



## Prevalence and Pattern of Birth Defects among Pediatric Surgical Patients: A 12-Year Analysis in a Tertiary-level Government Hospital in Bangladesh

Tanvir Kabir Chowdhury<sup>1\*</sup>, S. M. Humayun Kabir<sup>2</sup>, Md. Afruzul Alam<sup>1</sup>,  
Md. Tameem Shafayat Chowdhury<sup>1</sup>, Rumana Khan<sup>1</sup>, Tanzil Farhad<sup>1</sup>  
and Md. Abdullah Al Farooq<sup>1</sup>

<sup>1</sup>Department of Pediatric Surgery, Chittagong Medical College and Hospital, Chattogram, Bangladesh.

<sup>2</sup>Chattogram Medical College Hospital, Chattogram, Bangladesh.

### Authors' contributions

*This work was carried out in collaboration among all authors. Author TKC designed the study, managed the literature search, performed the statistical analysis and wrote the first draft of the manuscript. Authors MAA, MTSC, RK and TF complied data. Authors SMHK and MAAF reviewed and approved the final draft. All authors read and approved the final manuscript.*

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### ABSTRACT

**Aims:** Birth defects are structural or functional abnormalities which occur during the intrauterine life but can be identified at birth or a later age. Birth defects are major public health concerns and the World Health organization (WHO) is supporting countries for earlier diagnosis and better treatment of these conditions. The aim of the study was to assess the hospital-based prevalence and types of birth defects among the surgical children from a part of the world where these are underreported.

**Materials and Methods:** We retrospectively analyzed patients of birth defects admitted in the Department of Pediatric Surgery, Chattogram Medical College Hospital, Bangladesh for a period of 12 years (2008-2019). Data were extracted from annual admission reports, annual audits, mortality and morbidity audits, death registers and computerize data base. Yearly distribution of birth defects, type of defects and their systemic distribution and mortality were analyzed.

\*Corresponding author: E-mail: [ivan\\_tanvir@yahoo.com](mailto:ivan_tanvir@yahoo.com);

**Results:** Among a total of 30,301 admitted patients 30.72% (9307 patients) had birth defects. The most common system involved with birth defects was gastrointestinal (GI) system, 30.61% followed by genitourinary (GU), 29.15% system. Congenital inguinal hernia (17.50%) was the most common disease, followed by hypospadias (14.54%) and anorectal malformation (ARM); 13.98%. Mortality from birth defects were 53.25% of all mortalities. ARM was the most common cause of death (23%), followed by gastroschisis (16%) and intestinal atresia (11%). Highest mortality rates were in tracheo-esophageal fistula/esophageal atresia (TEF/EA)- 83.33%, gastroschisis-80.92%; intestinal atresia-42.53%; omphalocele-32.48%; and congenital diaphragmatic hernia (CDH)- 27.78%.

**Conclusion:** Pattern of birth defects and outcome varied from other countries. The prevalence is 31% among pediatric surgical admissions. Some conditions have extremely high mortality rates and need better management.

*Keywords: Bangladesh; birth defects; children; congenital anomalies; mortality; prevalence; surgery.*

## ABBREVIATIONS

*GI* : Gastro-intestinal  
*GU* : Genitourinary  
*ARM* : Anorectal malformation  
*TEF/EA*: Tracheo-esophageal fistula/Esophageal atresia  
*CDH* : Congenital diaphragmatic hernia  
*WHO* : World Health Organization  
*PUV* : Posterior urethral valves  
*PUJO* : Pelvi-ureteric junction obstruction  
*VID* : Vitello intestinal duct  
*UDT* : Undescended testis  
*VUJO* : Vesico-ureteric junction obstruction  
*CHD* : Congenital heart disease  
*CNS* : Central nervous system  
*LMICs* : Low-middle income countries  
*HICs* : High income countries  
*NICU* : Neonatal intensive care unit  
*TPN* : Total parenteral nutrition

congenital anomaly registries, birth defects commonly involved the following systems (per 10,000 births): congenital heart defects (79.17), limb defects (42.86), urinary anomalies (35.32), nervous system (26.20), Down syndrome (24.34), genital anomalies (21.87), digestive system (18.22), orofacial defects (14.26), and anterior abdominal defects (6.48) [3]. Some commonest defects per 10,000 births are: Hypospadias (18.12), congenital hydronephrosis (13.20), club foot (11.30), neural tube defects (10.08), polydactyly (9.63), cleft lip and palate (8.42), syndactyly (4.20), omphalocele (3.50), ARM (3.31), CDH (2.87), TEF/EA (2.65), and gastroschisis (2.53) [3].

