ORIGINAL RESEARCH

Pattern of Congenital Malformations in Neonates: A Cross-sectional Study

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ABSTRACT

Background and Aim: Congenital anomalies are a worldwide problem, causing perinatal and infant deaths and postnatal physical disabilities. The present study was planned to highlight the pattern of congenital anomalies and its incidence, in new-borns. This study also included the assessment of various risk factors during antenatal and intranatal period, and their association with occurrence of congenital malformations in newborn babies, so that some preventive measures could be formulated. **Material and Methods:** A cross-sectional study was conducted at neonatal intensive care unit (NICU) of Medical College and Hospital, tertiary care institute of India. The study period was for the duration of 1 year. All the new born babies were thoroughly examined for the presence of congenital malformations. Detailed maternal history was recorded so as to evaluate association of various maternal risk factors with the congenital malformations. **Results:** During the study period, congenital malformations were detected in 120 new born babies. Out of this, 110 (91.6%) new born babies were live births and 10 (8.33%) were Intrauterine deaths. Most of the deliveries were full term 64 (53.3%) and only 56 (46.6%) were pre term delivered between 32 to 36 weeks of gestation. Among maternal and fetal risk factors; parental consanguinity, maternal under nutrition/obesity, positive history of a congenital anomaly (CA) in the family, and still birth/intrauterine deaths and ambiguous sex of the newborn baby were significantly associated with higher frequency of CAs ($p<0.05$). **Conclusion:** Congenital anomalies in this study were significantly associated with having inadequate antenatal care. With adequate antenatal care, pregnant women are often provided with health education on various issues such as the importance of proper nutrition, how to avoid teratogens, and prevention of maternal infections.

Key Words: Congenital Malformations, Gestation, Infant, Neonatal

INTRODUCTION

Birth defects, encountered frequently by pediatricians, are important causes of childhood morbidity and mortality. They are 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]

Birth defects can be classified according to their severity, system involved and whether they are involving a single system or multiple systems. It is a major public health problem globally. Annual estimates show the presence of serious birth defects in 7.9 million newborns.[2] Middle and low income countries account for more than 94% of births with serious birth defects.[4] Genetics is the etiological factor in (30-40%), environmental in 5-10% and cause is unknown in nearly 50% of the congenital malformed babies.[5] Worldwide surveys have shown the prevalence of congenital anomalies between 1.07-4.3/100 births.[6] Data from India shows the prevalence between 2-3%.[7] Ecological, social, racial and

economic factors play an important role in the geographical variation of birth defects. Considerable variation in frequency of CAs in different populations has been reported, from as low as 1.07% in Japan to as high as 3% in Taiwan. This wide variability could be due to the ecological variations of health facilities used for data collection in different studies.[8] The burden of this problem is still underestimated in the developing world due to the lack of healthcare diagnostic facilities in rural areas and lack of accuracy and adequacy of health care statistics.[9] The birth of a congenitally malformed neonate imparts enormous stress and burden to the affected families.[10] Identification of the various risk factors and creating public awareness can help to reduce the burden of this problem.[11] Early diagnosis of CAs by level 3 and level 4 antenatal ultrasounds in 1st and 2nd trimester of pregnancy are strong preventive measures.[12]

Knowledge about the prevalence of congenital anomalies is useful to obtain baseline rates, documenting changes over time, and identifying clues to the etiology of conditions. This knowledge is also helpful to plan and assess antenatal screening for congenital anomalies, especially for high-risk populations.[13] The present study was planned to highlight the pattern of congenital anomalies and its incidence, in new-borns. This study also included the assessment of various risk factors during antenatal and intranatal period, and their association with occurrence of congenital malformations in newborn babies, so that some preventive measures could be formulated. Further this study stressed on the importance of carrying out a thorough clinical examination of all newborn babies before discharge from hospital.

MATERIAL AND METHODS

A cross-sectional study was conducted at neonatal intensive care unit (NICU) of Medical College and Hospital, tertiary care institute of India. The study period was for the duration of 1 year. All the babies born during this period were included in the study. Ethical approval was taken from the institutional ethical committee and written informed consent was taken from all the participants.

INCLUSION CRITERIA

All the babies delivered at the institute were included in the study.

EXCLUSION CRITERIA

VLBW babies less than 1000 gm, babies delivered before 32 weeks of gestation (very preterm babies) and still born babies were excluded from the study.

