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STUDY OF CONGENITAL MALFORMATION IN NEONATES BORN AT TERTIARY CARE HOSPITAL

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ABSTRACT

Introduction: Congenital malformations are major contributors of neonatal mortality or lifelong disability. Early diagnosis and early surgical treatment when required can prevent neonatal deaths and help for better survival.

Method: An observational prospective study was carried out at a tertiary care hospital. All neonates delivered from 1st July, 2007 to August, 2009 including still borns were included in the study. Detailed maternal history was taken, complete clinical examination and necessary investigations were carried out to look for congenital malformations. Babies were followed up till one week of life. Data was recorded and analysed with standard statistical software.

Results: Out of 6652 babies, 113 (1.69%) babies were diagnosed having congenital malformations. Amongst them, 98 were live births and 15 were still borns. Out of 113 babies, 34 babies (30.04%) were diagnosed antenatally. CNS malformations were the most common to be antenatally diagnosed. At the end of first week of follow up, 22 out of 98(22.44%) expired, 73(74.48%) survived and 3(3.06%) were lost in follow up.

Conclusion: Incidence of congenital malformations was significantly high in still born babies. Factors like prematurity, low birth weight, birth order four or more, liquor abnormalities and maternal age more than 35 years were found to carry higher risk for congenital malformations. Musculoskeletal system was most commonly affected. Congenital talipesequinovarus was the commonest malformation.

Keywords: Congenital malformation, neonate, musculoskeletal system, risk factors

INTRODUCTION

Congenital anomaly is an internal or external structural defect that is identifiable at birth[1]. Subtle interplay between genetic and environmental factors affects morphogenesis during early fetal life that may result into malformation, deformation, disruption or dysplasia[2] that eventually cause congenital anomalies. They are responsible for 9% of neonatal deaths[3]. It is estimated that 2.5% neonates have recognizable malformations at birth and they contribute up to 40% of surgical

pediatric admissions[4]. As tremendous efforts are being carried out to reduce mortality by improvement in perinatal and neonatal care, congenital malformations may become an important cause in further reduction in perinatal mortality in upcoming decades. Economical and psychological burdens of parents with malformed babies are beyond all imaginations. Early diagnosis and early surgical treatment when required can prevent neonatal deaths and help in better survival. Prenatal detection of an anomaly increases options of pregnancy management. Knowledge of association of various maternal risk factors helps for early diagnosis and organization of referral when indicated. This also helps in planning of preventive measures by health education and identification of high risk groups. The importance of congenital malformations is clearly recognized in the fields of both clinical medicine and public health. Hence, the need for a systemic approach to congenital malformations arises. So this study was opted to know the incidence, associated risk factors, various systems involved, antenatal diagnosis and outcome of congenital malformations.

METHODS

This prospective study was carried out in Department of Pediatrics of a teaching hospital of Ahmedabad, Gujarat. All intramural deliveries from 1st July, 2007 to 31st August, 2009 including still births comprise the study material. All neonates were thoroughly examined by an expert pediatrician within first 24 hours of birth. Detailed history including sex, birth weight, birth order, maternal age, maternal disease, maternal drug intake, previous bad obstetric history, substance abuse, consanguinity and maternal antenatal investigations including antenatal ultrasonography were noted down in Performa. Relevant radiological and hematological investigations were carried out including 2D echo and neuroimaging wherever indicated. Malformations were classified according to the systems involved as musculoskeletal, central nervous system, cardio vascular system, gastrointestinal system, genitor urinary system, multiple malformations and miscellaneous malformations. Babies were followed up till 7th day of life and outcome was noted as survived, expired or lost in follow up. Statistical analysis was done using appropriate tests. The study was concluded in August 2014.

RESULTS

There were total 6550 deliveries during study period including 239 still births. Total 6652 birth also include 101 twins and 1 triplet delivery. Incidence of congenital malformation was 113/6652(1.69%).