However, the disease burden in South East Asian regions is not properly studied due to a lack of national-level surveillance mechanism [6,7]. To combat this situation, WHO has been observing "World Birth defects Day" every year on 3rd March since 2015. This has now turned into a movement and the first priority of this movement is to increase visibility of birth defects and disseminate what is already known about the prevention and improvement of health and quality of life of the affected individuals [8]. There have been a number of studies and meta-analyses on conditions such as neural tube defects, orofacial clefts, congenital heart diseases, and club foot; but there are few epidemiological studies on GI and GU defects, which constitute a large number disease burden in pediatric surgical wards [9–12]. There is also scarcity of reports from pediatric surgical institutes globally about the surgical aspects of birth defects. As birth defects can present later in life according to WHO definition, it can be assumed that the actual prevalence is more than that was described in literatures. A five-year (2008-2012) analysis of birth defects from this department had been published earlier [13]. The

## 1. INTRODUCTION

Birth defects, also known as congenital anomalies, congenital disorders or congenital malformations, are major causes of mortalities and morbidities in neonates, infants and children. WHO defines birth defect as structural or functional anomalies that occur during intrauterine life that can be identified prenatally, at birth, or sometimes later in infancy [1]. Worldwide, about 8 million babies are born every year with birth defects and among them about 0.3 million neonates die [1]. About 95% of these deaths occur in the low-and middle-income countries. The prevalence of birth defects varies among regions and estimated to be 2.76% in USA, 2.59% in Europe, 6.9% in Eastern Mediterranean and 5.1% in South-East Asian countries [2–5]. According to the European Surveillance of Congenital Anomalies (EUROCAT) network of population-based

aim of the current study was to report a longer-duration (12 years) analysis of prevalence and spectrum of birth defects admitted in a pediatric surgical ward in a tertiary-level government hospital in Bangladesh.

## 2. MATERIALS AND METHODS

### 2.1 Study Design and Setting

This was a hospital based retrospective study carried out in the Department of Pediatric Surgery, Chattogram Medical College Hospital, which is a tertiary-level government academic hospital affiliated with Chittagong Medical College, in Chattogram, Bangladesh. Chattogram (former Chittagong) is the business capital and the second largest city in the country, situated at its south-east part. This hospital is the largest referral center for pediatric surgical services for about one fifth (about 28.4 million) of the population of the country (161.4 million). Patients are referred here from all the southern districts and part of some north-eastern districts. All admitted children of 0 to 12 years of age between January 2008 to December 2019 (12 years) were included in this study. The age limit for admission in this department is 12 years and patients older than that are admitted in adult surgical wards. The department primarily deals with patients of pediatric general surgery and pediatric casualty, pediatric urology, neonatal surgery and pediatric onco-surgery. It also provides outpatient and emergency services to the surgical children. Patients can be admitted round the clock. Pediatric patients with orthopedic, neurosurgical, cardiac surgical, burn and plastic surgical problems can be admitted in the respective adult departments or can be admitted in our department and later referred to them if needed. The hospital has a neonatal intensive care unit (NICU) and pediatric intensive care unit which are run by neonatologists and pediatricians, respectively. The general objective of the study was to evaluate the prevalence and pattern of birth defects in our institute; and the specific objectives were to find out the most common disease burdens, diseases with unsatisfactory outcome in terms of mortality and categorize diseases according to body systems.

### 2.2 Data Collection

Detailed records of any admitted patient are kept in the admission register which is maintained by nursing staffs, and an admission record book is maintained by the resident doctors which is cross checked and signed by a consultant on the

following day. During discharge, the details are recorded in another patient-register maintained by the resident doctors and signed by the respective consultants. It includes patient demography, registration numbers, treatment given and outcome of the patients. Every month these records are summarized and presented as monthly audit by a resident in presence of all the faculties and these records are preserved in Excel files. Every year these records are again compiled in a yearly audit. For the purpose of the study, we fed the period of admission, diagnosis and mortality records of all the patients who were admitted between 2008 and 2019 to an excel sheet. Patients, who had received treatment in the outpatient department or emergency room but were not admitted in the ward, were not included in the study.