Immediately after birth all the newborn babies were shifted to NICU and examined thoroughly by pediatrician on duty for the presence of any congenital malformations. All the babies were thoroughly examined at 24 hours and 48 hours of life to confirm the presence of birth defect and appearance of any new signs and symptoms related to CAs. Investigations like X-ray, ultrasound and echocardiography were done later for establishment of appropriate diagnosis. Based on International Classification of Diseases-10 (ICD-10) classification, system wise categorizations of all the CAs were done. Babies born before 37 weeks (less than 259 days) completed of gestation were considered as preterm, 37-41 completed weeks (259-293 days) as term NB and babies born after 42 weeks (after 294 days) of pregnancy were labeled as post term.[14] Depending on birth weight, neonate with birth weight less than 2.5 kg were classified as low birth weight (LBW), birth weight less than 1500 gm up to 1000 gram as very low birth weight (VLBW) and birth weight and less than 1000 gm as extremely low birth weight (ELBW) respectively.[14] Infants with birth weight 90th percentile of the expected weight for gestational age and sex of the newborn were considered as LGA babies. Infants with birth weight between 10th and 90th percentile were labeled as appropriate for gestational age (AGA) babies.[15,16] Neonate was considered as live birth when the product of conception, irrespective of weight or gestational age, and that, after separation from mother, shows signs of life such as breathing, heartbeat, pulsation of umbilical cord or definite voluntary muscles movements. When a product of conception that, after separation from mother, does not show any evidence of life is known as foetal death. A foetal death at a gestation of 22 weeks or more or weight 500 gram or more at birth is considered as stillbirths.[14] Detailed history was recorded from mother/attendant which included socio-demographic profile, age of both the parents, birth order, gestational age, mode of delivery, whether spontaneous conception or with treatment, family history of congenital malformation in siblings or in the family. Maternal history of fever with rash, exposure to drug/radiation, cigarette smoking and alcohol/substance abuse especially in 1st trimester of pregnancy was recorded. Details of medical/surgical ailments and any pregnancy related complications in mother were recorded. Details of antenatal visits and investigations done during this period with special attention to antenatal ultrasonographs for foetal well-being were recorded. Note was made of the any complication during intranatal period and delivery. History of consanguineous marriage was asked. Growth parameter record of the newborn included the weight, length and head circumference.

STATISTICAL ANALYSIS

The recorded data was compiled and entered in a spreadsheet computer program (Microsoft Excel 2007) and then exported to data editor page of SPSS version 15 (SPSS Inc., Chicago, Illinois, USA). For all tests, confidence level and level of significance were set at 95% and 5% respectively.

RESULTS

During the study period, congenital malformations were detected in 120 new born babies. Out of this, 110 (91.6%) new born babies were live births and 10 (8.33%) were Intrauterine deaths. Mode of delivery was normal in 70 (58.33%) and caesarean section in 50 (41.66%). Most of the deliveries were full term 64 (53.3%) and only 56 (46.6%) were pre term delivered between 32 to 36 weeks of gestation. Genders wise distribution of newborns with congenital malformations shows 65 (54.16%) were males, 55 (45.83%) were females. (Table 1) Average weight of the NBs with CAs recorded was 2.70 kg, average head circumference was 33.1 cm and average length was 48.2 cm. Age distribution of mothers showed that maximum number 95 (79.1%) of mothers were in the age group of 21 to 30 years; followed by 31-40 years 16 (13.3%) and only 9 (7.5%) mothers were less than 20 years of age group. Majority of neonates were 1st birth order 62 (51.6%) and 2nd in 58 (48.33%) in birth order. Conception was spontaneous in all the mothers. History of pregnancy induced hypertension was recorded in 29 (24.16%). They were taking drugs for high blood pressure in last trimester only.

Antenatal ultrasound showed congenital anomalies in 61 (50.83%) mothers. Anomalies detected on antenatal ultrasonography were those pertaining to musculoskeletal, genitourinary and cardiovascular system. The most common system involved was musculoskeletal 53 (44.16%) followed by central nervous system; 34 (28.33%) and genitourinary system in 10 (8.33%) study population (Table 2). There was history of siblings affected with congenital malformations in 16 (13.3%). Affected siblings had involvement of musculoskeletal system manifested as congenital talipes equinovarus. It is statistically significant. Congenital anomalies in new born with ambiguous genitalia were not compatible for life and caused intrauterine death of the baby. This was statistically significant.

