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Patterns of Clinically Identifiable Congenital Defects in Neonates

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ABSTRACT

Background: The study is aimed at highlighting the pattern of congenital defect in a tertiary care hospital. Congenital anomalies are recognized as a growing cause of neonatal morbidity and mortality in developing countries and a major cause of distress to parents.

Methods: This was a prospective descriptive study conducted between September 2019 and August 2020 with the objective to determine the types of congenital anomalies among live born neonates at Manipal Teaching Hospital (MTH), Pokhara and to determine their immediate outcome. Neonatal and maternal characteristics were noted.

Results: Twenty four out of 2515 live births had congenital anomalies during the study period, giving an incidence rate of 9.42 congenital anomalies per 1000 live birth per year. Single system involvement was seen in 79.2 % cases, remaining 5 (20.8%) neonates had involvement of more than one system; 54.2% of these newborns were discharged, 33.3% expired, 8.3% left against medical advice and 4.2% were referred out.

Conclusions: This study highlights the importance of clinical examination of neonates to detect anomalies in our setting.

Keywords: Congenital anomalies; incidence; newborn

INTRODUCTION

Congenital anomalies are emerging cause of perinatal and neonatal deaths. Global estimates of neonatal mortality due to birth defects according to World Health Organization in 2010 was 7%.[1,2]

The March of Dimes data reports showed 3.3 million children under five years die from birth defects yearly, 3.2 million of survivors may be disabled.[3] WHO (1972) states that the term "congenital birth defects" should be used to describe structural defects at birth.[4,5] Another definition includes structural defects, chromosomal anomalies, inborn errors of metabolism and hereditary diseases diagnosed before, at or after birth.[6,7] The incidence of congenital anomalies at birth ranges from 0.42% to 4.3% in various studies in Asia.[8-11] In least developed countries, underestimation of anomalies are increased due to unavailability of diagnostic tools, lack of medical reports, undocumented births and underreporting.[10]

This study aims to document the patterns of congenital anomalies at study site.

METHODS

This was a prospective descriptive study carried out to study the pattern of congenital anomalies in newborns born in Manipal Teaching Hospital (MTH), Pokhara, over the period of twelve months from September 2019 to August 2020. The variables studied were congenital birth defects, maternal characteristics and the immediate outcome of the babies with the congenital anomalies. All clinically identifiable congenital defects at birth, gestational age and prenatal sonographic diagnosis of anomaly were included in the study. When identified as having a birth defect, the defect was examined in detail and anomalies recorded were classified according to International Classification of Diseases- 10 criteria. The clinical examination, photographs, radiographs, ultrasound and echocardiography were done as indicated. Maternal history such as maternal age, parity, previous termination of pregnancy for malformation, previous still birth or spontaneous abortions, maternal illness, use of drugs, smoking, alcohol use and exposure to radiation were also recorded. The baby's gestational age, sex, mode of delivery, ethnicity, birth weight, birth order, occipito-frontal circumference and parental

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consanguinity were documented. Data were analyzed using descriptive analysis. Statistical Package for Social Sciences (SPSS) software, version 23 was used. Types of birth defects were documented and means and percentages were calculated. Ethical approval for this study was taken from the institutional review committee of Manipal College of Medical Sciences before the initiation of the study.

RESULTS

During the one year study period, there were a total of 2549 deliveries in MTH, out of which 34 were still born and 2515 were live births. Out of the live births, 1249 were males and 1264 were females and 2 were born with ambiguous genitalia. Congenital anomaly was detected in 24 live neonates and in one of the 34 stillbirths. This gives an incidence rate of 9.42 per 1000 live births per year. A single system was involved in 19 (79.2%) of the newborns whereas as the rest of the 5 (20.8%) had involvement of more than one system (Table1).

Incidence of congenital anomaly was slightly higher in males (0.96% vs 0.79%) among the total number of births. The mean birth weight in grams was 2503.3 ± 681.2 SD with a minimum of 1200 grams and a maximum of 3600 grams. Mean occipito-frontal circumference in cm was 32.60 ± 2.05 SD, with a minimum of 28cm and a maximum of 35cm. In reference to the immediate outcome of the neonates, 13 (54.2%) were discharged, 8 (33.3%) expired, 2 (8.3%) left on leave against medical advice, and one (4.2%) was referred on patient party's request (Table 2).

Maternal history revealed that all these mothers received folic acid during the course of their pregnancy but not prior to conception. None of the affected babies were born to a consanguineous marriage. Family history of previous congenital anomaly was seen in two (8.2%) of the mothers where they reported similar defects (cleft lip and palate and brittle bone disease respectively). (Table 3).

There was a history of previous early neonatal death in 4/24 mothers (16.7%), but the cause for the deaths could not be ascertained. Three mothers (12.5%) also reported spontaneous abortions during the first trimester in the past. There was a history of urinary tract infection in one of the mothers' during the course of her pregnancy and another mother reported of chronic hypertension and pre-eclampsia for which the pregnancy had to be terminated before term. None of the mothers who delivered a newborn with congenital anomaly gave history of exposure to offending drugs, like antipeiletics, antipsychotic drugs like lithium and antithyroid medications ,radiation, smoking or alcohol consumption during the course of the pregnancy.

Table 1. Description of the congenital anomalies and their outcomes.

