interviewing the parents. Maternal risk factors are identified, various hematological and radiological investigation were done as per the need. Birth defect were categorized according to system involved and whether single or multiple.

A marriage has been considered consanguineous, when it is occurred between a male and a female who are blood-related, e.g., between brother and sister, between 1st cousins etc. Birth weights <2.5 kg and <1.5 kg was termed as low birth weight (LBW) and very low birth weight (VLBW) respectively. Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as premature. Patients were referred to specialty services for immediate surgical management. Outcome in the form of morbidity and mortality were taken up to their hospital stay. Finally, all the data analyzed by Microsoft excel sheet and results were compared with other study.

RESULTS

This study was carried out at a tertiary care hospital over a period of 2 years. There were 9600 newborns delivered out of which 9064 live births and 536 stillbirths. Total 9600 births include 90 twins and one triplet delivery. Total no. of neonates with single malformation were 171(1.88%) and multiple malformations were 23 (0.25%). As shown in Table 1 incidence of malformations in stillbirth was higher (24.25/1000 live births) as compare to live births (19.96/1000 live births).

Table 1: Incidence of malformations in live births and stillbirth.

Cases	Births	Malformed babies	Incidence/1000 live births
Live births	9064	181	19.96
Stillbirths	536	13	24.25
Total	9600	194	20.20

Table 2 shows the correlation between birth weight and malformations. Incidence of birth defect was higher in babies with birth weight of <=1500 grams and 1501-2000 grams that was 57.28/1000 live births and 55.20/1000 live births respectively.

Table 2: Incidence of malformations in relation to birth weight.

Birth weight (gm)	Babies	Malformed babies	Incidence/ 1000 live births
<=1500	330	19	57.28
1501-2000	850	47	55.20
2001-2500	2100	66	31.4
> 2500	6320	62	9.81

Out of 194 babies with malformations, male babies 112 (57.7%) and female babies 82 (42.3%). Male to female ratio was 1.36. As shown in Table 3 incidence of malformation was more than 2 times higher in preterm babies as compare to full term delivered babies.

Table 3: Incidence of malformations in relation to maturity.

Maturity	Babies	Malformed babies	Incidence/1000 live births
Preterm	2101	76	36.17
Full term	7499	118	15.73

Regarding parity of the mothers 3980 were primi-paras and 5620 were multiparas. Incidence was higher in multiparas mothers. It has been seen that more than half of the mothers were aged 21-30 years. Incidence of congenital anomalies was 11.7/1000 live births for mothers age <=20 years, 17.31/1000 live births in 21-30 years and 34.18/1000 live births in >30 years (Table 4).

Maternal risk factors for congenital malformations were identified only in 26 (13.4%) cases, history of oligohydramnios in 8 (4.12%) babies, polyhydramnios in 3 (1.54%) babies, previous abortion in 5 (2.5%)babies

and eclampsia in 5 (2.5%)babies. Maternal diabetes was noted in 3 (1.54%) babies. History of third degree consanguinity was observed in 2 (1.03%) babies. Total 166 (85.1%) of pregnant women had taken regular antenatal visit but only 16.8% of babies with malformations were diagnosed antenatally.

Table-4. Incidence of malformation in relation to mother's age and parity.

Mother's age and parity	Babies	Malformed babies	Incidence/1000 live births
Age<=20 years	940	11	11.7
Age21-30 years	6700	116	17.31
Age>30 years	1960	67	34.18
Primiparas	3980	67	16.83
Multiparas	5620	127	22.59

As shown in Table 5 the predominant systems involved with malformations were cardiovascular system (23.4%) and musculoskeletal system (22.3%) followed by gastrointestinal (15.9%), genitourinary (15.4%) and central nervous system (9.7%). Acyanotic heart disease were most common amongst the all individual anomalies (19.1% of the total malformed babies) followed by CTEV (13.4% of the total malformed babies) followed by polydactyl (8.7% of all malformed babies).

There were total 31 babies with gastrointestinal system malformations out of which 17 babies (8 babies with trachea-esophageal fistula, 3 congenital diaphragmatic hernia, 2 jejunal atresia and 4 with ano-rectal malformations) required surgery immediately after birth but 13 babies expired in 3rd or 4th post-operative day the due to sepsis. In Central nervous system anomalies one baby with ruptured meningomyelocele was expired and all others discharged successfully. Maximum mortality occurred in babies with gastrointestinal system malformations (56.5% of the babies expired) followed by those with cardiovascular system malformations (21.7% of the all babies expired).

Majority of the babies with malformations were discharged (78.9% of the total malformed babies) only 11.8% of babies were expired and 2.6% of babies left against medical advice (LAMA).

Table 5: Systemic distribution of malformations and outcome.