### 2.3 Data Analysis

Upon entry on an excel sheet, we alphabetically sorted all the diagnoses and corrected any inaccurate data with the help of the paper records. Then we categorized the diseases with birth defects and without birth defects. The diseases were again categorized according to body system. Frequency and percentage of birth defects were calculated among all patients and among all diseases of respective system. Yearly total of all admitted patients, patients with birth defects, systemic distribution of all diseases and diseases with birth defects were analyzed. Year wise data were compiled and compared along these parameters. Data were evaluated to present the yearly percentage of patient bulk, systemic distribution and disease types. Data were analyzed using both Microsoft Excel 2019 and SPSS version 22 to analyze the prevalence and patterns of birth defects and cross checked to correct errors. Data were expressed as frequency and percentage.

## 3. RESULTS

During this 12-year period, a total of 30,301 patients were admitted in the department of pediatric surgery. Among them, 9307 (30.72%) patients had birth defects. The yearly percentage of birth defects among the yearly admitted patients ranged from 24.68% to 42.37%. Yearly number of admitted patients with birth defects ranged from 691 to 921 (Median 759).

The most common system involved with birth defects was GI system followed by GU system. A large number of patients (2096 patients, 22.52%)

had anterior abdominal wall defects and it was categorized separately. These three systems constituted 82.28% of all diseases with birth defects. Table 1 shows the systemic distribution of birth defects with their contribution among all admitted patients.

Congenital inguinal hernia (1629, 17.50%) was the most common disease, followed by hypospadias (1353, 14.54%) and anorectal malformations (1301, 13.98%). Other common diseases were Hirschsprung disease (823, 8.84%), hemangioma (396, 4.25%), posterior urethral valves (PUV-330, 3.55%), pelvi-ureteric junction obstruction (PUJO-314, 3.37%), undescended testis (296, 3.18%), tongue tie (218, 2.34%), and thalassemia with splenomegaly (186, 2%). These diseases constituted 73.56% of all patients with birth defects. Birth defects of GI (including hepatobiliary) system (2849 patients) were 23.22% of the patients with all diseases of the GI system (both congenital and acquired, 12,270 patients). Anorectal malformation was the most common GI birth defect which was 45.13% of all GI abnormalities. Other common GI conditions were Hirschsprung disease, intestinal atresia (duodenal, jejunoileal and colonic atresia combined), and malrotation of gut with or without volvulus. Table 2 shows a list of birth defects according to body system.

Birth defects of GU system (2713 patients) were 51.38% of all patients of GU system (both congenital and acquired, 5280 patients). Hypospadias (49.87%) was the most common GU defect, followed by posterior urethral valves (PUV), 12.16%; and pelvi-ureteric junction obstruction (PUJO), 11.57%. GU defects shown as others in Table 2 were torsion of penis (9), patent urachus(9), polycystic kidney disease (8), horse shoe kidney (7), ectopic kidney (6), renal

agenesis (5), vaginal tag (4), urethral duplication (4), web penis (4), aphelia (3), vaginal duplication (3), Cobb's collar (2), buried penis (2), primary megaureter (2) and ectopic penis (1).

The most common birth defect of anterior abdominal wall was congenital inguinal hernia (77.72%) and that of vascular/hematological system was hemangioma (50.38%). Tongue tie (33.13%), syndactyly (48.11%), and meningocele (29.29%) were the most common birth defects of orofacial, musculoskeletal and head neck regions, respectively. Orofacial defects, shown as others in Table 2, were macro stoma (4), facial cleft (3), cleft uvula (1), cleft ear lobule (1), deformed ear (1), preauricular sinus (1), duplication of tongue (1), and macroglossia (1). Other skeletal defects were limb deformities (5), spina bifida (5), flat foot (1), and osteogenesis imperfecta (1). Other head neck defects were thyroglossal fistula (5), anencephaly (4), thyroglossal cyst (3), branchial cyst (3), and microcephaly (1).

