

# Original Articles

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## Frequency of Birth Defects in a Tertiary Care Hospital in Bangladesh

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### Abstract

**Background:** Birth defects usually present at birth and may cause morbidity and mortality in the newborn. These defects may involve any organ system of the body. Gradually the incidence is rising due to various etiologies. The objective of the study was to find out the frequency, types and etiology of birth defects.

**Materials & Methods:** This cross-sectional study was done in a tertiary care hospital in Bangladesh. In this study, 100 neonates with birth defects were enrolled, thoroughly examined, investigated and evaluated. Then collected data were analyzed.

**Results:** The frequency of birth defects in this study was 1.54%. The most frequent birth defect was club foot deformity i.e. 11% and the most frequently involved organ system was cardiovascular system i.e. 28%. Among the birth defects 78% were major and only 22% were minor. Among those children with birth defect mortality was 17%; 41% were referred to advanced center and 42% were improved and discharged. In some of the cases with birth defect chromosomal anomaly, congenital infection (TORCH), maternal diabetes was found as causing factor.

**Conclusion:** The frequency of birth defects was found 1.54% and most of the birth defects were major defect. Club foot deformity was found as the most common birth defect.

**Key words:** Birth defects, frequency.

### Introduction:

The overall incidence of birth defects is about 2-4% of all neonatal population and affects the newborn health with long term morbidity and mortality.<sup>1</sup> Although there are many types of birth defects, some are common, e.g. neural tube defect, cleft lip-cleft palate, congenital heart disease, club foot deformity, etc.<sup>2</sup> There is a sharp rise in the prevalence of birth defects recently.<sup>3</sup> Genetic or chromosomal diseases, toxins, drugs, chemicals, radiation, maternal diabetes, congenital infections e.g. TORCH, Zika or

HIV and maternal habit disorders e.g. smoking, alcohol, drug abuse etc. are responsible for increased number of congenital anomalies in newborns.<sup>4</sup>

A birth defect is a problem that occurs when a baby is developing in utero.<sup>5</sup> Birth defects can be minor or major.<sup>6</sup> They may affect appearance, organ function, and physical and mental development.<sup>7</sup> Most birth defects are present within the first three months of pregnancy, when the organs are still forming.<sup>8</sup> Minor birth defects e.g. syndactyly, polydactyly are usually harmless; while major birth defects e.g. anencephaly, congenital heart defect, gastroschisis or hydronephrosis may cause death. Major birth defects are the leading cause of infant death in the United States, accounting for 20 percent of deaths.<sup>9</sup> Among the numerous birth defects, some are more common than the others such as the most common is those related to musculoskeletal system,

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followed by genitourinary system, cardiovascular system and gastrointestinal system.<sup>10</sup> Birth defects are typically classified as structural or functional and developmental.<sup>11</sup> Structural defects are when a specific body part is missing or malformed. The most common structural defects are: heart defects, cleft lip-cleft palate, spina bifida and club foot deformity.<sup>12</sup> Functional or developmental birth defects include metabolic defects, sensory problems, and nervous system problems causing a body part or system not to work properly. These often cause disabilities of intelligence or development.<sup>12</sup> The most common types of functional or developmental birth defects include: Down syndrome, sickle cell disease and cystic fibrosis.<sup>12</sup> All pregnant women have some risk of delivering a baby with a birth defect.<sup>13</sup> Risk increases with any of the following conditions: family history of birth defects or other genetic disorders, drug abuse, alcohol consumption, or smoking during pregnancy, maternal age of 35 years or older, inadequate prenatal care, untreated viral or bacterial infections, including sexually transmitted diseases, use of certain high-risk medications, such as isotretinoin and lithium.<sup>14</sup> Women with pre-existing medical conditions, such as diabetes, are also at a higher risk of having a newborn with a birth defect.<sup>15</sup> Prenatal ultra sounds, blood tests and amniocentesis may help diagnosis of certain birth defects in utero.<sup>16-18</sup> Physical examination and hearing test may also help to diagnose birth defects after the baby is born.<sup>19</sup> A blood test called the newborn screening can help doctors to screen some birth defects shortly after birth, before the symptoms occur.<sup>20</sup> Most birth defects can be diagnosed with certainty after birth.<sup>21</sup> Banu T et al.<sup>22</sup> and Chowdhury K et al.<sup>23</sup> studied on birth defects in Bangladesh and musculoskeletal system (37.88%) was found as the most commonly affected system and the most frequent lesions were club foot and arthrogyroposis.<sup>23</sup> The objective of the study was to find out the frequency, etiology and types of birth defects in a tertiary care hospital.

