



# Prevalence and Pattern of Birth Defects in a Tertiary Care Hospital in Bangladesh

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## Key words:

Birth Defects, Newborn

## Abstract:

**Background:** Birth Defect is one of the most important causes of neonatal mortality and morbidity worldwide. The pattern and prevalence of Birth Defects may vary overtime or with geographic location. The objective of the study to determine the prevalence and pattern of Birth Defects in live newborn admitted in Hospital.

**Materials & Methods:** This cross-sectional descriptive study was carried in Neonatology and OBS-gynae department of Sir Salimullah Medical college Hospital during the period of July 2016 to December 2019. A total 306 babies with Birth Defects were examined and detailed history for any risk factor was taken. A through physical examination was performed. Confirmation of internal defect was done by various imaging modalities; i.e, radiography, ultrasound, echocardiography and CT scan. A detailed history was taken regarding maternal age, gestational age and previous delivery of abnormal baby.

**Results:** During this period total 27799 was born alive, of which 306 had Birth Defects, the prevalence of Birth Defects was 1.1%. The predominant system involved was Musculo-skeletal system (30%) followed by CNS (19%), oro-facial clefts (13%), GIT, CVS, Ear-nose-throat(7%) and least involved system was urinary system(4%). Talipes was most common one in musculo-skeletal system defect and Anencephaly in CNS defect.

**Conclusion:** Prevalence of Birth Defects is common among newborn admitted in hospital and musculoskeletal system is most commonly affected system. Birth Defects is upcoming issue in current newborn situation which need to be prioritized.

## Introduction:

Birth defects, also known as congenital anomalies, are structural or functional anomalies, including metabolic disorders, which are present at the time of birth<sup>1</sup>. Birth defects can be isolated abnormalities or part of a syndrome and continue to be an important cause of neonatal and infant morbidity

and mortality<sup>2</sup>. With significant improvement of child health care overall childhood mortality has been decreased but the impact of birth defect is increasing day by day specially in developing countries<sup>2</sup>. In developed countries birth defects cause 5-7% of perinatal, neonatal and childhood mortality but in developing countries, they

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contribute to about 30-35% of mortality and this proportion is progressively increasing. Every year more than 8.14 million children are born with serious birth defect due to genetic or environmental causes. Hundreds of thousands are born with serious birth defects of prenatal origin, including maternal exposure to environmental agents (teratogens) such as alcohol, rubella, syphilis and iodine deficiency that can cause harm a developing fetus<sup>3</sup>. Around 40% to 60% of congenital anomalies are of unknown etiology; 20% are attributed to a combination of heredity and other factors; 7.5% due to single gene mutations; 6% is caused by chromosomal abnormalities; and another 5% is due to maternal illnesses, such as diabetes or infection, or use of anticonvulsant or other drugs<sup>4</sup>. Serious birth defects can be lethal. For those who survive, these disorder can cause lifelong mental, physical, auditory or visual disability<sup>3</sup>. The pattern of congenital anomalies varies from region to region and also over time<sup>4</sup>. About 60% of the causes of congenital anomalies in humans are still unknown. However, in about 25% of congenital anomalies, the causes seem to be “multifactorial”, indicating a complex interaction between genetic and environmental risk factors. A wide range of environmental risk factors have been associated with the occurrence of congenital anomalies<sup>1</sup>. The occurrence of congenital anomalies has also been associated with advanced maternal age, parental consanguinity, increasing birth order and low birth weight<sup>5</sup>. According to the literatures, birth defects are a major source of infant and child morbidity and mortality and single or multiple defects can occur in one or several organs of the children<sup>6</sup>. Some of the congenital anomalies may be life threatening, may impair function, hence an immediate management is required. The long-term disability caused by congenital anomalies may have a significant impact not only on a child’s wellbeing and development, but also on families, health care systems and societies<sup>3</sup>.

