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Research Article

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# Prevalence and Analysis of Inborn Birth Defects in A Tertiary Level Hospital in Bangladesh

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## Abstract

***Objective:*** *To assess the prevalence of congenital defects and to investigate the maternal and perinatal aspects in relation to the detailed ICD-10 coding of each individual case using The New Born Data base NBBD data collection system under Global surveillance in collaboration with Center for Disease Control CDC, Atlanta and All India Institute of Medical Science AIMS, New Delhi and Bangabandhu Sheikh Mujib Medical University BSMMU as the Focal point of investigation.*

***Methods:*** *All births and terminations of pregnancy beyond 24 weeks with structural and sonographically detectable birth defects from October,2014 to October, 2018 in the Department of Obstetrics and Gynaecology of Bangladesh Medical College and Hospital were carefully scrutinized and detailed information regarding the maternal and associated clinical risk factors were compiled using the NBBD (New Born Birth Defects) surveillance system. Among that period all births (Live birth and still birth) were counted to have a prevalence data of birth defects using the total number of births as the denominator and the number of birth defects as the numerator.*

***Results:*** *The prevalence of detectable birth defects among the 2002 total births (which includes 110 still births) was found to be 4.34% (87/2002 x 100). According to birth defect category using the ICD-10 coding system, 11 broad categories were found. Musculoskeletal deformities Q65-Q79 were the highest (25/87), followed by congenital malformation of the nervous system Q00-Q07(15/87) and congenital malformation of eye, ear, face and neck Q10-Q18(14/87). The birth defects were categorized as isolated, syndrome and sequence; among the 87 cases, 44 were isolated defects, 40 were syndromic / multiple birth defects and 3 were result of Potter sequence.*

*Regarding maternal variables, maternal age<18 years was 23.4%, 18-25 years was 48.93% ,26-33 years was 23.4% and ≥ 34 years was 6.4%; father's age < 35 yearswas 74.5% and ≥ 35 years 25.5%%, parental consanguinity was present in 4.3% of case. Analyzing the variables relating to labour conditions, majority of pregnancies were singletons 95.7% leaving only 4.3% of pregnancies being Twin pregnancies. Reviewing babies according to gestational age, 69 (73.4%%) of babies were less than 34 weeks and 26.6% remaining were equal to/more than 34 weeks of gestation reflecting a higher frequency of prematurity or pre-term delivery either induced or spontaneous onset. Regarding the mode of delivery, vaginal birth was conducted in approximately 74% of cases and C-Section was performed in remaining cases, the indication of C-section was guided by obstetric causes such as previous C-section and maternal desire for an elective abdominal delivery.*

*Results of the foetal variables by sex distribution showed a significant male predominance (51/87) 51 male, 26 female and 10 ambiguous. Reviewing babies according to gestational age, 64 (73.4%%) of babies were less than 34 weeks and 26.6% remaining were more than 34 weeks of gestation reflecting a higher frequency of prematurity. The studied foetal variable as categorized by weight, as ≤1500gm (extreme low birth weight ELBW) was 23.4%, 1501-2499gm (Low birth weight LBW) was 50% and ≥2500g (Average birth weight) was 26.6 %. The studied foetal variable as categorized by percentage of babies that were born live birth was 87%, 17 % were stillbirth: a significant portion of those terminated late were found macerated. Data was also compiled regarding the following risk factors: Previous history of birth defects/ previous still birth/ previous spontaneous abortions/ terminations for birth defects which did not reveal significant differences.*

***Conclusion:*** *The study notified only the most visible defects in most cases. However, the study is part of an ongoing surveillance program which has incited much alertness among the participants regarding documentation. The prevalence records and the type of defects may help in the expansion of these programs for the development of future preventive strategies.*

## Introduction

Birth defects (also called congenital anomalies) include all functional and structural anomalies arising from factors originating before birth, even when the abnormality is not apparent in the new born, and is later recognized. From a biological standpoint, birth defects (BDs) represent a heterogeneous group of embryonic-fetal development disorders with distinct etiologic factors often involved simultaneously. The monogenic genetic or chromosomal conditions are responsible for 15-20% of cases. The multifactorial etiology, with polygenic genetic component associated with environmental factors, is implicated in another 20%. Maternal conditions, such as diabetes and obesity, advanced maternal age, environmental exposure or teratogens, especially congenital infections, and exposure to drugs, alcohol, and illicit drugs, are known to account for a significant number of defects [1]. The etiology of the disorder is unknown in about 50 to 60% of cases [2]. Consanguinity contributes to the load of autosomal recessive disorders with variants of phenotypic expressions.