System involved	Malformed babies (n=194)	Stillbirth (n=13)	Live birth (n=181)	Survived	Expired	LAMA
Cardiovascular	45(23.4%)	2(15.4%)	43(23.7%)	38(24.8%)	5(21.7%)	0
Musculoskeletal	43(22.3%)	0	43(23.7%)	43(28.1%)	0	0
Gastrointestinal	31(15.9%)	0	31(17.1%)	18(11.7%)	13(56.5%)	0
Genitourinary	30(15.4%)	3(23%)	27(14.9%)	26(16.9%)	0	1(20%)
Central nervous system	19(9.7%)	2(15.4%)	17(9.39%)	15(9.8%)	1(4.34%)	1(20%)
Multiple malformation	23(11.8%)	5(38.4%)	18(9.94%)	11(7.18%)	4(17.3%)	3(60%)
Miscellaneous	3(1.5%)	1(7.7%)	2(1.10%)	2(1.30%)	0	0
Total	194	13	181	153(78.9%)	23(11.8%)	5(2.6%)

DISCUSSION

Several studies in India and birth defect registry have addressed the prevalence of birth defects in the country.[4] Their frequency varies from 1.94% to 2.03% of birth.[5] In the present study incidence of congenital malformation in live births was 1.99% and stillbirths 2.42%. That was comparable with study by Taksande A which shows incidence 1.9% in live births and 4.68% in stillbirths. Singh A shows incidence 1.5% in live births and 8.7% in stillbirths. Malla B shows incidence 0.36% in live births and 2.0% in stillbirths.[6-8] In the present study only clinically identifiable malformations were taken, so some subclinical finding may be missed. That could be the reason for low incidence of malformations in stillbirth babies.

In the present study 39% of malformed babies were preterm and 60.5% babies were full-term. Study by Malla B8 and Dutta H showing the similar results (36% preterm and 64% full-term, 40.6% preterm and 59.4% full-term babies respectively).[9] In the present study 9.7% of malformed babies had birth weight <=1500 grams that was similar with study by Patel Z (9.8% of malformed babies).[10] In this study 68.1% of babies with malformations were low birth weight while 31.9% of babies with weight >2500 grams. Study by Patel Z showing the similar showing the similar results (59.8%)

babies with weight <=2500 grams and 40.2% babies with weight >2500 grams.[10]

In this study, male babies were more affected with malformations. 57.7% of total malformed babies were male and 42.3% female babies. Study by Taksande A and Dutta H showing the similar results (61% male babies and 37.4% female babies, 64.7% male babies and 34% female babies respectively).[6,9]

Incidence of malformation was higher (34.18/1000 live births) in mother aged of >30 years, that is comparable with study by Taksande A and Saiyad S (incidence of malformation 36/1000 live births and 20/1000 live births respectively).[6,11]

Taksande A reported higher incidence of malformations among the multiparas (19.5/1000 live births). In the present study incidence was 22.59/1000 live births. Our result is consistent with this finding, indicates a positive correlation between the birth order and the incidence of congenital anomalies.[6]

The most common systems involved in this study were Cardiovascular system (23.4%) and musculoskeletal system (22.3%), followed by gastro-intestinal tract (15.9%), genitourinary system (15.4%) and Central nervous system (9.7%). This was comparable with study conducted by Taksande A which shows cardiovascular system (23%), musculoskeletal system (21.9%), Gastrointestinal tract (14%), genitourinary (18.9%) and central nervous system (9.1%).[6]

Central nervous system malformations were predominantly seen in study by Saguna bai and Malla B (44% and 40% respectively).[8] Gastrointestinal system malformations are predominantly seen in study by Desai and Desai.[12,13] Differences between studies might be the effect of different racial, ethnic, and social factors in various parts of the world.

As congenital anomalies are 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 includes obtaining data on prevalence, nature of birth defects, genetic contributions, morbidity and mortality.

As majority of the health expenditure in India (69%) is from private sector, prevention and early detection of birth defect should be a priority. Prevention is practically feasible with the existing National health programs. Other measures include identifying risk pregnancies such as those in women >35 years of age, family history of birth

defects, maternal medical conditions, nutritional and lifestyle profile.[8] Antenatal diagnosis is possible with maternal biochemical screening and ultrasonography. Antenatal diagnosis with termination of affected pregnancies would be a cost saving strategy.

Due to their low prevention and high mortality, birth defects are not considered to be a significant health problem in India. There are no well-accepted preventive measures in India. Increasing awareness about maternal risk factors during pregnancy and educational programs on congenital malformations need to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

In tertiary care hospital, complicated cases are more commonly encountered. Hence prevalence calculated in this type of hospital-based study cannot be projected to the total population. Community based study should be ideal for true estimation of prevalence of congenital anomalies in a population. Another limitation of this study is the small size of the population.

congenital malformations represent one of the causes of neonatal mortality. Antenatal ultrasonography and maternal risk factors has important role to identify malformations. Early detection and timely management required to decrease the mortality.

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