Congenital Anomaly bserved	N= 24	Outcome
Ambiguous genitalia	2 (8.3%)	LAMA*
Hypospadias (coronal type)	2 (8.3%)	Discharged/ Alive
Aplastic cutis congenital	1(4.2%)	Referred out
Atrial Septal Defect with moderate pulmonary arterial hypertension	1(4.2%)	Discharged/ Alive
Brittle bone disease	1(4.2%)	Died
Cleft lip, cleft palate, choanal atresia, duodenal atresia	1(4.2%)	Referred out
Cleft lip	1(4.2%)	Discharged/ Alive
Cleft lip with complete cleft palate	1(4.2%)	Discharged/ Alive
Cleft palate	1(4.2%)	Discharged/ Alive
Club foot	1(4.2%)	Discharged/ Alive
Congenital hydrocele	1(4.2%)	Discharged/ Alive
Congenital hydronephrosis with bilateral PUJ obstruction with posterior urethral valve with mysenteric cyst	1(4.2%)	Died
Congenital rubella	1(4.2%)	Died
Down syndrome phenotype with unspecified congenital heart disease	1(4.2%)	Died
Facial defect (non-fusion of left side of lips)	1(4.2%)	Discharged/ Alive
Holoprosencephaly	1(4.2%)	LAMA*
Hypoplastic left heart syndrome	1(4.2%)	Died
Myringocele with imperforate anus	1(4.2%)	Died
Polydactyly	1(4.2%)	Discharged/ Alive
Sacrocoxygeal teratoma	1(4.2%)	Died
Single umbilical artery	1(4.2%)	Discharged/ Alive
Spina bifida occulta with sacral dimpling	1(4.2%)	Discharged/ Alive

**LAMA – Leave against medical advice*

Table 2. Characteristics of babies with congenital defects (N=24).

Characteristics	n (%)

Gender	
Males	12 (50)
Females	10 (41.7)
Ambiguous	2 (8.3)
Birth Weight (grams)	
1000 - 1500	5 (20.8)
1501 - 2500	4 (16.7)
>2500	15 (62.5)
Gestational Age (weeks)	
28-32	2 (8.3)
32-35	8 (33.3)
35-37	3 (12.5)
>37	11 (45.8)
Occipitofrontal circumference (cm)	
<33	8 (33.3)
≥33	16 (66.7)

Table 3. Maternal Characteristics (N=24).

Characteristics	n (%)
Maternal age (years)	
<20	3 (12.5)
20-30	16 (66.7)
>30	5 (20.8)
Mode of delivery	
Normal vaginal delivery	9 (37.5)
LSCS	14 (58.3)
Instrumental delivery	1 (4.2)
Parity	
Primipara	15 (62.5)
Multipara	9 (37.5)
Ethnicity	
Bhramin/Chhetri	12 (50.0)
Janajati	8 (33.3)
Dalit	4 (16.7)
Previous stillbirth	
Yes	1 (4.2)
No	23 (95.8)
Previous abortion	
Yes	3 (12.5)
No	21 (87.5)

DISCUSSION

In the present study, the incidence of congenital anomalies was almost 1%, which is lower than most other studies.[6,12-14] However, a study done in the same region as ours by Sharma et al showed a lower incidence than ours at 0.42%.[11] The low incidence of congenital anomalies in our study can be attributed to the factors like single hospital based study and only defects that were clinically apparent or needed intervention in the immediate neonatal period were noted in this study.

In this study congenital anomaly was seen slightly more in the males (50%), with a male to female ratio of 1.2:1. Two of the babies were identified as having ambiguous genitalia (8.3%). These findings are compatible with studies done by Patel et al and Taksande et al.[2,15] It was observed that congenital anomalies were more common in mothers in the age group of 20-30 years (66.6%) and least seen in those who were <20 years old (12.5%). The possible explanation for this could be that in developing countries this age group is considered to be a safe conception period hence pregnancies mostly occur in this age group. This was in contrast to a study done by Kokate et al which showed that maternal age of >30 years was a major risk factor in their study.[16] However, a similar study done by Sharma et al in the same region had similar findings as that of ours.[11]

Analysis of the overall distribution of congenital anomalies showed that the commonest system involved in our study was the genitourinary system (25.9%) followed by cardiovascular system (14.8%). These findings differ from that of other studies done in Pokhara by Sharma et al and Bastola et al.[11,17] The commonest systems involved in their settings were musculoskeletal and anomalies of the ear, eye, face and neck. The difference in the findings could be attributed to different study protocols.

In our study we noted that the distribution of incidence of anomalies among babies with normal birth weight, low birth weight (LBW) and very low birth weight (VLBW) were 62.5%, 16.79% and 20.8% respectively. Our findings of highest incidence of congenital anomalies in normal weight babies were contrary to findings from other studies where an association between LBW and congenital anomalies has been noted.[18-20]

The detection and reporting of only external/overt anomalies was possible in this study as majority of the diagnosis of anomalies was based on clinical examinations since there is lack of facilities for cytogenetic analysis or autopsy in our setting. We only included babies born in the institute so the incidence we reported may very well not reflect the true nature of incidence in the community. Furthermore, anomalies which may not be apparent at birth (such as some congenital heart disease) or needed chromosomal analysis for diagnosis were missed and therefore not recorded; It is also to be noted that anomalies that might have been detected

through antenatal scans and aborted are not reflected in this study.

CONCLUSIONS

Severe congenital anomalies must be identified by thorough clinical examinations and supplemented with necessary investigations as early diagnosis and curative interventions might give them their only chance of survival.

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