Globally, about 7.9 million children are born annually with a serious birth defect. WHO estimates that out of 1 million neonatal deaths in 2012 in South East Asia Region, about 46000 (4.6%) was caused by birth defects. Approximately 2-4% of all infants are born with a major birth defect (CDC, 2013) [3]. Birth defects are one of the leading global causes of infant mortality, accounting for more than 20% of all infant deaths [4].

The leading causes of neonatal death in South East Asia are still neonatal sepsis, perinatal asphyxia and complications of prematurity. With the emergence of comprehensive obstetric and neonatal care in regard to the prevention of perinatal asphyxia, labour dystocia and prematurity, birth defects will emerge as the leading cause of neonatal intensive care/pediatrics admission even in resource -restricted set-up. The birth defects spectrum involves different organ systems, the most common being congenital heart disease (CHD) (0.5-0.8% of all live births), neural tube defects (NTD) (0.2-0.4% of all live births), Trisomy 21 followed by Haemoglobinopathies and musculoskeletal disorder [5]. The genetics of most of these defects are multifactorial; therefore, antenatal screening for all birth defects is not possible. However, some disorders are amenable to prenatal screening and diagnosis. These include aneuploidies (Trisomy 21, Trisomy 18.), Cystic Fibrosis and Sickle Cell Disease [6].

Adequate and verified data and information on birth defects is not available in South East Asian region. Such valid data is important to understand the public health implication and design preventive and awareness strategies in the country which may include preconceptional vaccination against rubella, folic acid supplementation and fortification of staple foods with micronutrients (iodine and folic acid), prevention and management of syphilis and preconceptional genetic counseling for affected/ high risk cases [6].

This study is part of an ongoing hospital-based surveillance program which was conducted with the aim to evaluate the prevalence of birth defects and to investigate certain maternal and perinatal associated facts in all inborn births and terminations of structural and sonographically detectable birth defects from October, 2014 to October, 2018 in the Department of Obstetrics and Gynaecology of Bangladesh Medical College and Hospital.

## Methods

The present study was a descriptive/analytical study conducted in the Department of Obstetrics & Gynaecology and Department of Pediatrics, Bangladesh Medical College, the first Private /non-government medical college in Bangladesh working under the Parent institution BMSRI (Bangladesh Medical Service and Research Institute) in Dhaka, capital of Bangladesh. It serves as a private center for at least 10 sub-urbs or localities bearing a majority of middle -income inhabitants with a population density of above 2000 person/km$^2$ and a human development index of 0.6. The research was based on data collected using a data collection sheet/questionnaire which is provided in the Appendix. The Department of Obstetrics& Gynaecology of Bangladesh Medical college has 54 beds which includes antenatal and post-natal beds, eclampsia ward and labour observation suite. The hospital is equipped with a neonatal intensive care unit with 12 beds, supervised by neonatologists and specialists. It has an average 75 births per month, contributing to a small scale in comparison to the large population burden.

The study considered all inborn births (live born and still born) and terminations of pregnancy beyond 24 weeks with structural and sono-graphically detectable birth defects from October, 2014 to October, 2018 in the Department of Obstetrics and Gynecology of Bangladesh Medical College and Hospital. Among that period all births (Live birth and still birth) were counted to have a prevalence data of birth defects using the total number of births as the denominator and the number of birth defects as the numerator. The out born cases were excluded, as including those cases would hamper the prevalence record. The following information were collected using a data collection, sheet/questionnaire: mother's age, father's age, parental consanguinity, baby's gender, birthweight, Gestation, History of birth defects: Previous termination of pregnancy for birth defects, previous pregnancy affected with Birth defect, previous still birth, previous spontaneous abortion(s), the birth defect with full description and photographs and ICD-10 coding. After gaining consent from the parents and maintaining confidentiality, new born with birth defect was photographed which included a comprehensive coverage and detailing of the birth defect? This photograph along with the gross physical description were verified and if needed corrected by the Surveillance committee. In certain cases, photography was not feasible.

