Original Research Article

Clinical profile of neonates with congenital malformation born at a tertiary teaching hospital in a Himalayan state of India

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ABSTRACT

Background: Congenital malformations are assuming prominence as a contributor to neonatal and childhood mortality. This study is aimed at identifying the pattern and profile of congenital malformations among neonates delivered in a tertiary teaching hospital in Sikkim which will provide baseline data for future studies.

Methods: A hospital based cross sectional observational study was conducted from 15 October 2015 till 15 April 2017 among live newborns delivered in the only teaching hospital of Sikkim. Out of 2521 neonates delivered, 96 neonates met the inclusion criteria. These neonates underwent a detailed clinical evaluation and appropriate investigations. Data collected were entered into Microsoft excel worksheet 2017, SPSS (version 22) was used for data analysis.

Results: The occurrence of congenital malformations in the hospital was 3.8%. Neonates with major anomalies were 50% and majority of the anomalies were of genitourinary system (23.96%). The relation between gender and congenital malformation was found to be statistically significant with p value <0.05, predominantly affecting male. Only 6.25% of the malformations were diagnosed prenatally.

Conclusions: This study gives data on the pattern and profile of malformation from a geographically isolated place in the Himalayan regions of India which showed some significant variation. It also highlights the lack of adequate facilities for prenatal diagnosis of congenital malformation in these regions.

Keywords: Congenital malformations, Live neonates, Patterns, Prenatal diagnosis

INTRODUCTION

Congenital malformations, as per WHO fact-sheet of September 2016, is defined as structural or functional anomalies that occur during intrauterine life and can be identified prenatally, at birth or sometimes detected later in infancy.[1] The prevalence in birth defects in India is 6-7% which translates to around 1.7 million annually.[2] Congenital malformation is the fifth leading cause of neonatal mortality in India, accounting for 9% of neonatal deaths and in the coming decades it is going to be one of the leading causes of morbidity and mortality among neonates.[3]

The cause of congenital malformations in humans is still elusive but some are contributed by genetic and environmental causes.[4] The prevalence and pattern of congenital malformations varies between regions and over time. The most common body systems involved in congenital malformations include musculoskeletal, central nervous system, gastrointestinal system and

cardiovascular system with the least affected system being the urogenital system.[5-7]

It is important to do epidemiological surveys in various parts of the world with different environment, socioeconomic status and ethnic groups with widely varying habits which will give information on the pattern and factors for congenital anomalies in different areas. So far, there is no scientific data regarding the pattern and factors associated with congenital anomalies in Sikkim, a Himalayan state of India with difficult terrain and limited health resources. It is one of the North Eastern states of India sharing international borders with China, Nepal and Bhutan. The population is ethnically distinct from the people of other regions of India.

The objective of this study was to describe the occurrence and pattern of congenital malformations in live neonates delivered in the only teaching hospital of Sikkim which would provide a baseline data for future research.

METHODS

This hospital based observational, cross-sectional study was carried out in the department of Pediatrics of Central Referral Hospital (CRH), teaching hospital of Sikkim Manipal Institute of Medical Sciences (SMIMS), Gangtok. Gangtok is the capital of the state of Sikkim. Sikkim Manipal Institute of Medical Sciences is the only medical college in Sikkim. After the approval of Research Protocol Evaluation Committee and Institutional Ethics Committee of SMIMS, the study was conducted from October 2015 to April 2017.

The decisions of the parents/caretakers were respected and after the acquisition of an informed consent, the live newborns with congenital malformations fulfilling the inclusion criteria were included in the study and those who failed to do so, were excluded.

After stabilization following birth, the new-borns were examined systematically for the presence of any congenital malformations and followed up till discharge. Diagnosis of the congenital malformations was based on thorough clinical evaluation of new-borns. A face to face interview was done with the mother and caregiver of participating new-borns to collect socio-demographic data and clinical details of antenatal period and visits. Investigations such as radiography, ultrasonography, echocardiography and blood tests were done as per the need of clinical diagnosis. Anomalies were classified as per the European Surveillance of congenital anomalies (EUROCAT) as major and minor.[8]

Data collected were entered in Microsoft Excel Work sheet 2017 which was imported to SPSS (version22) for data analysis. Excel and SPSS were used to create tables and figures. Percentages, proportions, means, charts and tables were used for description of the data. Analysis of qualitative data was done using Chi-square test with

Yate's correction and level of significance is expressed as p value <0.05.

