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Original Research Article

Different Types of Abnormalities of Newborn with Congenital Anomalies

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Abstract

Introduction: Congenital anomalies are structural, behavioral, functional, or metabolic defects that occur before the birth of a baby, and their nature and type are highly dependent on the causative agent. It refers to changes in embryonic or fetal development caused by genetic, environmental, or unknown factors that result in physical and/or mental impairment. This study aims to evaluate the various types of abnormalities in newborns with congenital anomalies. ***Methods:*** 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 evaluate the various types of abnormalities in newborns with congenital anomalies. 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. ***Results:*** Eighty neonates were presented with congenital anomalies. Male infants were 52(65.0%) and female infants were 28(35.0%). Among congenital heart diseases TGA found in 4(5.0%), TOF 2(2.5%), VSD 4(5.0%), PDA 5(6.3) cases. According to the patient's abnormalities, hydrocephalus was found in 14(17.5%), encephalocele in 4(5.0%) & meningocele, myelomeningocele was found in 13(16.3%) cases. Club foot in 8(10.0%) syndactyly, polydactyly was in 6(7.5%) and cleft lip and palate were in 16(20.0%) cases. Anorectal malformations were observed in 11(13.8%) cases. Ambiguous genitalia were 11(13.8%) cases. Previous maternal history of abnormal delivery shows Encephalocele, meningocele, myelomeningocele, club foot was 9(11.4%), IUGR 3(3.8%), preterm birth 7(8.8%), stillbirth 3(3.8%) and infants death was 3(3.8%). ***Conclusion:*** Most frequent birth defect was cleft lip and/or palate followed by congenital heart diseases, hydrocephalus, myelomeningocele, ambiguous genitalia, and anorectal malformations. The study of birth defects in underdeveloped countries should continue.

Keywords: Abnormalities, Congenital Anomalies, Neonates, Parental Consanguinity, Malformations.

INTRODUCTION

Congenital anomalies are structural, behavioral, functional, or metabolic defects that occur before the birth of a baby, and their nature and type are highly dependent on the causative agent. It refers to changes in embryonic or fetal development caused by genetic, environmental, or unknown factors that result in physical and/or mental impairment. It has been estimated that about 15 to 25% of congenital anomalies are due to recognized genetic conditions, 8 to 12% to environmental factors, and 20 to 25% to multifactorial inheritance. The majority of congenital anomalies, 40 to 60% are unexplained [1]. Another consequence of birth defects is the high death rate within the first year of life. Infant mortality is an important indicator of a country's or community's health, and it is linked to factors such as maternal health, the quality and accessibility of health care, socioeconomic conditions, and public health practices [2]. Except for chromosomal abnormalities, twin pregnancies have a higher rate of congenital anomalies than singleton pregnancies for all major types of anomalies. Birth defects are the leading cause of infant mortality in developed countries. Most of the

time, they limit the newborns' ability to adapt to life outside the womb and may significantly reduce their quality of life. Cleft lip/cleft palate, neural tube defects (occurring singly or in combination with other abnormalities), limb abnormalities (often in combination with neural tube defects of various types), omphalocele, umbilical hernia, anorectal malformations, and dysmorphism associated with multiple congenital abnormalities were the most common abnormalities seen in these babies. The damages and handicaps that result from congenital abnormalities usually pose serious problems to the health, the social and financial well-being of the children, their family, and the healthcare system. A similar trend is being observed in the developing world [3]. Among the risk factors are advanced maternal and paternal ages, parental consanguinity, teratogenic agents such as infectious agents and drugs, and nutritional deficiencies [4-6]. Congenital anomalies make an important contribution to infant mortality and remain a leading cause of death in many countries of the world. Many babies also died in Bangladesh due to congenital anomalies. Information on birth defects is becoming increasingly more important throughout the world so that preventive measures can be taken.

OBJECTIVE

- To evaluate the various types of abnormalities in newborns with congenital anomalies

METHODOLOGY AND MATERIALS

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 the purposive sampling method. The study was conducted during the time from April 2012 to September 2012. Both the major and minor congenital malformations were taken into account by a questionnaire. Informed consent was obtained from the legal guardians of the infants. 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 the 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.

RESULTS

Table 1 shows the age distribution of the study patients. Three fourth (75.0%) of the study patients were between 1-and 5 days of age, and their mean age was 4.04±3.43 days with ages ranging from 1 to 16 days. Figure 1 demonstrated the sex distribution of the study patients. Male infants were 52(65.0%) and female infants were 28(35.0%). Table 2 showed the distribution of the study patients according to their abnormalities. Among congenital heart diseases. TGA found 4(5.0%), TOF 2(2.5%), VSD 4(5.0%), PDA 5(6.3) cases. Hydrocephalus 14(17.5%), encephalocele 4(5.0%) & meningocele, myelomeningocele was 13(16.3%) cases. Club foot 8(10.0%) syndactyly, polydactyly was 6(7.5%) and cleft lip and palate were 16(20.0%) cases. Anorectal malformations were 11(13.8%) cases. Ambiguous genitalia were 11(13.8%) cases. Table 3 shows the distribution of the study patients according to previous maternal history of abnormal delivery. Encephalocele, meningocele, myelomeningocele, and club foot were 9(11.4%), IUGR 3(3.8%), preterm birth 7(8.8%), stillbirth 3(3.8%), and infant's death was 3(3.8%). Table 4 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 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)