Congenital malformation were significantly high in still born babies as compared to live born babies(p value < 0.05).Out of 113 malformed babies 59(52.21%) were male and 54(47.79%) were female. There was no sex predilection in Congenital malformation (p value > 0.05). Frequency of Congenital malformation in preterm babies was 9.04% (37 out of 409) and in full term babies it was 1.21% (76 out of 6243). Frequency in preterm babies was 7.5 times higher than term babies which was statistically significant (p value < 0.05%).

Congenital malformations were highest amongst very low birth weight babies, followed by large for gestational age babies. Incidence of congenital malformation was highest (32.25/1000 births) in babies born to mothers with age more than 35 years. Incidence was also higher in babies born to mothers with age < 20 years (21.85/1000). Incidence of congenital malformations was 15.82, 16.64 and 13.93 per 1000 births in maternal age groups of 21-25 years, 26-30 years and 31-35 years respectively. Incidence of congenital malformation was 16.34 in primigravida, 13.18 in 2nd gravida, 17.17 in 3rdgravida and 55.94 in 4th or higher gravida mothers per 1000 births respectively. So incidence of congenital malformation was highest in 4th or higher gravida mothers.

History of parental consanguinity was present in 3/113 (2.6%) malformed babies. History of sibling with congenital malformation was present in 6/113(5.31%) babies. Previous history of still birth was present in 6/113(5.31%) babies. Congenital talipesequinovarus was commonest anomaly amongst musculoskeletal malformations. Out of 23 babies with multiple malformations 19 were live births and 4 were still born. 6 out of 23 babies with multiple malformations were of down's syndrome out of which 4 babies had associated congenital heart disease. 9 cases of Miscellaneous malformations included sacrococcygealteratoma, jaw cyst, sacral dimple, ear tag, deformed ear, ear canal stenosis and anotia. History of sibling with congenital malformation was present in 6 out of 113(5.31%) babies.

Table 1: Frequency of congenital malformation in live birth and still birth

Cases	Births	Malformed babies (%)
Live birth	6413	98 (1.52)
Still birth	239	15 (6.27)
Total	6652	113 (1.69)

Table 2: Incidence of congenital malformation at different birth weights

Birth weight (in gm)	Babies	Malformed babies (%)	Incidence per 1000 births
< 1499	267	17 (15.04%)	63.67
1500-1999	541	20 (17.70%)	36.96
2000-2499	1506	29 (25.66%)	19.25
2500-3999	4319	46 (40.71%)	10.65
>4000	19	1 (0.89%)	52.63
Total	6652	113 (100%)	

Table 3: Maternal Risk Factors and Congenital Malformations

Risk factor	Malformed babies	% of total malformed babies
Pre eclampsia	12	10.62
Severe anemia	21	18.58
Polyhydramnios	11	9.73
Oligohydramnios	12	10.62
Consanguinity	3	2.65
Infertility	3	2.65
Twins	5	4.42
Maternal fever during 1st trimester	4	3.54

Out of 113 babies, 34 babies (30.04%) were antenatally diagnosed by ultrasound examination. Central nervous system malformations were the most common to be antenatally diagnosed and musculoskeletal malformations were the least common to be antenatally diagnosed.

All 98 live born babies were followed up till one week of life. Out of these 98, 52 (53.06%) were male and 46 out of 98 (46.93%) were female babies. At the end of 1 week of follow up 22 expired, 73 survived and 3 babies were lost in follow up. Out of the 22 expired babies, 13 were male and 9 were female babies. Mortality at 1 week follow up was highest amongst babies having CNS and CVS malformations.

Table 4: Antenatal Diagnosis of Congenital Malformations

System involved	Malformed babies	Antenatally diagnosed
Musculoskeletal	31	1 (3.2)
CNS	19	16 (84.2)
GIT	19	3 (15.8)
GUS	10	3 (30.0)
CVS	2	0 (0)
Multiple	23	9 (39.1)
Miscellaneous	9	2 (22.2)
Total	**113**	**34 (30.04)**