A total of 657 patients with birth defects died during this period. This was 53.25% of all the deaths during this period and 7.06% of total admission with birth defects. While overall mortality from all diseases during this period was 4.07%; mortality from birth defects was higher (7.06% of all birth defect admissions). Among the mortalities from birth defects, ARM was the most common cause of death (23%), followed by gastroschisis (16%) and intestinal atresia (11%). Fig. 1 shows the most common causes of mortality from birth defects. Mortality rate for ARM was 11.38% of all ARM admissions; and for gastroschisis and intestinal atresia was 80.92% and 42.53%, respectively. Top five diseases with highest mortality rates were TEF/EA- 83.33%; gastroschisis-80.92%; intestinal atresia-42.53%; omphalocele-32.48%; and CDH- 27.78%.

**Table 1. Systemic distributions of birth defects**

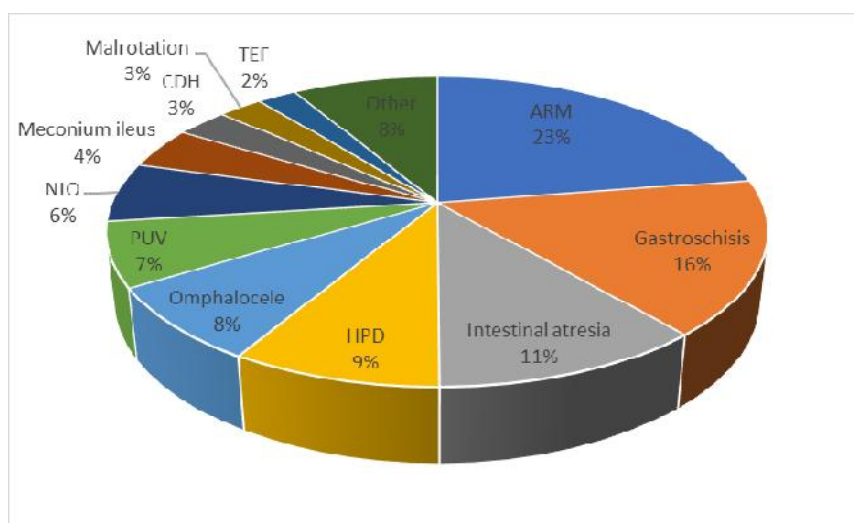
Body Systems	Frequency of birth defects (n=9307)	% of total admissions with birth defects (n=9307)	% of total admissions with all diseases (N=30,301)
Gastrointestinal	2849	30.61%	9.40%
Genitourinary	2713	29.15%	8.95%
Anterior abdominal wall	2096	22.52%	6.92%
Vascular & Hematological	786	8.45%	2.59%
Orofacial	658	7.07%	2.17%
Musculoskeletal	106	1.14%	0.35%
Head & Neck	99	1.06%	0.33%
Total birth defects	9307	100.00%	30.72%