RESULTS

During our study period, 2521 babies were born; out of which 96 neonates were documented to have congenital anomalies hence the occurrence was found to be 3.8%. 50% of the newborns were affected with major defects. The spectrum of congenital malformations seen was that the most common system involved was genitourinary system (23.96%). Out of the genitourinary defects hydrocele was found to be the commonest followed by hydronephrosis, undescended testes, hypospadias. The other anomalies seen were minor cutaneous defects (17.71%), cardiovascular system (16.67%), musculoskeletal system (12.5%), and gastrointestinal system (10.42%) (Figure 1).
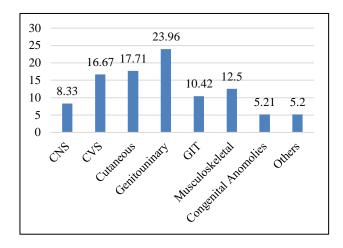

Figure 1: Pattern of congenital anomalies

Out of 96 affected neonates, 64 were male and leading at a ratio of 2:1 from their female counterpart. The relation between gender and congenital malformation was found to be statistically significant with p value <0.05(0.0037). Chi square 8.448, RR=1.875, CI=1.236 to 2.846 (Table 1).

The mean birth weight (kg) was 2.97±0.56SD (Standard deviation), with a minimum value of 1.52 Kg and a maximum value of 4.19 Kg. The mean gestational age (weeks) at delivery was 38.47±1.80SD, with minimum gestational period being 31 weeks and maximum as 42 weeks (Table 2).

Majority of the neonates with congenital malformation were Appropriate for gestational age (Figure 2).
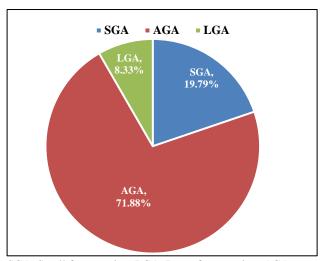
The frequency of congenital anomalies was highest amongst the maternal age group of 21-24 years (75%). The mean age was 28.32±5.60 SD years, with minimum age being 14 and maximum being 40 year. More than 30 % of the mothers had irregular antenatal visits, 8% of mothers had not taken folic acid tablets adequately during

antenatal period, 5.88% of mothers had history of previous abortion, 1.07% of the mothers had diabetes, 2.08% had multiple pregnancy. 5.2% of the babies were born by artificial conception (Table 3).

Table 1: Distribution of cases based on gender.

Gender	Congenital Malformation		Normal		Total	
	Count	Percentage	Count	Percentage	Count	Percentage
Male	64	3.00	1237	49.00	1301	52.00
Female	32	1.00	1188	47.00	1220	48.00
Total	96	4.00	2425	96.00	2521	100.00

SGA: Small for gestation, LGA: Large for gestation, AGA: Appropriate for gestation

Figure 2: Distribution based on weight for age.

Table 2: Distribution of cases with respect to weight and gestational age.

Weight(g)	Number (n=96)	Percentage(%)
<=1500g	0	0
1501 - 2000g	3	3.13
2001 - 2500g	20	20.83
2501 - 3000g	33	34.38
3001 - 3500g	21	21.88
>3500g	19	19.79
Gestational age		
<35 weeks	4	0.0417
35-37 weeks	17	0.1771
38-40 weeks	69	0.7188
>40 weeks	6	0.0625

In this study, about 55% of reported cases were of first birth order and only 2 had family history of congenital anomalies. Out of 96 neonates, 11 (12%) expired in the neonatal period. Congenital malformation was diagnosed in only six (6.25%) of the neonates in the antenatal period (Table 4).

Table 3: Distribution of cases with respect to various maternal factors.

Maternal Profile (96)	Yes	No
Booked Case	66	30
Folic Acid Intake	89	7
Multiple Pregnancy	2	94
Assisted Conception	5	91
Diabetes Mellitus	1	95
Consanguinity	2	94
Previous Abortion	6	96

Table 4: Distribution of cases in relation to prenatal diagnosis.

Prenatal diagnosis	Numbers (n=96)	Percentage (%)
Yes	6	6.25
No	90	93.75
Total	96	100.00

DISCUSSION

The incidence of congenital anomalies worldwide is estimated at 3-7%, but actual numbers vary widely between countries.[9] There is paucity of data from India as there is no systematic surveillance system for congenital malformations. There is no scientific data regarding congenital anomalies from this region till date. This study will provide baseline data for planning preventive and management policies for these disorders.