Figure in parenthesis indicate percentage

Table 5: Systemic Distribution of Congenital Malformations and Outcome

System involved	Malformed babies (n=113)	Still Birth (n=15)	Live birth (n=98)	Follow up on 7th day		
				Expired	Survived	Lost to Follow up
Musculoskeletal	31 (27.43)	0	31 (31.64)	4 (12.90)	27 (87.10)	-
CNS	19 (16.81)	9 (60)	10 (10.20)	5 (50)	4 (40)	1 (10)
GIT	19 (16.81)	2 (13.33)	17 (17.35)	3 (17.65)	13 (76.47)	1 (5.88)
GUS	10 (8.85)	0	10 (10.20)	1 (10)	9(90)	-
CVS	2 (1.77)	0	2 (2.04)	1 (50)	1(50)	-
Multiple system	23 (20.35)	4 (26.67)	19 (19.39)	6 (31.58)	13 (68.42)	-
Miscellaneous	9 (7.96)	0	9 (9.18)	2 (22.22)	6(66.67)	1 (11.11)

Figure in parenthesis indicate percentage

DISCUSSION

Significance of congenital malformation lies not only in their contribution to neonatal and perinatal mortality but, also in causing disabilities and handicaps in infant and children. Incidence of congenital malformation in present study was 113 out of 6652 (1.69%) as compared to 1.5% in Arjunsingh's study , 1.9% in Aman's study and 1.78% in N. Gover's study.[6,7,8] Incidence of congenital malformation was highest (63.67 per 1000 births) amongst very low birth weight babies followed by (52.63 per 1000 births) large for gestational age babies. Patel's study has also documented higher incidence of congenital malformation at extremes of weight.[9]These may be because factors causing malformation may also affect somatic growth. Present study has documented marginally higher incidence of malformation in male babies than female babies. Aman's study has also documented similar results.[7] Incidence of congenital malformation was

90.46 and 12.17 per 1000 birth in pre term and full term babies respectively. Stastically significant higher incidence of malformation in preterm babies was also reported by Patel and Aman's study[7,9]. Higher maternal age is a well known risk factor for malformations. Highest incidence of malformation was (32.25 per 1000 birth) in babies of mothers more than 35 years of age. Swain's study has also documented highest incidence (16.6 per 1000 births) in mothers more than 35 years of age[11]. Higher maternal age is a well known risk factor for down's syndrome[4]. Out of 3 babies born to mother with age more than 35 years, 2 had down's syndrome in the present study. Previous study has reported highest incidence of congenital malformation in babies of birth order 4 or more [11]. Present study has also reported highest incidence (55.94 per 1000 birth) in babies born to mothers who were 4th or higher gravida. Out of 11 malformed babies born to mother with history of polyhydramnios, 4(36.36%) had gastrointestinal and 4(36.36%) had central nervous system malformations. So gastro intestinal and central nervous system malformations were common with polyhydramnios. Out of 12 malformed babies born to mother with history of oligohydramnios, 6(50%) had congenital malformations of musculoskeletal system. Dutta's study has reported maternal history of liquor abnormality as statistically significant in causation of congenital malformations[12]. 30 out of 113 (30.04%) babies were diagnosed antenatally by ultrasound. With the advent of newer therapeutic modalities including fetal medicine, early antenatal diagnosis may help in some cases that are amenable to intrauterine treatment. Central nervous system malformations were the most common to be diagnosed antenatally (16 out of 19, 84.21%) while musculoskeletal malformations were the least common to antenatally diagnosed (1 out of 31, 3.2%). In Eurofetus study also best detected anomaly were of central nervous system (88.3%) and the lowest rate of detection were for musculoskeletal system (18%)[13]. Musculoskeletal system abnormalities were commonest one but least commonly antenatally detected. This may be because of oligohydramnios present in babies of mother with musculoskeletal anomaly, as oligohydramnios has negative effect of accuracy of antenatal ultrasound examination[14].

Incidence of congenital malformation was 98 out of 6423 (1.52%) and 15 out of 239 (6.27%) in live birth and still birth respectively. So statistically significant higher incidence of congenital malformation was seen in still born babies, which was comparable to 4.46% in Arjunsinghs study and 6.06% in chaturvrdis study respectively[6,10]. This may be because severe malformation may result in still birth. Incidence of congenital malformations in pre term babies was 37 out of 409 (9.06%) and in full term babies was 76 out of 6243(1.21%). So, significantly higher incidence of congenital malformations was found in pre term babies as compared to full term babies which is comparable to Patel's study[9].

