Prevalence and Clinical Presentation of Congenital Anomalies in Neonates

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Abstract: Introduction: The impact of congenital anomalies on newborn mortality is determined by a number of factors, including the prevalence of congenital anomalies, the prevalence of other opposing causes of death, the superiority and accessibility of medical and surgical care, and the presence and efficacy of primary prevention policies. The aim of the study was to assess the prevalence and evaluate the clinical presentation of congenital anomalies in neonates. Material & Methods: This prospective observational study was conducted at the Pediatric Medicine and Pediatric Surgery department of Dhaka Shishu Hospital, Dhaka, Bangladesh during the period from April 2012 to September 2012. Eighty (80) newborns were included in the study using the purposive sampling method. A pre-designed questionnaire was completed for every neonate including H/O regular maternal antenatal care with the taking of TT and MMR vaccine, any maternal disease or fever with rash, taking any offending drug, use of abortifacient, exposure to radiation or industrial hazards, feeding habit including smoking or use of alcohol and clinical and anthropological examination. After collecting, the data were processed and analyzed using computer-aided statistical software SPSS (Statistical Package for Social Sciences) version 16.0 for Windows (SPSS Inc., Chicago, Illinois, USA). Ethical clearance was taken from the Ethical Review Committee (ERC) of BICH, Dhaka Shishu Hospital, Dhaka, Bangladesh. Results: Prevalence of congenital anomalies were found at 7.2%. The mean age was 4.04 ± 3.43 days with a range from 1 to 16 days. Male infants were 52 (65.0%) and female infants were 28 (35.0%). The newborn mean weight was 2437.5 ± 324.4 gm, and the mean length was 51.89 ± 2.71 cm. Mean OFC was 35.19 ± 1.21 cm (normal OFC just after birth 35 cm and normal increment in 1st month 2 cm). The mean gestational age was 35.59 ± 2.33 weeks. The distribution of the studied patients according to their immediate outcome shows, that almost two-thirds (66.3%) of patients had been discharged with advice, DORB was 11 (13.7%) and expired 16 (20.0%). Conclusion: The hospital prevalence of birth defects is 7.2% in newborns. Birth defects are more frequent in mothers with diabetes mellitus, hypertension, consanguineous marriage, and those with irregular antenatal care.

Keywords: Congenital Anomalies, Newborn, Clinical Presentation, Prevalence

1. Introduction

The impact of congenital anomalies on newborn mortality is determined by a number of factors, including the prevalence of congenital anomalies, the prevalence of other opposing causes of death, the superiority and accessibility of medical and surgical care, and the presence and efficacy of primary prevention policies. In some developing countries, newborn mortality remains extremely high (as high as 10% in 1995). Each year, eight million children are born worldwide with congenital malformations, of which 3.3 million die before the age of five; 3.2 million of the survivors may be mentally and/or physically disabled. [1] The rate of congenital anomalies in twins was 405.8 per 10 000 twins

versus 238.2 per 10 000 singletons [rate ratios (RR) = 1.7, 95% confidence interval (CI) 1.5–2.0]. Newborn mortality attributable to congenital anomalies decreased by 33.4 percent and accounted for an increasing segment of the total newborn mortality. Birth defects comprise a complex and heterogeneous group of embryonic and/or fetal development disorders, which, in about 50% of cases, have no known cause, although genetic and environmental or a combination of these two may be involved. [2] However, the underlying causes of the majority of congenital anomalies remain unknown, and multifactorial inheritance is thought to be the underlying etiology of the majority of common congenital anomalies. It has been estimated that about fifteen to twenty-five percent of congenital anomalies are due to recognized genetic conditions, eight to twelve percent to environmental factors, and 20%-25% to multifactorial inheritance. The majority of congenital anomalies, 40%-60% are unexplained. [3] At the first valuation by the clinician, details from a checklist of symptoms were requested, counting the presence of feeding difficulties, constipation, physical dormancy, prolonged jaundice, cold or mottled skin, macroglossia, hypothermia, umbilical hernia, and other abnormalities. Newborns with one or more of these symptoms or signs were considered to be 'symptomatic'. Although different studies have been undertaken in different parts of the world, no such study has been undertaken in Bangladesh.