The occurrence of congenital anomalies in this study was found to be 3.8%. A similar study was done, to look for the pattern of congenital anomalies in neonates delivered in a hospital in Kathmandu and the incidence was note as 0.36% among the total live births, which is comparatively less.[7] The reason could be primarily because our hospital is the only tertiary/referral center in the state which offers pediatric surgery care. A study done by Wills V et al, in Kerala showed that 149 mothers gave birth to 151 newborns with congenital anomalies in their study period.[10] Another research by Sarkar S et al, in a tertiary center in East India revealed the incidence to be 2.22%.[11]

These variations in prevalence of Congenital anomalies in various parts of the world might be explained by social and racial influences that are commonly known in genetic disorders. Also, the results may vary according to the background of the investigators, the type of sample chosen and the period of study.[12]

Regarding the pattern of congenital malformation in this study, the most common single system involved was genitourinary system. This was in accordance to a study conducted by Wills V et al, which showed that anomalies related to genitourinary system were most common followed by other system related anomalies.[10] However, many studies reported musculoskeletal anomalies to be most common whereas studies done by Patel KG et al, reported cardiovascular system related anomalies to be most common.[9,11,13,14] Hence the spectrum of malformations varies in different regions.

In this study the relation between congenital malformation and gender was found to be statistically significant with p value <0.05. Male preponderance has been reported in many studies done in country like Nepal and in Eastern India too.[7,11-13] The reason for greater numbers in male population is thought to be caused by the fact that male embryos are more vulnerable to oxidative stress which could partly be explained by the biological fragility of the male embryo. Oxidative stress has been implicated in the pathogenesis of several congenital anomalies.[15]

Our study showed that babies weighing 2.5 to 3 kg were maximally affected with congenital anomalies. This is in contrast with most studies which usually reports higher incidence among low birth weight babies.[16,17] The reason could be because of the non-inclusion of stillbirths in this study who, most of the time turn out to be either premature or IUGR babies. Study done by Malla BK. et al, in Kathmandu, reported that the incidence of congenital anomalies was significantly higher in full term babies with low birth weights as compared to pre-term babies.[7] In this study, the incidence was higher in babies delivered at 38-40 weeks with 71.88%.

Advanced age of the mother didn't seem to influence the incidence of congenital anomalies in this study. This is in agreement with study done by Sarkar et al, and Wills V.[11] et al, where most of the mothers (55.7% and 69.8% respectively) belonged to the age group between 21 and 30 years.[10] Many studies report a very close and significant association of congenital anomalies with advanced maternal age which was in contrast with our study which could be explained by the fact that we included only the absolute numbers and did not include the rest of the mother in these age groups who delivered normal babies.[13,17,18]

Only 2% of mothers of neonates with congenital anomalies in this study had multiple pregnancy which indicated an insignificant association. Similar studies done in hospitals or maternity care found no significant association.[7, 19] In this study 5% newborn with anomalies were conceived with assisted conception methods as most of the mothers in this study belonged to 21-30 years of age where difficult conception is not usually encountered.

Even though more than 65% of the mother of these newborns was booked, antenatal diagnosis was noted in 6.25% and the results were informed much later in pregnancy where termination was not possible. As most of the cases were referred since it's the only tertiary center in state, it is difficult to gauge how effective the anomaly scans are in remote parts of the state and even more difficult to comment on how many of them had undergone the scan. Moreover, as there were no facilities available for fetal echocardiography, majority of cardiovascular defects were missed during neonatal period.

A study like this further reinforces the importance of regular and effective antenatal visits. In a country like India, with limited resources at hand, providing an anomaly scan is not always possible. Numerous studies have proven how an effective antenatal scan can help reduce the infant mortality rate of the nation. Study done by Liu S. et al, in Canada has concluded that increases in prenatal diagnosis and pregnancy termination for congenital anomalies are related to decreases in overall infant mortality at the population level.[20] Similar study done in Europe showed that prenatal diagnosis does affect the Infant Mortality Rate.[21] Study done in Eastern India by Shatanik Sarkar et al, has stressed upon the importance of prenatal diagnosis.[11] Prenatal diagnosis helps to be prepared for better management. A study done by Agarwal A. et al, has shown the importance of antenatal diagnosis of anterior abdominal wall defects on patient management and prognostication.[22] Similarly, study done in AIIMS, New Delhi on management of antenatal hydronephrosis has shown the importance of antenatal scan on the outcome of the patient's morbidity.[23]

CONCLUSION

The study has highlighted the pattern and profile of congenital malformations among neonates delivered in the only tertiary teaching hospital of Sikkim. The importance of antenatal diagnosis to both parents and doctors needs to be stressed upon, as it's the only preventive measure to reduce the incidence of these malformations. This study reiterates the fact that there is still a lack of essential modern antenatal facilities in most regions of our country. Hence, there is a need for better provision of technology and trained personnel, which will help in better and early management of these new-borns.

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