In total, we analyzed 9 variables:6 maternal variables, 3 foetal variables. The variable analyzed were: maternal age, categorized as <18 years, 18-25 years, 26-33 years and ≥ 34 years; father's age < 35 years and ≥ 35 years, parental consanguinity. The variables relating to labour conditions were: type of pregnancy, categorized as single or multiple; gestational age, categorized as less than 34 weeks and more than 34 weeks and mode of delivery, categorized as vaginal or cesarean section. The studied foetal variable were gender,

categorized as male, female and ambiguous; weight, categorized as, 1500g, 1501-2499g and ≥2500g and outcome, categorized as livebirth, stillbirth: fresh /macerated.

The statistical analysis was conducted with Graphpad Prism software, version 4.0. There were descriptive statistics of the data and the chi-square and Fisher exact test for comparison of variables, according to the number of categories of each variable analyzed, establishing significance at 5%.

## Results

The prevalence of detectable birth defects among the 2002 total births (which includes 110 still births) was found to be 4.34% (87/2002x100). Among the 2002 live births, 87 birth defects with ICD-10 coding of each individual case were identified and verified by the New Born Birth Defect surveillance program in collaboration with Center for Disease Control CDC, Atlanta and All India Institute of Medical Science AIMS, New Delhi and Bangabandhu Sheikh Mujib Medical University BSMMU as the Focal point of investigation.

According to birth defect category using the ICD-10 coding system,11 broad categories were found. Musculoskeletal deformities Q65-Q79 were the highest, followed by congenital malformation of the nervous system Q00-Q07 and congenital malformation of eye, ear, face and neck Q10-Q18. The birth defects were categorized as isolated, syndrome and sequence; among the 87 cases, 44 were isolated defects, 40were syndromic / multiple birth defects and 3 were result of Potter sequence.

Regarding maternal variables, maternal age<18 years was 23.4%, 18-25 years was 48.93% , 26-33 years was 23.4% and ≥ 34 years was 6.4%; father's age < 35 yearswas 74.5% and ≥ 35 years 25.5%%, parental consanguinity was present in 4.3% of case. Analyzing the variables relating to labour conditions, majority of pregnancies were singletons 95.7% leaving only 4.3% of pregnancies being Twin pregnancies. Reviewing babies according to gestational age, 69 (73.4%%) of babies were less than 34 weeks and 26.6% remaining were equal to/more than 34 weeks of gestation reflecting a higher frequency of prematurity or pre-term delivery either induced or spontaneous onset. Regarding the mode of delivery, vaginal birth was conducted in approximately 74% of cases and C-Section was performed in remaining cases, the indication of C-section was guided by obstetric causes such as previous C-section and maternal desire for an elective abdominal delivery.

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**Table 1:Type of Birth Defects According To The Icd-10 Code And Percentage**

| Birth defect Category | Birth defect type sub-category | ICD -10 code | n (87) | %(≈) |
|---|---|---|---|---|
| Congenital malformation and deformation of the musculoskeletal system (25) 28.7% | Congenital deformity of the hip, unspecified | Q65.9 | 2 | 2.3 |
| | Talipes equinovarus | Q66.0 | 4 | 4.6 |
| | Congenital pes planus | Q66.5 | 1 | 1.15 |
| | Rocker bottom foot | Q66.8 | 2 | 2.3 |
| | Dolicocephaly | Q67.2 | 1 | 1.15 |
| | Congenital deformity of the hand | Q68.1 | 1 | 1.15 |
| | Clinodactyly | Q68.10 | 3 | 3.45 |
| | Congenital genu recurvatum | Q68.21 | 1 | 1.15 |
| | Accessory finger(s) | Q69.0 | 1 | 1.15 |
| | Other reduction defects of lower limb(s) | Q72.8 | 1 | 1.15 |
| | Longitudinal reduction defect of femur | Q72.4 | 1 | 1.15 |
| | Other reduction defects of upper limb(s) | Q71.8 | 1 | 1.15 |
| | Thanatophoric short stature | Q77.1 | 2 | 2.3 |
| | Congenital diaphragmatic hernia | Q79.0 | 2 | 2.3 |
| | Exomphalos | Q 79.2 | 2 | 2.3 |
| Congenital malformation of the nervous system (15) 17.2% | Nasofrontal encephalocele | Q 01.1 | 1 | 1.15 |
| | Occipital encephalocele | Q01.2 | 1 | 1.15 |
| | Microcephaly | Q 02 | 4 | 4.6 |