Present study has reported highest numbers of congenital malformations were of musculoskeletal system followed by multiple malformations. Congenital talipesequinovarus was the commonest congenital anomaly. Commonly diagnosed system with congenital malformation may vary as per available diagnostic modalities at different institutes. Aman's study has reported cardiovascular system anomalies as commonest[7]. In Aman study 2D echo of all the babies of mothers with risk factors was carried out within first 24 hour of birth while in our study 2D echo was carried out of those babies who presented with clinical signs and symptoms suggestive of congenital heart disease within first week of life or who were diagnosed antenatally, as bedside 2D echo was not available.

CONCLUSION

Frequency of congenital malformations is significantly high in still born babies as compared to live born babies. Factors like prematurity, low birth weight, birth order four or more, liquor abnormality and maternal age more than 35 years were found to carry higher risk for congenital malformations. Musculoskeletal system was most commonly affected and congenital talipes equine varus was the commonest malformation. Only 34 out of 113 (30.04%) babies were diagnosed antenatally underlying the need for better antenatal diagnostic facilities. CNS malformations were the most common antenatally diagnosed malformations. At one week follow up, highest mortality was noted amongst the babies having CNS and CVS malformations.

LIMITATIONS OF STUDY

Antenatal ultrasonography of all mothers were not done as some of them were not registered for antenatal care. Specific investigations e.g 2D echo, neuroimaging were done only in babies

with clinical signs and symptoms. so some sub clinical malformations may have remain undetected. Genetic tests and autopsies were also not done.

REFERENCES

1. Michele C walsh, Fanaroff and martin's neonatal perinatal medicine,9th edition, Elseveir, Mosby missouri 2010, 29, 531.
2. Smith; Recognizable patterning of human malformation kennethlyons jones, Elseveirsaunders, philadelphia2004 ,1, 2.
3. K.Park (MBBS, MS), Park textbook of preventive and social medicine, 22 nd edition, Bhanot publisher, Jabalpur India, 2013. 523.
4. Kliegman, Stathon, Behrman, Nelson text book of pediatrics ,19th edition, 2012, ElseveirSaundey, philadelphia, pg 0629
5. B. Vishnu, Bhat ,Lokeshbabu et al ., Congenital malformation at birth. A prospective study from south India. Indian J. Pediatrics 1998; 65; 873-878.
6. Arjunsingh and Ravindra Gupta, Pattern of anomaly in new born; A hospital based prospective study ,J.K.science vol.11, Jan-March 2009. 34-36.
7. AmanTaskade, Krishna, Vilhelar at el. Congenital malformation at birth in central India, A rural medical college based data. Indian Journal of human genetics 2010. Sep-Dec. 16.159-163.
8. N groven . Congenital malformation in Shimbi, Indian J. pediatrics 2000; 67, 240-250.
9. Patel Z. M., Adhia R.A., Birth defect survillance study Indian J. Pediatrics Volume 72 June 2005; 489-490.
10. Chaturvedi P, Banerjee K. S, An epidermiologcal study of congenital malformation in newborn, Indian Journal Pediatrics 1993; 60, 645-653.
11. Swain S, Agrawal A, Bhatia B.D., Congenital malformation at birth, Indian Pediatrics 1994, 31, 1187-1191.
12. Dutta V Chaturvedi P. Congenital malformation in rural Maharashtra, Indian paediatrics 2000; 37: 998-1000
13. Grandjean H, Haroque D et al, The performance of routine ultrasonographic screening of pregnancies in the eurofetus of pregancies in the eurofetus study, Am j. ObstractGynecol 1999:181;446-454.
14. Carol M rumack, Stephanic Wilson at el.diagnostic ultrasound. 4th edition, Elsevier Mosby, Philadelphia. 2011. 28; 1056-1057.