With significant improvement of child health care overall childhood mortality has been decreased but the impact of birth defect is increasing day by day specially in developing countries. Birth defects are one of the four leading causes of neonatal mortality in Bangladesh which is about 12% of all neonatal deaths<sup>7</sup>. Birth defect are an important cause of still births and neonatal deaths in Bangladesh and contribute to chronic illnesses and disability among children. The burdens of birth defects are felt at a greater magnitude due to the increased presence

of risk factors associated with birth defects compared to other countries. In addition, the problem of birth defects is overshadowed by other causes of child mortality and morbidity such as low birth weight, asphyxia, maternal and childhood infections, under-nutrition and other factors for which studies and actions regarding congenital anomalies are largely lacking. There has been a significant decline in infant and childhood mortality rates in most countries in the past two decades. This has primarily been due to extensive and successful use of immunization, control of diarrhoeal disorders, acute respiratory tract infections and improvement in health-care services through a focus on primary health care<sup>3</sup>. In Bangladesh, due to the low socio-economic status of most households, adverse environmental conditions, widespread existence of maternal infections and lack of awareness about birth defects, congenital anomalies have a higher rate of occurrence compared to developed countries. Many of the congenital anomalies can be prevented and treated. Important aspects of prevention and treatment include early recognition, micronutrient supplementation, vaccination, antenatal care and surgical intervention or reconstruction. For example simple preventive measures like the intake of folic acid during pregnancy can drastically reduce the incidence of neural tube defects. However, despite having the expertise and facilities to tackle the problem to some extent, the burden of congenital anomalies among children in Bangladesh poses a significant health problem largely due to insufficient studies and data especially on the variations or spectrum of the anomalies, inadequate prevention and intervention strategies and lack of awareness among people. Therefore this research, exploring the prevalence and various patterns of distribution of congenital anomalies, gives valuable epidemiologic information based on which further analytic studies can be undertaken and appropriate prevention and intervention strategies can be designed.

#### **Methodology:**

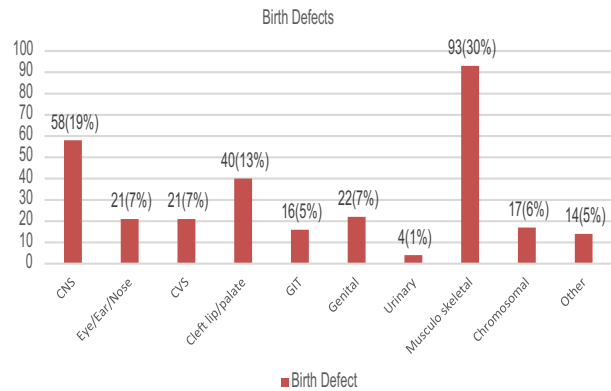
This was a prospective hospital based cross-sectional study with consecutive sampling of newborn babies with birth defect admitted in NICU of Sir Salimullah medical college and Mitford hospital from July 2016 to December 2019 . Total 306 newborn with birth defect were enrolled in the study. After inclusion, detailed relevant history was taken from the mother as well as from antenatal record which included maternal age,

consanguinity, gestational age, previous spontaneous abortion, birth defect in previous live birth, maternal history of diabetes mellitus, hypertension, hypothyroidism, infection with TORCH, history of received any anti convulsant, exposure to radiation and smoking during antenatal period and history of receiving peri conceptional folic acid during first trimester. All birth defect babies were categorized at birth as having major or minor anomalies, single or multiple anomalies by inspection by a neonatologist and classify according to ICD- 10 (International classification of Disease-10). The International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10) has described birth defects or congenital malformation in chapter XVII (Q00-Q99) which is titled as 'Congenital malformations, deformations and chromosomal abnormalities' and excludes: inborn errors of metabolism (E70-E90). It has classified birth defects into several body systems along with chromosomal abnormalities and for each category there is a specific code. To confirm the birth defect necessary radiological, ultrasonography, hematological and when possible genetic investigation were done. Clinical factors like associated complication in terms of co-morbidities during hospital stay, management option and outcome (discharge/.referral/death) were documented. The babies with birth defect who need consultation from other department was taken and who requiring surgery were followed up in post operative ward. All other management was continued as per institutional guideline.

### Result:

This cross sectional descriptive study was carried out in the department of Neonatology of Sir Salimullah Medical college from July 2016 to December 2019. During the study period total 28220 babies was born of which 27799 was live born. A total 306 neonates had one or more congenital anomalies accounting a prevalence of 1.1% (Table I). The demographic characteristics of study subject were describe in Table II. The congenital anomalies were more in male (M:F=2:1) and term inborn babies ranging birth weight between 2500 gm to 4000gm.

The most prevalent birth defect were musculoskeletal system defect (30%) followed by central nervous system defect (19%), cleft lip/palate (13%) while the least were urinary system (1%) (Figure 1).