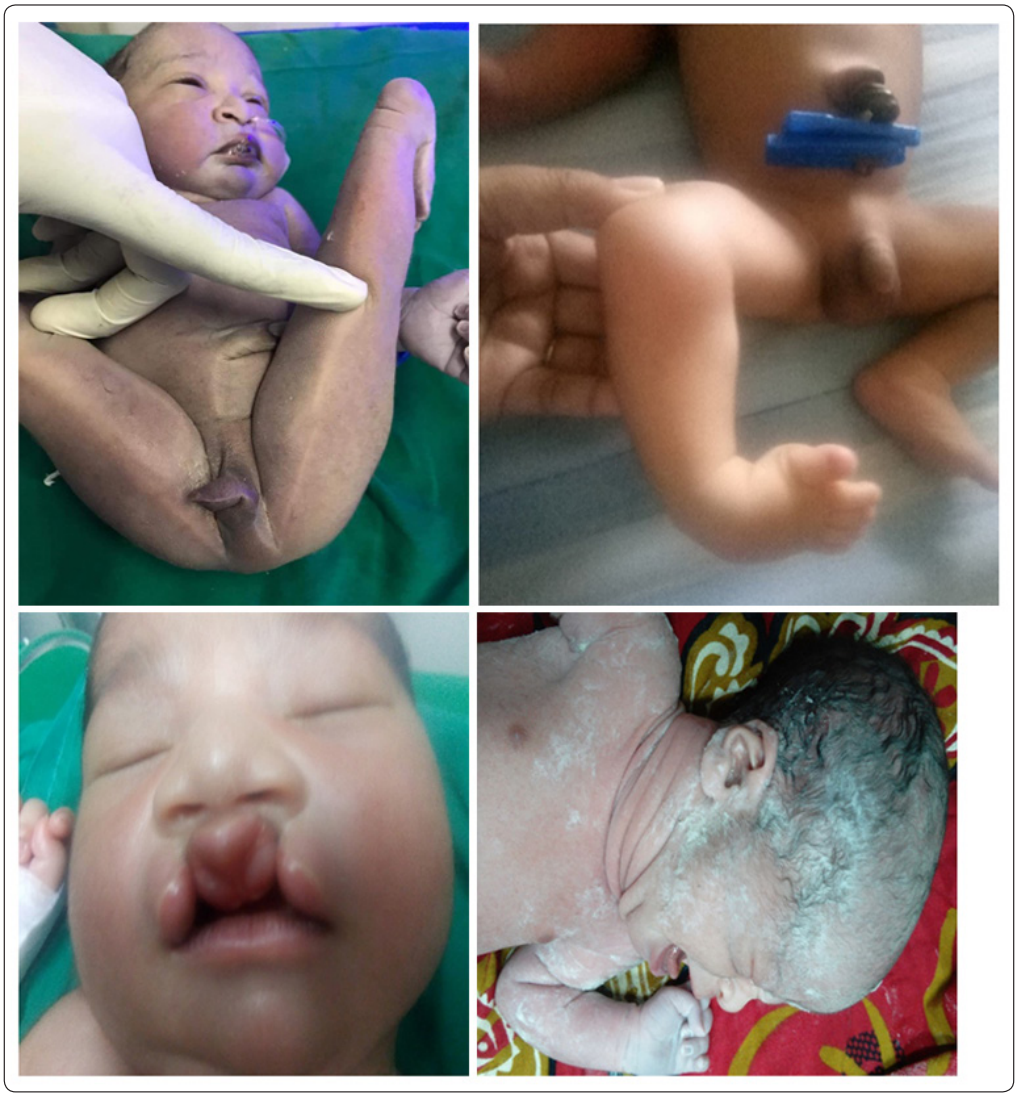
| | | | | |
|---|---|---|---|---|
| | Congenital hydrocephalus | Q 03 | 4 | 4.6 |
| | Holoprosencephaly | Q04.2 | 3 | 3.45 |
| | Sacral spina bifida with hydrocephalus | Q05.3 | 1 | 1.15 |
| | Lumbar spina bifida without hydrocephalus | Q05.7 | 1 | 1.15 |
| Congenital malformation of the eye, ear, face and neck (14) 16.09% | Congenital ectropion | Q10.1 | 1 | 1.15 |
| | Other congenital malformations of the eyelid | Q10.3 | 2 | 2.3 |
| | Anophthalmos,microphthalmos and macrophthalmos | Q11 | 3 | 3.45 |
| | Misplaced ear | Q17.4 | 3 | 3.45 |
| | Other congenital malformation of the face and neck | Q18 | 1 | 1.15 |
| | Sinus, fistula and cyst of the brachial cleft | Q18.0 | 2 | 2.3 |
| | Unspecified malformation of the face and neck | Q18.9 | 2 | 2.3 |
| Congenital malformation of the genital organs (7)8.04% | Congenital malformation of the genital organ, unspecified | Q52.9 | 1 | 1.15 |
| | Undescended testis | Q 53 | 1 | 1.15 |
| | Undescended testis, bilateral | Q53.2 | 1 | 1.15 |
| | Hypospadias, balanic | Q54.0 | 1 | 1.15 |
| | Congenital absence and aplasia of the penis | Q55.5 | 1 | 1.15 |
| | Indeterminate sex, unspecified | Q56.4 | 1 | 1.15 |
| Cleft lip and palate (6) 7% | Cleft hard palate, unspecified | Q35.19 | 1 | 1.15 |
| | Cleft lip, bilateral | Q36.0 | 1 | 1.15 |
| | Cleft hard palate and hard lip, | | | |
| | bilateral | Q36 | 1 | 1.15 |
| | Cleft hard palate with cleft lip, unilateral | Q37.1 | 3 | 3.45 |
| Congenital malformation of the urinary system (5) 6% | Renal agenesis, unspecified | Q60.2 | 1 | 1.15 |
| | Potter's syndrome | Q60.6 | 1 | 1.15 |
| | Congenital malformation of the kidney, unspecified | Q63.9 | 1 | 1.15 |
| | Congenital posterior urethral valves | Q64.20 | 1 | 1.15 |
| | Congenital malformation of the urinary system, unspecified | Q64.9 | 1 | 1.15 |
| Congenital malformation of the circulatory system (3) 4% | Ventricular septal defect | Q 21.0 | 1 | 1.15 |
| | Common atrioventricular canal | Q21.21 | 1 | 1.15 |
| | Other specified congenital malformation of the circulatory system | Q 28.8 | 1 | 1.15 |
| Congenital malformation of the respiratory system (1) 1.14% | Agenesis and under-development of the nose | Q30.1 | 1 | 1.15 |
| Congenital malformation of the digestive system (1) 1.14% | Atresia of oesophagus without fistula | Q39.0 | 1 | 1.15 |
| | Congenital absence, atresia, stenosis of anus without fistula | Q42.3 | 2 | 2.3 |
| | Hirschsprung's disease | Q43.1 | 1 | 1.15 |