#### Materials & Methods:

The study was a cross-sectional study conducted at the Neonatal ward in the Sher-e-Bangla Medical College Hospital, Barishal from July 2016 to December 2017. The inclusion criterion was live born neonates with apparent birth defects, irrespective of gestational age, birth weight, sex, race or ethnicity. The only exclusion criterion was stillbirth. The study subjects were included in the study after obtaining informed consent from the parents. Ethical permission was obtained from the ethical committee

of the hospital to conduct the study. Total 100 consecutive neonates with apparent birth defects were included in this study. All the neonates with apparent birth defects were thoroughly examined clinically by the principal investigator himself within 24 hours after admission in the ward. Radiological or imaging studies, echocardiography, hematological and biochemical studies were done. The birth defects were evaluated according to the objective of the study. Etiology of the birth defects were evaluated by history (e.g. drug abuse, exposure to toxin, smoking, tobacco or alcohol), maternal medical record (e.g. maternal diabetes), clinical examinations, laboratory investigations (e.g. TORCH screening) and echocardiography. In case of newborn with suspected chromosomal disorder karyotyping was done. Gestational age was calculated by New Ballard score; anthropometric measurements e.g. weight, length and head circumference were measured by neonatal weighing scale, infantometer and non-elastic measuring tape respectively. Statistical analysis was done by using SPSS version 16.

#### Results:

During the study period, the total number of birth in the hospital was 6613, the number of live birth was 6480 and still birth was 133. Among those 100 neonates with birth defects were included in the study. The demographic data including gestational age, weight at admission, length, head circumference, sex, mode of delivery were shown in Table I. The frequency of birth defects was 1.54% shown in Table II. Among total birth defect (n=100), major birth defects were 78% and minor birth defects were 22%. In some newborn more than one birth defect was found. In this study, the birth defects were most commonly found involving the cardiovascular system (i.e. 28%), followed by musculoskeletal system (24%).

**Table I**

*Base-line data of the study subjects (n=100).*

Characteristics	Clinical data
Gestational age (wk)	35.28 ± 2.3
Weight at admission (gm)	2156 ± 134
Length (cm)	47.74 ± 0.6
Head circumference (cm)	33.42 ± 1.4
Male / female	53/ 47
Mode of delivery (cesarean/vaginal)	65/35

**Table II**  
*Frequency of birth defects.*

No. of birth defect	No. of live birth
100	6480
Frequency of birth defects	100/6480 = 1.54%

**Table IV**  
*Frequency of major and minor birth defects among the study subjects (n=100).*

Type of birth defect	No. (%)
Major birth defect	78 (78%)
Minor birth defect	22 (22%)

**Table III**  
*Etiology of the birth defects.*

Etiology of birth defect	No.	Clinical birth defect
Chromosomal	05	Down syndrome, Edward syndrome, Turner syndrome
Maternal diabetes	05	Congenital Heart defects (VSD, ASD, PDA)
Congenital infection (TORCH/HIV)	02	Congenital rubella syndrome (CRS), Toxoplasmosis