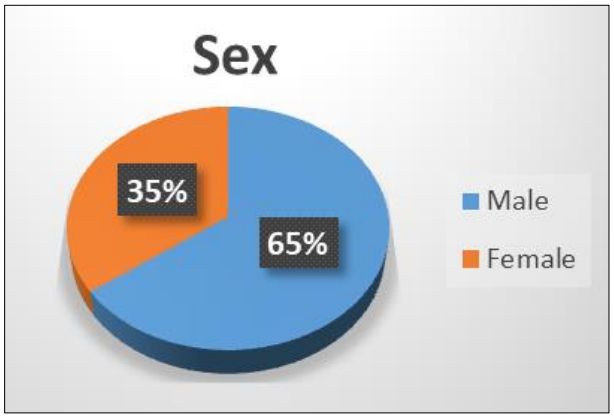

Figure 1: Pie chart showing the sex distribution of the study patients

Table 2: Distribution of the study patients according to their abnormalities (n=80)

Abnormalities	N (%)
Congenital heart diseases	
TGA	4 (5%)
TOF	2 (2.5%)
VSD	4 (5%)
PDA	5 (6.3%)
Hydrocephalus	14 (17.5%)
Neural tube defects	
Encephalocele	4 (5%)
Meningocele, Myelomeningocele	13 (16.3%)
Musculoskeletal defects	
Club foot	8 (10%)
Syndactyly, polydactyly	6 (7.5%)
Cleft lip and palate	16 (20%)
Spina bifida	5 (6.3%)
Gastrointestinal defects	
Umbilical hernia	3 (3.8%)
Gastroschisis	2 (2.5%)
Anorectal malformation	11 (13.8%)
Urogenital defects	
Polycystic kidney disease	3 (3.8%)
Ambiguous genitalia	11 (13.8%)
Skin defects	5 (6.3%)
Facial defects	3 (3.8%)
Others	2 (2.5%)

Note: Multiple abnormalities were present in one patient.

Table 3: Distribution of the study patients according to previous maternal history of abnormal delivery (n=80)

Previous history	N (%)
Specific birth defects	
Encephalocele	3 (3.8%)
Meningocele/Myelomeningocele	3 (3.8%)
Club foot	3 (3.8%)
No defects	71 (88.6%)
IUGR	
Present	3 (3.8%)
Absent	77 (96.2%)
Preterm births	
Present	7 (8.8%)
Absent	73 (91.2%)
Stillbirths	
Present	3 (3.8%)
Absent	77 (96.2%)
Infant deaths	
Present	3 (3.8%)
Absent	77 (96.2%)

Table 4: 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

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

DISCUSSION

Congenital abnormalities are defined as obvious structural or form abnormalities that are either present at birth or become apparent within a few days of birth. Limb defects were the most common external anomalies, followed by craniofacial anomalies, which accounted for more than 68 percent of all cases. In this present study, it was observed that three fourth (75.0%) of the study 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. Researchers previously observed congenital anomalies in newborns of a similar age range [8-10]. Few other studies have determined the pattern of major congenital malformations in neonates admitted to NICU and evaluated their early outcomes [7, 11, 12]. 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 *et al*, 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 [10]. Similar findings were also observed in previous studies [7, 9, 11, 13, 14]. Regarding the abnormalities, it was observed in this present series that multiple abnormalities were present in one patient. It was also found that congenital heart diseases such as transposition of the great arteries (TGA) were 5.0%, tetralogy of Fallot (TOF) at 2.5%, *ventricular septal defect (*VSD) at 5.0%, and *patent ductus arteriosus (*PDA) 6.3% cases. Okoromah *et al*, mentioned in their study that ventricular septal defect (VSD) was the leading cardiac lesion among all cases of CHD (35.6%), patent ductus arteriosus (PDA) 13.7%, and accounted for 54.2% of cases in the acyanotic group tetralogy of Fallot (TOF) was the most frequent cyanotic lesion among all cases of CHD 15.1%, transposition of the great arteries (TGA) + VSD without PS 4.1% and the cyanotic group 44.0% [15]. Regarding the previous maternal history of abnormal delivery, it was observed in this present series that specific birth defects were in 11.4%, out of which encephalocele was 3.8%, meningocele/ myelomeningocele 3.8%, club foot 3.8%. IUGR was present in 3.8%, preterm birth in 8.8%, stillbirth in 3.8%, and infant death was 3(3.8%) in this study. Jehangir *et al*, showed term 16.66%, pre-term 83.34%, and stillbirths 2.95% [16]. In this series, it was observed that multigravida was found in 41.3%, regular antenatal checkups received only 16.4%, 42.5% underwent LUCS, and birth injury was observed in 6.3% cases. Two other studies also observed multigravida 88.89% and 96.6% respectively [16, 17]. Fatema *et al*, showed only 8.0% made their antenatal visit regularly [9]. Almost similar findings were obtained by Singh and Gupta [10].

LIMITATIONS OF THE STUDY

The present study was conducted in a very short period due to time constrain and funding limitations. The small sample size was also a limitation of the present study. No control was taken.

CONCLUSION AND RECOMMENDATIONS

Congenital anomalies are a major cause of miscarriages and infant mortality. The most frequent birth defect was cleft lip and/or palate followed by congenital heart diseases, hydrocephalus, myelomeningocele, ambiguous genitalia, and anorectal malformations. The study of birth defects in underdeveloped countries should continue. The identification of occurrence, risk factors, and significances are important for planning defensive measures and effective treatments. To control the factors underlying the various types of congenital abnormality encountered in this area more research is needed. Developed maternal health, pre-conception care including folic acid supplementation, and early diagnosis of most of these anomalies are recommended.

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