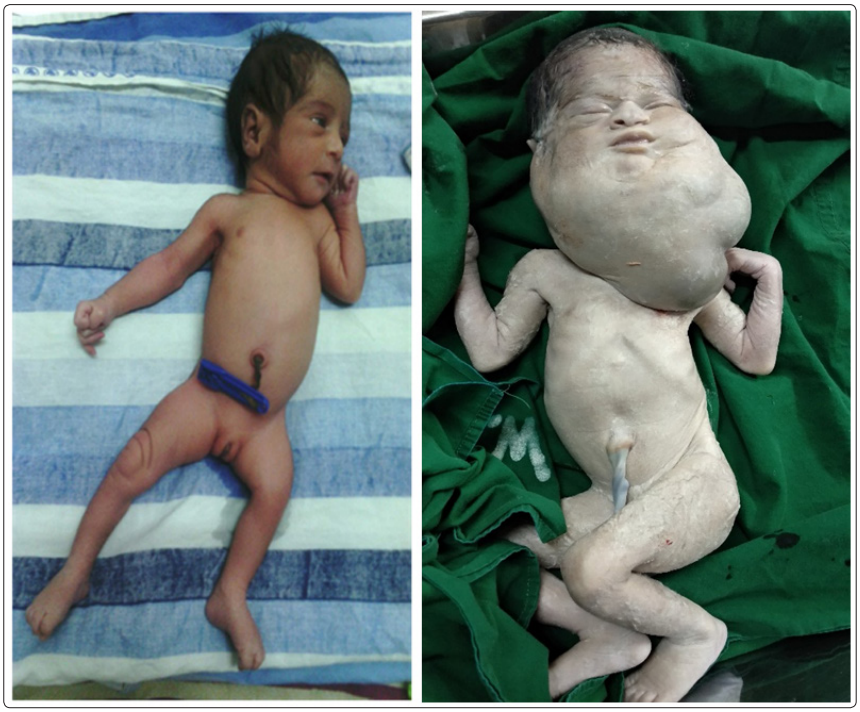
| | | | | |
|---|---|---|---|---|
| Othercongenital malformations (4) 4.56% | Other congenital malformation of the skin | Q82 | 2 | 2.3 |
| | Congenital malformation syndromes affecting facial appearance | Q87.0 | 2 | 2.3 |
| Chromosomal abnormalities, not elsewhere classified (3) 3.42% | Down's syndrome, unspecified | Q90.9 | 2 | 2.3 |
| | Chromosomal abnormality, unspecified | Q99.9 | 1 | 1.15 |
| Total | | | 87 | 100 |