Table 2. List of birth defects

Body system and birth defects	Number of patients	Percent of respective system	Percent of total birth defects (n=9307)
<b>Gastro-intestinal defects (n=2849)</b>			
ARM	1301	45.67%	13.98%
Hirschsprung disease	823	28.89%	8.84%
Intestinal atresia	174	6.11%	1.87%
Malrotation	150	5.27%	1.61%
CDH	72	2.53%	0.77%
Biliary atresia	55	1.93%	0.59%
Choledochal cyst	55	1.93%	0.59%
Patent VID	37	1.30%	0.40%
Meconium ileus	61	2.14%	0.66%
Meconium plug	56	1.97%	0.60%
Eversion of diaphragm	21	0.74%	0.23%
Meckel's diverticulum	21	0.74%	0.23%
TEF/EA	18	0.63%	0.19%
Duplication cyst	2	0.07%	0.02%
Meconium pseudocyst	2	0.07%	0.02%
Antral web	1	0.04%	0.01%
Total GI defects	2849	100.00%	30.61%
<b>Genito-urinary defects (n=2713)</b>			
Hypospadias	1353	49.87%	14.54%
PUV	330	12.16%	3.55%
PUJO	314	11.57%	3.37%
UDT/ectopic testis	296	10.91%	3.18%
Ambiguous genitalia	136	5.01%	1.46%
Ectopia vesicae	62	2.29%	0.67%
Neurogenic bladder	36	1.33%	0.39%
Epispadias	28	1.03%	0.30%
Ureterocele	21	0.77%	0.23%
Ectopic ureter	18	0.66%	0.19%
VUJO	14	0.52%	0.15%
Duplex urinary system	13	0.48%	0.14%
Urogenital sinus anomaly	12	0.44%	0.13%
Micro penis	11	0.41%	0.12%
Other urological defects	69	2.54%	0.74%
Total GU diseases	2713	100.00%	29.15%
<b>Anterior abdominal wall defects (n=2096)</b>			
Inguinal hernia	1629	77.72%	17.50%
Omphalocele	157	7.49%	1.69%
Gastroschisis	131	6.25%	1.41%
Umbilical hernia	73	3.48%	0.78%
Bladder exstrophy	62	2.96%	0.67%
Ventral & other hernias	31	1.48%	0.33%
Cloacal exstrophy	13	0.62%	0.14%
Total anterior abdominal wall defects	2096	100.00%	22.52%
<b>Vascular /Hematological defects (n=786)</b>			
Hemangioma	396	50.38%	4.25%
Thalassemia with splenomegaly	186	23.66%	2.00%
Cystic hygroma	114	14.50%	1.22%
Vascular malformation	64	8.14%	0.69%
Hemophilia	26	3.31%	0.28%

Body system and birth defects	Number of patients	Percent of respective system	Percent of total birth defects (n=9307)
Total vascular defects	786	100.00%	8.45%
<b>Orofacial defects (n=658)</b>			<b>0.00%</b>
Tongue tie	218	33.13%	2.34%
Cleft lip with palate	146	22.19%	1.57%
Cleft lip	140	21.28%	1.50%
Cleft palate	117	17.78%	1.26%
Preauricular skin tag	24	3.65%	0.26%
Other orofacial defects	13	1.98%	0.14%
Total orofacial defects	658	100.00%	7.07%
<b>Musculo-skeletal defects (n=106)</b>			
Syndactyly	49	46.23%	0.53%
Polydactyly	30	28.30%	0.32%
club foot	9	8.49%	0.10%
Amniotic band	6	5.66%	0.06%
Other skeletal defects	10	9.43%	0.11%
Total musculoskeletal defects	106	100.00%	1.14%
<b>Head-neck defects (n=99)</b>			
Meningocele	29	29.29%	0.31%
Myelomeningocele	22	22.22%	0.24%
Branchial fistula	14	14.14%	0.15%
Lipo-meningomyelocele	9	9.09%	0.10%
Hydrocephalus	9	9.09%	0.10%
Other head-neck defects	18	18.18%	0.19%
Total head-neck defects	99	100.00%	1.06%

Abbreviations: ARM: anorectal malformation; CDH: congenital diaphragmatic hernia VID: vitello-intestinal duct; TEF: trachea-esophageal fistula; EA: esophageal atresia, PUV: posterior urethral valves; PUJO: pelvi-ureteric junction obstruction; UDT: undescended testis; VUJO: vesico-ureteric junction obstruction



**Fig. 1. Most common causes of mortality from birth defects**

Percent values are among all mortalities from birth defects (n=657). Abbreviations: ARM: anorectal malformation; HPD: Hirschsprung disease; PUV: posterior urethral valves; NIO: neonatal intestinal obstruction; CDH: congenital diaphragmatic hernia; TEF: tracheoesophageal fistula/esophageal atresia

#### 4. DISCUSSION

This study showed that birth defects constituted about 31% of all pediatric surgical admissions

and 53.25% of all pediatric surgical mortalities. This is a large disease burden and represents patients from surgical ward of only one center. When we consider the undiagnosed cases in the

community, it can be estimated that birth defects are major public health concerns. Worldwide, 3% of the 134 million annual births are associated with major structural abnormalities [7]. To reduce the morbidity and mortality from this huge disease burden, WHO has increased its efforts in prevention and treatment of birth defects over the last five years as evident by the observation of “world birth defect day” since 2015. WHO is also encouraging epidemiologic and basic research in birth defects [8].