**Table V**  
*Frequency and types of birth defects.*

Organ system	Birth defect	N (%)
Musculoskeletal system	Talipesquinovarus (club-foot deformity)	11 (11%)
	Arthrogryposis multiplex congenita	3 (3%)
	Polydactyly	2 (2%)
	Syndactyly	2 (2%)
	Ectrodactyly	2 (2%)
	Pierre-robin syndrome	2 (2%)
	TAR (thrombocytopenia, absent radius) syndrome	1 (1%)
	Sirenomelia	1 (1%)
	Gastrointestinal system	Cleft-lip, cleft-palate
Ano-rectal malformation		4 (4%)
Gastroschisis		2 (2%)
Omphalocele		2 (2%)
Duodenal atresia		1 (1%)
Diaphragmatic hernia		1 (1%)
Hirschprung disease		1 (1%)
Genitourinary system	Congenital polycystic kidney disease	2 (2%)
	Hydronephrosis due to PUJ obstruction	2 (2%)
	Exstrophy of the bladder	1 (1%)
	Posterior urethral valve	1 (1%)
	Hypospadias	3 (3%)
	Micropenis	2 (2%)
	Undescended testis (cryptorchidism)	2 (2%)
	Ambiguous genitalia	2 (2%)
	Hydrocele	1 (1%)
	Cloacal exstrophy	1 (1%)
Polythelia (supernumerary nipple)	1 (1%)	
Central nervous system	Hydrocephalus	4 (4%)
	Neural tube defect (spina bifida, myelomeningocele)	4 (4%)
	Anencephaly	2 (2%)

Organ system	Birth defect	N (%)
	Microcephaly	2 (2%)
	Craniosynostosis	2 (2%)
	Encephalocele	1 (1%)
Cardiovascular system	Ventricular septal defect (VSD)	8 (8%)
	Atrial septal defect (ASD)	6 (6%)
	Persistent ductus arteriosus (PDA)	5 (5%)
	Bicuspid aortic valve (i.e. Turner syndrome)	1 (1%)
	Pulmonary stenosis (i.e. congenital rubella syndrome)	1 (1%)
	Tetralogy of Fallot (TOF)	3 (3%)
	Transposition of Great arteries (TGA)	2 (2%)
Dextrocardia	2 (2%)	
Genetic / chromosomal	Down syndrome (VSD)	3 (3%)
	Edward syndrome (PDA, VSD, etc.)	2 (2%)
	Turner syndrome (bicuspid aortic valve)	1 (1%)
	Congenital infection (TORCH) (PS, cataract, etc.)	1 (1%)
Eye	Cataract	2 (2%)
	Microphthalmia	1 (1%)
	Aniridia	1 (1%)
	Hypertelorism	1 (1%)
	Hypotelorism	1 (1%)
	Enophthalmos	2 (2%)
	Ear	Microtia
Anotia		1 (1%)
Skin-soft tissue, vascular	Hemangioma	2 (2%)
	Collodion baby	2 (2%)
	Port-wine stain (i.e. Sturge-Weber syndrome)	1 (1%)

Total 126 birth defects were found among those 100 neonates in the study, since there were multiple birth defects present in 27 neonates. Sometimes, multiple birth defects were found in the same subject and in this study it was 27% and single birth defects was 73%. Among the 100 neonates, 41% neonates were referred to advanced center for further management, 42% neonates were discharged and 17% died.

### Discussion:

The frequency of birth defects in this study was 1.54%. In the United States, they occur in about 3% of newborns.<sup>21</sup> Worldwide the incidence of birth defects ranges between 2-4%.<sup>1</sup> In the study done by Yang et al.<sup>18</sup> the prevalence of birth defect was 1.5% and was 2.9% in the study by Mosquera et al.<sup>24</sup> and in Bangladesh, it was 3.3% in the study conducted by Chowdhury K et al.<sup>23</sup> There was a male predominance in birth defects i.e. 53% vs. 47% found in this study; the male-female ratio was 2.1:1 in the study done by Banu T et al.<sup>22</sup> and 53% vs. 43.8% found in the study by Chowdhury K et al.<sup>23</sup> and similar result was found in study conducted by Jain SR et al.<sup>27</sup> (51.6% vs. 48.4%) and also by Zheng et al.<sup>25</sup>