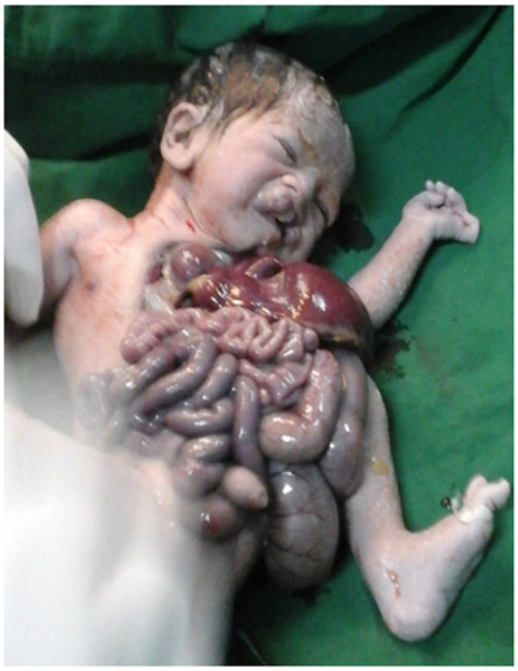
**Table 2: Distribution of Maternal and Delivery Variables among Newborns with Birth Defects**

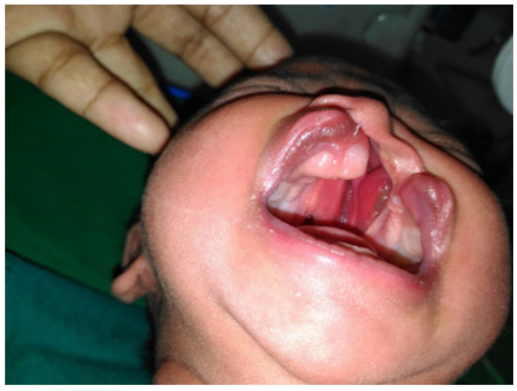
| **Maternal variables** | | | |
|---|---|---|---|
| Maternal Age | n (87) | Percentage % | p-value |
| ≤18 years | 18 | 23.4% | NS |
| 18-25 years | 42 | 48.93% | |
| 26-33 years | 18 | 23.4% | |
| ≥ 34 years | 07 | 6.4% | |
| **Parental consanguinity** | | | |
| Yes | 04 | 04.3% | NS |
| No | 83 | 95.7% | |
| Father's age | | | NS |
| < 35 years | 65 | 74.5% | |
| ≥ 35 years | 22 | 25.5% | |
| **History of birth defects** | | | |
| Previous termination of pregnancy for birth defects: | | | NS |
| Yes | 07 | 8.1 % | |
| No | 80 | 91.9% | |
| **Previous pregnancy affected with BD** | | | |
| Yes | 07 | 08.1% | NS |
| No | 80 | 91.9% | |
| **Previous Still birth** | | | |
| Yes | 10 | 11.7% | NS |
| No | 77 | 88.3% | |
| **Previous spontaneous abortion(s)** | | | |
| Yes | 25 | 28.7% | NS |
| No | 62 | 71.3% | |
| **Foetal variable** | | | |
| **Foetal Weight** | | | |
| ≤1500gm (extreme low birth weight ELBW) | 21 | 23.4% | NS |
| 1501-2499gm (Low birth weight LBW) | 43 | 50% | |
| ≥2500g (Average birth weight) | 23 | 26.6 % | |
| **Gestation** | | | |
| ≤ 34 weeks | 64 | 73.4%% | NS |
| >34 weeks | 23 | 26.6% | |
| **Sex Distribution** | | | |
| Male | 51 | 54% | NS |
| Female | 26 | 35% | |
| Ambiguous | 10 | 11% | |
| **Multiple Birth** | | | |