Table 1: Demographic Distribution of study participants

Variable	Number	Percentage (%)
Mode of Delivery		
Normal	70	58.33
Caesarean section	50	41.66
Type of Delivery		
Full term	64	53.3
Pre term	56	46.6
Gender		
Male	65	54.16
female	55	45.83

Table 2: Distribution of congenital malformations based on ICD-10 classification

System/malformation	Number	Percentage (%)
Musculoskeletal system	53	44.16
Congenital talipes equinovarus	13	10.83
Right foot	12	10
Left foot	9	7.5
Both sides	17	14.16
Achondroplasia	3	2.5
Polydactyle	9	7.5
Nervous system	34	28.33
Hydrocephalous	14	11.6
Meningomyelocele + spina bifida + hydrocephalus	6	5
Spina bifida with chest wall deformity	2	1.66
Anencephaly	2	1.66
Anencephaly + encephalocoele + gastrochiasis	4	3.33
Corpus calosum absent	3	2.5
Microcephaly	3	2.5
Genitourinary	10	8.33
Multicystic dysplastic kidney	7	5.33
Undescended testis	3	2.5

DISCUSSION

As improvement in antenatal, perinatal and neonatal care, lead to reduction in neonatal infections, preterm births and number of LBW babies. All these factors helped in reduction of neonatal morbidity and mortality due to neonatal sepsis and congenital pneumonias.[17] However, the proportion of neonatal deaths due to congenital malformations is increasing. Hence in coming period, congenital malformations will be the leading cause of neonatal morbidity and

mortality.[18] With improved control of infections and nutritional deficiency diseases, CAs have become important causes of perinatal mortality in developed countries and will very soon become increasingly important determinates of perinatal mortality in developing countries.

Congenital malformation were more in newborns with birth order of 4 or more, but in present study Majority of neonates were 1st birth order 62 (51.6%) and 2nd in 58 (48.33%) in birth order.[18] In our study CAs were more in preterm 2.40% of total deliveries as compared to term babies (2.39%) which was comparable to earlier studies which showed higher incidence of congenital malformations in preterm babies.[18] In a study from Lebanon, family history of congenital malformations was present in 12 (13.1%) of study population, similar results were obtained in our study also.[19] 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.[20,21] 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).[22,23]

There was a statistically significant association between maternal malnutrition, history of congenital malformations in family in present study. Luck in his study value of routine ultrasound scanning at 19 weeks found that some CAs were not compatible for life lead to intrauterine devices (IUDs) or still births.[24] Similar results were obtained from present study, neonate with ambiguous genitalia had 100% mortality (IUD). Cassell and Golden studied maternal obesity as a risk factor for the development of CAs in the newborn.[14] They reported that maternal obesity is significantly associated with an increased risk of selected birth anomalies such as spina bifida and heart defects.

Systems involved were musculoskeletal, genitourinary and cardiovascular. Similar anomalies were detected on antenatal ultrasound i.e. gastroschisis, extra renal dilated pelvis with mild calyceal separation, left multicystic dysplastic kidney and cardiomegaly.[25-27] A study by Bai in year 1982 reported that CAs were more in low birth weight infants and in neonates born to mothers with age above 35 years but opposite results were obtained in our study, majority of mothers of congenitally malformed babies were in age group of 21-30 years and no specific birth weight group predilection recorded.[11] This suggests focusing more on younger age group mothers in the form of preventive health care measures like compulsory institution of healthy diet rich in folic acid and other nutrients to women who are planning pregnancy and in first trimester of pregnancy. Antenatal ultrasound for detection of congenital malformations in early pregnancy should be available at a nearby health facility, so as to reduce the burden of problem. Hence health awareness among the women in reproductive age group should be provided at all levels of health care.

CONCLUSION

Congenital anomalies in this study were significantly associated with having inadequate antenatal care. With adequate antenatal care, pregnant women are often provided with health education on various issues such as the importance of proper nutrition, how to avoid teratogens, and prevention of maternal infections. The birth of a congenitally malformed baby imparts enormous stress to the affected families. Institution of preventive measures with more focus on young mother's nutrition, provision of health education and early diagnosis of congenital malformations during antenatal period can help to curb the burden of this problem.

SOURCE OF FUNDING

None

CONFLICT OF INTEREST

None

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