In this study, etiology of birth defects was chromosomal anomaly e.g. Down syndrome, Edward syndrome etc. congenital infection (TORCH) e.g. congenital rubella syndrome, Toxoplasmosis and maternal diabetes. Similar etiology were found in the studies done by Mosquera et al.<sup>24</sup> and Wiener-Megnazi et al.<sup>26</sup> In this study major birth defects were 78% and minor birth defects were 22%; it was similar to (60.71% vs. 33.29%) the study conducted by Chowdhury K et al.<sup>23</sup> In this cross-sectional study, 27% neonates had multiple birth defects while 73% newborns had only single birth defects, which were similar to (15% vs. 85%) study done by Chowdhury K et al.<sup>23</sup> and (8.3% vs. 91.7%) in study by Jain SR et al.<sup>27</sup> and study by Mosquera et al.<sup>24</sup> showed 37% babies with multiple birth defects and 63% cases with an isolated defect. Accordingly, in this study, 126 birth defects were found among 100 study subjects. The most frequent birth defect in this study was club foot deformity i.e. 11%, similar to 7% in study by Chowdhury K et al.<sup>23</sup> but Yang et al.<sup>18</sup> showed heart defects were the most frequent (13 per 1000). In this study, the birth defects were most commonly found in the cardiovascular system (i.e. 28%) among the system wise distribution of birth defects, followed by musculoskeletal system (24%), genitourinary system (18%) and gastrointestinal system (17%); similar results were found in study conducted by Mosquera

et al.<sup>25</sup> showed the most common birth defect was cardiac defects (75.2 per 10000) and by Jain et al.<sup>27</sup> showed cardiac defects as the most common (29.6%). However, study by Zheng XY et al.<sup>25</sup> showed the most common birth defect was neural tube defects (168 per 10000) and Desai NA et al.<sup>28</sup> found that birth defects were most common in the musculoskeletal system (31.65%). In this study among the study population, mortality was 17%, however 5.67% mortality was found in study by Wazir A et al.<sup>29</sup>

### Conclusion:

The frequency of birth defects in Bangladesh is 1.54%. Club foot deformity was the most frequent congenital anomaly; and the cardiovascular system was the most commonly affected. Some of the study subjects had multiple birth defects while majority neonates had single birth defect and most of them were major birth defects (78%). This study was a small, single center one, so large population based, multi-center studies are required to validate the results further.

### References:

1. Centers for Disease Control and Prevention (CDC) and the National Birth Defects Prevention Network. Key Findings: Updated National Birth Prevalence Estimates for Selected Birth Defects in the United States, 2004–2006. Retrieved October 1, 2014.
2. Centers for Disease Control and Prevention. Update on Overall Prevalence of Major Birth Defects—Atlanta, Georgia, 1978-2005. *MMWR Morb Mortal Wkly Rep.* 2008;57:1-5.
3. Ruth A. Hannon. *Porth pathophysiology: concepts of altered health states* (1st Canadian ed.). Philadelphia, PA: Wolters Kluwer Health/Lippincott Williams & Wilkins. 2010. p. 128.
4. Graham, John Whichello. *Smith's Recognizable Patterns of Human Deformation*, 3rd Edition. Philadelphia: Saunders. 2007. p. 3.
5. Marco DS, Cesari E, Cavaliere A, Ligato MS, Nobili E, Visconti D et al. Paternal exposure and counselling: Experience of a Teratology Information Service. *Reproductive Toxicology.* 2008; 26: 42–6.
6. Sartorius GA, Nieschlag E. Paternal age and reproduction. *Human Reproduction Update.* 2009; 16: 65–79.
7. Cui W, Ma CX, Tang Y, Chang V, Rao PV, Ariet M et al. Sex differences in birth defects: A study of opposite-sex twins. *Birth Defects Research Part A: Clinical and Molecular Teratology.* 2005; 73: 876–80.