| | | | |
|---|---|---|---|
| Single | 83 | 95.7% | NS |
| Twin | 04 | 04.3% | |
| Triplet | 00 | 0.0% | |
| Higher order | 00 | 0.0% | |
| **Mode Of Delivery** | | | |
| Vaginal | 64 | 73.4% | NS |
| C-section | 23 | 26.6% | |
| **Foetal Outcome** | | | |
| Livebirth | 72 | 83% | NS |
| Stillbirth | 15 | 17% | |









## Discussion

Birth defects have adverse effects on the wellbeing and survival of children born with those anomalies. In analysis of birth defects in relation to pre-maturity and birth weight, a study concluded that birth defects are associated with preterm birth and low birth weight after controlling for multiple confounding factors, including shared risk factors and pregnancy complications. In that study, a singleton liveborn infant with a birth defect was 2.7 times more likely to be delivered preterm before 37 weeks of gestation (95% confidence interval [CI] 2.3-3.2), 7.0 times more likely to be delivered preterm before 34 weeks (95% CI 5.5-8.9), and 11.5 times more likely to be delivered very preterm before 32 weeks (95% CI 8.7-15.2). A singleton liveborn with a birth defect was 3.6 times more likely to have low birth weight at less than 2,500 g (95% CI 3.0-4.3) and 11.3 times more likely to be very low birth weight at less than 1,500 g (95% CI 8.5-15.1) [3].In our study 64 (73.4%%) of babies were less than 34 weeks and 26.6% remaining were more than 34 weeks of gestation reflecting a higher frequency of prematurity or pre-term delivery either induced or spontaneous onset and 74% of babieshad birth weight below 2500gms.

Multiple pregnancies have about twice the risk of congenital abnormalities including Down syndrome, club foot, neural tube defects (such as spina bifida), gastrointestinal, and heart abnormalities as comparing to singleton pregnancies making the risk of 6-8%. However, in our study, twin pregnancies contributed to only 4.3% of cases of birth defects [7].

A study conducted by the Department of Surgery, Chittagong Medical College on Birth defects from 2008-2012 revealed there were 5661 patients of birth defects admitted in this department [8]. Of these, 5156 had a single congenital anomaly and 505 had multiple congenital anomalies. The male to female ratio was 2.1: 1. Birth defects comprised 44.61% of all Pediatric surgical admissions and 0.90% of total hospital admissions. The gastrointestinal system was the most common organ system involved, followed by the genitourinary system. Inguinal hernias were the most common gastrointestinal abnormality and hypospadias were the most common genitourinary case. Most corrective operations were done for gastrointestinal and genito-urinary defects. Inguinal herniotomy was the most commonly performed operation followed by laparotomy for various indications. There were 225 deaths for birth defects which represents 51.49% of all (pediatric) deaths during the study period. The most common cause of death was anorectal malformations followed by gastroschisis.This study conducted in the department of surgery reflects the value of documentation in surgical sub-

specialities. It also can be commented that birth defects can manifest in a variety of departments at varying ages; particularly congenital heart disease (acyanotic varieties).

Potential teratogens/factors that have been identified in developing countries as contributors to birth defects include low socioeconomic and educational levels, malnutrition (mineral and vitamin deficiencies), intrauterine infections, lack of environmental protection policies, environmental pollution, unsafe working conditions during pregnancy, access to medicines without medical indication or prescription (self-medication), and common use of home remedies of unknown composition. Clearly documenting the magnitude of known risk factors that influence the occurrence of birth defects will help to develop preventive strategies [7]. Globally there is a scarcity of prevalence data of structural birth defects. From this incentive to compile global data the CDC has started this surveillance program with the future intent to build capacity of birth defect surveillance in hospitals around the world and set up contextual preventive strategies with long-term health impact.