2. Methodology

This prospective observational study was conducted at the Pediatric Medicine and Pediatric Surgery departments of Dhaka Shishu Hospital, Dhaka, Bangladesh. Eighty (80) newborns were included in the study using a purposive sampling method. The study was conducted during the time from April 2012 to September 2012. The aim of the study was to assess the prevalence and evaluate the clinical presentation of congenital anomalies in neonates. Both the major and minor congenital malformations were taken into account by a questionnaire. Immediately after admission, a detailed history of the newborn baby and mother was taken including all familial and gestational factors, and a meticulous examination of the baby was done. Thereafter, the newborn remained under continuous observation along with regular follow-up during a hospital stay. A pre-designed questionnaire was completed for every neonate including H/O regular maternal antenatal care with the taking of TT and MMR vaccine, any maternal disease or fever with rash, taking any offending drug, use of abortifacient, exposure to radiation or industrial hazards, feeding habit including smoking or use of alcohol and clinical and anthropological examination. After collecting, the data were processed and analyzed using computer-aided statistical software SPSS (Statistical Package for Social Sciences) version 16.0 for Windows (SPSS Inc., Chicago, Illinois, USA). Ethical clearance was taken from the Ethical Review Committee (ERC) of BICH, Dhaka Shishu Hospital, Dhaka, Bangladesh.

3. Results

A total number of 1017 newborns were admitted to the Pediatric Medicine and Pediatric Surgery departments of Dhaka Shishu Hospital, Dhaka from April 2012 to September 2012 (n=1017). Out of which, 80 neonates were presented with congenital anomalies and a prevalence of 7.2% was found. Table 1 shows the age distribution of the studied patients. Three fourth (75.0%) of the total patient's ages belonged to 1-5 days, and their mean age was 4.04±3.43 days with a range from 1 to 16 days. Figure 1 suggests the sex distribution of the studied patients. Male infants were 52 (65.0%) and female infants were 28 (35.0%). Table 2 shows that the newborn mean weight was 2437.5±324.4 gm, and the mean length was 51.89±2.71 cm. Mean OFC was 35.19±1.21 cm (normal OFC just after birth 35 cm and normal increment in 1st month 2 cm). The mean gestational age was 35.59±2.33 weeks. The distribution of the studied patients according to their immediate outcome. Table 3 shows the distribution of the study patients according to maternal pregnancy, labor & delivery of the study patients. Primipara was found in 47 (58.7%), regular antenatal checkups 13 (16.4%), mean antenatal checkups 2.05±1.57, normal delivery 46 (57.5%) among them breech presentations were 7 (8.7%). Birth injuries were 5 (6.3%). Table 4 shows, that almost two-thirds (66.3%) of patients had been discharged with advice, DORB was 11 (13.7%) and expired 16 (20.0%).

Table 1. Age distribution of the study patients (n=80).

Age (in days)	n (%)
1 – 5	60 (75%)
6 – 10	17 (21.2%)
>10	3 (3.8%)
Mean±SD	4.04 ±3.43
Range (min-max)	(1-16)

Table 2. Distribution of the study patients according to their anthropometric measurements (n=80).

Clinical presentation	Mean± SD	(Min-Max)
Weight on admission (gm)	2437.5±324.4	(1800-3000)
Length (cm)	51.89±2.71	(49-57)
OFC (cm)	35.19±1.21	(30-39)
Gestational age (weeks)	35.59±2.33	(32-39)

Table 3. Distribution of the study patients according to maternal pregnancy, labour & delivery (n=80).

Pregnancy, labour & delivery	n (%)
Parity	
Primi	47 (58.7%)
Multi	33 (41.3%)
Antenatal checkup	
Regular (4 or Above)	13 (16.4%)
Irregular (<3)	52 (64.4%)
None (0)	15 (19.2%)
Mean±SD	2.05±1.57
Range (min-max)	(0-5)
Mode of Delivery	
Normal	46 (57.5%)
Vertex presentation	39 (48.8%)
Breech presentation	7 (8.7%)
LUCS	34 (42.5%)

Pregnancy, labour & delivery	n (%)
Birth injury	
Present	5 (6.3%)
Absent	75 (93.7%)

Table 4. *Distribution of the study patients according to their immediate outcome (n=80).*