**Fig.-1:** Distribution of Birth Defects by system affected as per ICD-10

**Table II**  
Distribution of Neonatal variables in Newborn with Birth Defect

Variable	No	Frequency
<b>Sex</b>		
Male	181	59%
Female	115	38%
Ambiguous	10	3%
<b>Birth weight</b>		
1000- <1500	36	11.8%
1500- <2500	110	35.9%
2500- 4000	160	52.3%
<b>Gestation</b>		
Term	185	60%
Preterm	116	38%
Post term	1	0.3%
Preivable	4	1.3%
<b>Place of Delivery</b>		
Inborn	204	66.6%
Outborn	102	33.3%

**Table I**  
Prevalence of Birth Defects among Live born Newborn

Total birth July 2014 to December 2019	Total live birth	Total still birth	Total birth defect	Prevalence of Birth defects
28220	27799	421	306	1.1%

In musculo-skeletal system, the most common defect identified were Talipes equinovarus ( 26) (Table-III). Regarding central nervous system defect, Anencephaly (17) were more prevalent followed by Lumber spina bifida without

hydrocephalus (14) (Table IV). The common visible major defects were Neural tube defects (19%) followed by Oro-facial clefts, Talipes, abdominal wall defects (Table V).

**Table III**

*Summary of most common system, Musculo-skeletal system of Birth Defect cases according to ICD-10 code*

Birth Defect Type	Ambiguous	Female	Male	Total
Q65.8 Other congenital deformities of hip	0	0	1	1
Q66.0 Talipes equinovarus	0	12	14	26
Q66.4 Talipes calcaneovalgus	0	1	1	2
Q66.5 Congenital pes planus	0	0	1	1
Q66.6 Other congenital valgus deformities of feet	0	0	1	1
Q66.8 Other congenital deformities of feet	0	1	1	2
Q66.80 Rocker bottom foot	0	0	1	1
Q68.8 Other specified congenital musculoskeletal deformities	0	0	1	1
Q69.0 Accessory finger(s)	0	2	2	4
Q69.02 Accessory finger (s) –Postaxial	0	2	0	2
Q69.2 Accessory toe(s)	0	1	0	1
Q69.9 Polydactyly, unspecified	0	2	1	3
Q70.2 Fused toes	0	0	1	1
Q71.2 Congenital absence of both forearm and hand	0	1	0	1
Q71.3 Congenital absence of hand and finger(s)	0	0	1	1
Q71.30 Congenital absence of finger(s)	0	1	2	3
Q71.31 Absence or hypoplasia of thumb	0	0	1	1
Q71.4 Longitudinal reduction defect of radius	0	0	2	2
Q71.6 Lobster-claw hand	0	2	0	2
Q72.1 Congenital absence of thigh and lower leg with foot present	0	0	1	1
Q72.30 Congenital absence or hypoplasia of toe(s) with remainder of	0	1	0	1
Q72.4 Longitudinal reduction defect of femur	0	1	0	1
Q72.5 Longitudinal reduction defect of tibia	0	1	0	1
Q72.6 Longitudinal reduction defect of fibula	0	1	0	1
Q72.8 Other reduction defects of lower limb(s)	0	1	0	1
Q74.0 Other congenital malformations of upper limb(s), including	0	0	1	1
Q74.3 Arthrogyrosis multiplex congenita	0	1	3	4
Q76.3 Congenital scoliosis due to congenital bony malformation	0	2	1	3
Q76.30 Single hemivertebra with congenital scoliosis	0	0	1	1
Q76.4 Other congenital malformations of spine, not associated with	0	1	0	1
Q77.1 Thanatophoric short stature	0	0	1	1
Q77.4 Achondroplasia	0	1	1	2
Q78.9 Osteochondrodysplasia, unspecified	0	1	0	1
Q79.0 Congenital diaphragmatic hernia	0	0	1	1
Q79.2 Exomphalos	2	3	4	9
Q79.3 Gastroschisis	1	3	1	5
Q79.4 Prune belly syndrome	0	0	1	1
Q79.80 Congenital constriction bands	0	1	0	1
<b>Total</b>	<b>3</b>	<b>43</b>	<b>47</b>	<b>93</b>