Major musculoskeletal birth defects, including craniosynostosis, gastroschisis, diaphragmatic hernia, and transverse limb reduction deficiencies, are an important public health issue of largely unknown etiology with evidence of increased prevalence for gastroschisis. However, majority of musculoskeletal defects can be improved with the help of surgical treatment and usually involves the reconstructing of disfigured or missing parts of the body [10]. In this study, newborns born with isolated talipes and certain limb reduction defects were subsequently referred to Orthopeadic subspeciality.

Facial dysmorphisms sometimes can go unnoticed. Therefore, the awareness of the attending physician is paramount to detect such subtle external features. A list of facial dysmorphism that should be clinically suspected in the neonatal period may lead to the diagnosis of Down syndrome/Trisomy 21, Edwards syndrome/Trisomy 18 and Patau Syndrome/Trisomy 13,Turner syndrome,Congenital syphilis, Laurence-Moon-Biedl syndrome, Di George Syndrome, Noonan syndrome [11]. A study conducted in Campinas, in 2004, with 2,843 live births, with 92 cases of birth defects suggesting that minor anomalies are hardly recognized or are neglected during the filling of the questionnaire. Many minor BDs are dysmorphias, i.e., normal variants of the phenotype without clinical relevance. Although they may indicate general changes in morphogenesis, they eventually serve as a diagnostic clue for severe BDs or even malformation syndromes. In fact, 90% of infants with three or more minor BDs present a major BD, and minor BDs often occur in malformation syndromes; 43% of patients with idiopathic intellectual disability present three or more BDs, of which 80% are minor [11].

We encountered 2 cases of neck cysts over this period, one case of thyroid goiter and one case of multiple lymphangioma/cystic hygroma.

We encountered a case of holoprosencephaly with gross facial disfiguration at 35 weeks with moderate growth retardation (Birth weight1700 gm, female). The intracranial defect was diagnosed on antenatal sonography and the baby was fresh still born. This newborn was later suspected to have Patau syndrome/Trisomy 13.

We encountered a female baby of 1900 gm at 34 weeks vaginally with suspected achondroplasia as suspected by gross abduction of femurs with extension of both kneesand spastic limbs.

Among the cases of cleft lip, we encountered a case of gross unilateral cleft lip and palate We encountered a female of 1600gm at 29 weeks with omphalocele/exomphalos which was delivered liveborn later succumbed due to non-intervention.

A study about consanguinity and its relevance to clinical genetics suggested that autosomal recessive and multifactorial disorders had the highest values of consanguinity (78.8%, 69.8%, respectively), while chromosomal disorders had the lowest one (29.1%). However, we found no significant percentage of parental consanguinity among the cases of Birth defects [12].

## Conclusion

All women who present for antenatal care before 20 weeks gestation should be offered screening for fetal birth defects and aneuploidy. Patients should be counseled on the difference between screening and diagnostic testing. The choice of tests depends on several factors including gestational age at the initial prenatal care visit, patient history, number of fetuses, and availability of nuchal translucency measurementand the facility of corrective surgery in cases of certain birth defects. Regardless of which tests are offered, information provided about each test should include the purpose of the test, the detection and false-positive rates, and the limitations of testing. In addition, information on each test's risks and benefits should be given to the patient so she can make an informed decision regarding the prognosis in case of major birth defect.

Preconception folic acid supplementation of 400 µg per day for low-risk women and 4 mg/day for high-risk (previous pregnancy with NTD, patient or partner with NTD) women reduces the risk of NTD by approximately 60-70%.All pregnant women should be offered second-trimester MSAFP screening for NTD or should have a careful anatomic assessment of the CNS anatomy by a detailed anomaly scan.Women with elevated AFP should be referred for genetic counseling and offered a diagnostic test such as a targeted sonographic evaluation and potentially amniocentesis. The fetus with a Neural Tube Defect should be delivered at a tertiary care facility capable of managing all of the neonatal issues.Avoidance of teratogenic drugs particularly retinoic acid, tetracycline and misoprostol are among a few medications that should be avoided with care.

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