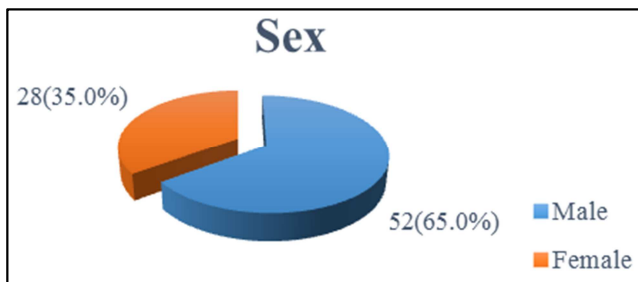| Immediate outcome	n (%)
DORB	11 (13.7%)
Discharge with advice	53 (66.3%)
Expired	16 (20%)

Figure 1. *Pie chart showing the sex distribution of the study patients.*

4. Discussion

This prospective observational study was carried out with an aim to find out the prevalence and clinical presentation of congenital anomalies, to identify different types of abnormalities of newborns having congenital anomalies, to find out the maternal risk factors, family history, and consanguinity of newborns with congenital anomalies as well as to find out the mortality and immediate hospital outcome of these newborns admitted in the Hospital. In this present study, it was observed that three fourth (75.0%) of the studied patients' age belonged to 1-5 days and their mean age was 4.04±3.43 days with a range from 1 to 16 days. Gillani et al.; Fatema et al.; Singh and Gupta et al observed the congenital anomalies in newborns of similar age. [4-6] Several other studies determined the pattern of major congenital malformations in neonates admitted to NICU and evaluated their early outcomes. [7-9] It was observed in this current series that congenital anomalies were predominant in male patients, where the male to female ratio was almost 2:1. Singh and Gupta mentioned in their study that the number of congenital anomalies was more in males, where the male to female ratio was 1.6:1.4. [6] Similar findings were also obtained by studies by various other researchers, which closely resembled the current study. [4, 5, 7, 9-11] In this current study, it was observed that the mean weight of the newborn was 2437.5±324.4 gm, and the weight varied from 1800 – 3000 gm. Patel et al. showed mean birth weight was 2879.3±655.9 gm. [12] Tootoonchi showed 30.9% and 27.3% cases were low birth weight (LBW) and premature, respectively. [7] Swain, Agrawal, Bhatia, et al. and Fatema et al. found 29.02% and 53.33% of a newborn have congenital anomalies with low birth weight respectively. [5, 13] Fida et al found body weight (kg) varied from 1011 gm to 5200 gm. In this present study, it was observed that the mean length was 51.89±2.71 cm with a

range from 49 – 57 cm. [10] Their study showed mean length was 50.30±5.36 cm with a range from 31.0 to 59.0 cm, which was similar to the current study. [10] In our study, it was observed that the mean OFC was 35.19±1.21 cm (normal OFC just after birth 35 cm and normal increment in 1st month 2 cm) with a range from 30 – 39 cm and also mean gestational age was 35.59±2.33 weeks varied from 32 – 39 weeks. Patel obtained the mean gestational age was 37.1±3.5 weeks. [12] In another study, Tootoonchi found the mean gestational age of 38.20±2.60 weeks varied from 23 to 44 weeks. [7] Regarding the immediate outcome, it was observed that almost two-thirds (66.3%) of patients were discharged with advice, DORB 13.7%, and expired 20.0%. Gillani et al. reported in their study that most of the admitted patients (40.0%) were discharged after necessary investigations and counseling, 25.0% expired, and 20% were referred to other hospitals. [4]

Limitations of the study

The present study was conducted in a very short period due to time constrain and funding limitations.

5. Conclusion and Recommendations

The hospital prevalence of birth defects in newborns is 7.2 percent. Birth defects are more common in mothers who have diabetes, hypertension, consanguineous marriage, or irregular antenatal care. Cleft lip and/or palate was the most common birth defect, followed by congenital heart diseases, hydrocephalus, myelomeningocele, ambiguous genitalia, and anorectal malformations. Long-term follow-up programs for newborns with an obvious life-threatening event subsidize the adaptation of medical attitudes to the child's needs and the confirmation of the medical diagnosis. A thorough diagnostic evaluation, combined with a comprehensive treatment plan, improves survival and quality of life for the most precocious newborns. To reach a meaningful conclusion, it is recommended that all neonates be thoroughly examined for overt and occult congenital anomalies. Furthermore, it is required.

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