8. GBD 2015 Disease and Injury Incidence and Prevalence, Collaborators. Global, regional, and national incidence, prevalence, and years lived with disability for 310 diseases and injuries, 1990-2015: a systematic analysis for the Global Burden of Disease Study 2015. *Lancet*. 2016; 388 (10053): 1545–602.
9. van Gelder MM, van Rooij IA, Miller RK, Zielhuis GA, de Jong-van den Berg LT, Roeleveld N. Teratogenic mechanisms of medical drugs. *Hum Reprod Update*. 2010; 16 (4): 378–94.
10. Ronan O'Rahilly, Fabiola Müller. *Human embryology & teratology*. New York: Wiley-Liss. 2001. ISBN 0-471-38225-6.
11. Zhu JL, Madsen KM, Vestergaard M, Olesen AV, Basso O, Olsen J. Paternal age and congenital malformations. *Human Reproduction*. 2005; 20: 3173–7.
12. Anderson D, Schmid TE, Baumgartner A. Male-mediated developmental toxicity. *Asian Journal of Andrology*. 2014; 16: 81–8.
13. Chia SE, Shi LM. Review of recent epidemiological studies on paternal occupations and birth defects. *Occupational and Environmental Medicine*. 2002; 59: 149–55.
14. Ritz B, Yu F, Fruin S, Chapa G, Shaw G, Harris J. Ambient Air Pollution and Risk of Birth Defects in Southern California (PDF). *American Journal of Epidemiology*. 2002; 155: 17–25. Retrieved 7 December 2014.
15. Aubard Y, Magne I. Carbon monoxide poisoning in pregnancy. *British Journal of Obstetrics and Gynaecology*. 2005; 107: 833–8. Retrieved 7 December 2014.
16. Rushton L. Health hazards and waste management. *British Medical Bulletin*. 2003; 68 :183–97.
17. Lary JM, Paulozzi LJ. Sex differences in the prevalence of human birth defects: A population-based study. *Teratology*. 2001; 64: 237–51.
18. Yang Q, Wen SW, Leader A, Chen XK, Lipson J, Walker M. Paternal age and birth defects: how strong is the association? *Human Reproduction*. 2006; 22: 696–701.
19. Savitz DA, Schwingl PJ, Keels MA. Influence of paternal age, smoking, and alcohol consumption on congenital anomalies. *Teratology*. 1991; 44: 429–40.
20. De Graaf JP, Steegers EAP, Bonsel GJ. Inequalities in perinatal and maternal health. *Current Opinion in Obstetrics and Gynecology*. 2013; 25: 98–108.
21. Vos AA, Posthumus.Anke G, Bonsel GJ, Steegers EP, Denkta<sup>o</sup> S. Deprived neighborhoods and adverse perinatal outcome: a systematic review and meta-analysis. *Acta Obstetriciaet Gynecologica Scandinavica*. 2014; 93: 727–40.
22. Banu T, Chowdhury T K, Das S K, Chowdhury M Z, Hoque M M, Rahman MAM. Birth Defects: A Hospital Based Study in Chittagong, Bangladesh. *Chattagram Maa-O-Shishu Hospital Medical College Journal*. 2014; 13: 5-10.
23. Chowdhury K, Mollah MAH, Choudhury AM, Parvin R, Begum M. Pattern and frequency of congenital anomalies among newborn: a hospital based study. *Journal of Bangladesh College of Physicians and Surgeons*. 2013;31: 84-7.
24. Mosquera TC, Riano GI, Rodriguez DC, Fernandez TJ, Moro BC, Rodriguez FA. Prevalence and secular trend of congenital defects in Austria, Spain. The need for clinic-epidemiological surveillance. *GacSanit* 2009; 23: 300-5.
25. Zheng XY, Song XM, Chen G, Ji Y, Wu JL, Lin JM. Epidemiology of birth defects in high prevalence area of China. *Zhonghua li xingbingxuezhazhi*. 2007; 28: 5-9.
26. Wiener-Megnazi Z, Auslender R, Dirnfeld M. Advanced paternal age and reproductive outcome. *Asian Journal of Andrology*. 2011; 14: 69–76.
27. Jain SR, Naik JD, Dhakne BR, Prabhu PM, Kamble SV, Mathurkar MP. Pattern of congenital malformations in newborn: a hospital-based study. *Int. J Res Med Sci*. 2016; 4: 524-8.
28. Desai NA, Desai A. Congenital anomalies: A prospective study. *Bombay Hospital J*. 2006; 48: 442-5.
29. Wazir A, Dey D, Farid R. Prevalence and Pattern of Congenital Anomalies and its Outcome at Chattagram Maa-O-Shishu General Hospital. *Chattagram Maa-O-Shishu Hospital Medical College Journal*. 2017; 16: 22-5.