**Table IV**  
Summary of 2<sup>nd</sup> common system, CNS Defects according to ICD-code 10

Birth Defect Type	Ambiguous	Female	Male	Total
Q00.00 Anencephaly	0	10	7	17
Q00.1 Craniorachischisis	0	1	0	1
Q01.0 Frontal encephalocele	0	1	0	1
Q01.2 Occipital encephalocele	0	1	3	4
Q02 Microcephaly	0	0	1	2
Q03 Congenital hydrocephalus	0	0	6	6
Q03.0 Malformations of aqueduct of Sylvius	0	0	1	1
Q03.8 Other congenital hydrocephalus	0	1	0	1
Q03.9 Congenital hydrocephalus, unspecified	0	0	1	1
Q04.00 Agenesis of corpus callosum	0	1	0	1
Q04.35 Hydranencephaly	0	1	0	1
Q04.61 Single congenital cerebral cyst	0	0	1	1
Q05.2 Lumbar spina bifida with hydrocephalus	0	2	1	3
Q05.5 Cervical spina bifida without hydrocephalus	0	0	1	1
Q05.6 Thoracic spina bifida without hydrocephalus	0	0	2	2
Q05.7 Lumbar spina bifida without hydrocephalus	0	4	10	14
Q05.9 Spina bifida, unspecified	0	1	0	1
Q06.1 Hypoplasia and dysplasia of spinal cord	0	1	0	1
Total	0	24	34	58

### Discussion:

Congenital anomalies are among the major causes of childhood morbidity and mortality in many countries. This study was attempt to find out the congenital anomalies among the newborn admitted in NICU and post natal ward of Gynae-Obstetric department of Sir Salimullah Medical college. The prevalence of Birth defects in our study was 1.1% which were similar with that of Singh et al. study from India<sup>8</sup> who reported 1.5% and Gopalipour et al. from Iran who reported 1.01%<sup>9</sup> and Desai et al. from Bombay<sup>10</sup> and Tahsinul et al. from Bangladesh.<sup>11</sup> The low prevalence in our study was due to exclusion of stillborn and aborted cases which were included in other studies. The pattern of birth defect may vary over time or with geographical location reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variable<sup>12</sup>.

In this study, the most prevalent birth defect were seen in musculoskeletal system (30%) followed by

central nervous system(19%), oro-facial clefts(13%), genital, ear/eye/nose and cardiovascular system (7%) and least involved system were urinary system (4%) defects.. In musculoskeletal system, most common defect was Talipes (9.4%). These finding was consistant with Sankar S et al. study done in India who found musculoskeletal system was most common system involved (33.2%) of which Talipes was more common.<sup>13</sup> Similar finding was observed in other studies done by Sing et al. , Dasie et al. in India and Masuda et al. in Iran studies (30.6% and (30.1%) respectively.<sup>8,9,10</sup> Another study done by Amin T et al. from Bangladesh, found that Talipes was the most frequent birth defect in his study population<sup>11</sup> Some studies however recorded higher incidence of CNS malformation followed by musculoskeletal system. Hossain et al. found that CNS malformation were (20.35%) more common followed by musculo-skeletal system(18.58%)<sup>15</sup> and Fatema et al. from Bangladesh observed that CNS was the most commonest defect of which hydrocephalus was

more common<sup>16</sup>. Other studies done by Amin T et al. and Afroze s et al. found that cardiovascular system defect was most common defect followed by musculoskelstal system defect<sup>11,17</sup>.

Birth defects were more common in male (male 59%, female-38%, M:F- 2:1) in this study .Similar male preponderance were observed in Amin T et al<sup>11</sup>, Banu et al.<sup>14</sup>, Fatema et al.<sup>16</sup> and Afroze s et al.<sup>17</sup> studies from Bangladesh and Hossain et al. and Sanker et al. studies from India<sup>13,15</sup>. It may be because of the fact that the females were afflicted with more lethal congenital malformations and could not survive to be born with signs of life.

This present study showed that Birth Defect was more prevalent in term babies (60%) and babies with birth weight 2500-4000 gm had birth defects more (52.3%) and this finding was consistant with other studies<sup>19,20,21</sup>.

### Conclusion:

This study has highlighted the prevalence and types of Birth defects in our hospital. Musculoskeletal system defects was the most common and Talipes was the most frequent congenital anomalies. Improvement in obstretic care, prenatal diagnosis and early neonatal care is advocated as these can improve the survival in this group. As this is a single centered study, large scale multicetered study should be needed to find out the actual prevalence of Birth Defects in Bangladesh.

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