The most common system involved with birth defects in this study was GI system followed by GU system. However, the most common disease (congenital inguinal hernia) involved a defect of anterior abdominal wall. Most reports on birth defects, that were studied on new born babies or fetuses, did not report inguinal hernia; because most cases of inguinal hernia usually do not present until later in infancy or childhood. Inguinal hernia in children occurs due to persistence of patent processus vaginalis and is a major cause of morbidity in children worldwide [13,14]. According to WHO, the most common and severe birth defects are heart defects, neural tube defects and Down's syndrome [1]. However, in our institute, these patients are treated in the departments of cardiac surgery, neurosurgery and pediatric wards respectively; and they were not well-presented in this study. Moreover, many cardiac anomalies and some closed neural tube defects, which are compatible with life, are not diagnosed until later age in this country.

The prevalence of birth defects, types of defects and their mortality varied among literatures from both same and different geographical areas. Socio-economic conditions, race, treatment spectrum of health care facilities, support for neonatal care, selection of patient groups, are some of the reasons for these diverse findings. Prenatal data among 18,931 fetuses with birth defects from 52 registered hospitals in Hunan, China found that congenital heart disease (CHD) was the most common defect followed by malformations of kidney, polydactyly and cleft lip and palate. Among these, 9,343 (49.3%) pregnancies were terminated [15]. A study conducted among 13,414 antenatal women, who came for ultrasonography anomaly scan in Maharashtra, found central nervous system (CNS) defects (42%) to be the most common anomaly, followed by urologic malformations (38%) [16]. Another Indian study followed a cohort of 2,107 pregnant women in Pune till outcome and the prevalence of birth defect was

about 351 per 10,000 births with CHD being the most common anomaly [17]. Analysis of 357 pregnant women attending at a fetal medicine service in Brazil found CNS and GU abnormalities most frequently [18]. A study conducted among 6,134 deliveries in an obstetrics and gynecology department in Pondicherry, India found that majority of congenital anomalies involved CNS (28.5%) followed by GI system (20.71%) and musculoskeletal system (20%) [19].

Birth defect surveillance data of perinatal infants from 56 hospitals of Changzho, China reported that the ten leading defects were polydactyly, CHD, syndactyly, microtia, cleft lip and palate, hypospadias, cleft palate, other malformation of external ear, ARM and club foot [20]. A study conducted in neonatal ward among 49 babies with birth defects in BSMMU, Bangladesh found that CVS anomalies (21%) were most common, followed by CNS and musculoskeletal anomalies [21]. A 10-year nationwide population-based cohort study in Taiwan reported prevalence of birth defect to be 271.66 per 10,000 births; CVS anomaly was the most common system involved and ventricular septal defect was the most common disease [22]. Another study used medical insurance benefit data of 403,250 infants, aged less than one year, from the National Health Insurance Corporation from seven metropolitan areas in Korea and found that CVS anomalies were most common followed by GU anomalies (particularly, obstructive uropathy and undescended testis) [7].

On the other hand, A meta-analysis of 25 studies from 9 sub Saharan countries showed that the most prevalent birth defect was musculoskeletal abnormalities while the least was Down's syndrome. The pooled prevalence of birth defects was 20.40 per 1,000 births [23]. Another meta-analysis estimated the national prevalence of birth defects in India from 878 articles (52 hospital based and three community based) and found that anomalies of the musculoskeletal system were highest among live births but CNS defects were highest when stillbirths were included in the analysis [24]. A metanalysis of 42 studies in Iran found urologic disorders to be the most common defects followed by musculoskeletal and cardiovascular malformations [5].

Studies which involved report from pediatric surgical wards are scarce. A meta-analysis of 41 studies from 21 low-middle income countries

(LMICs) for surgically correctable birth defects from 2006 to 2017 found that there is great discrepancy between reported epidemiological data in LMICs and high-income countries' (HIC) literature, partly due to varying quality of data collection in LMICs. They found that orofacial cleft and neural tube defect were the two most frequently studied congenital anomalies, and CDH, intestinal atresia, congenital hydronephrosis, omphalocele, syndactyly, ARM, and bladder exstrophy were among the least frequently studied anomalies [25]. However, studies from Bangladesh were not included in that study.

It has been estimated that birth defects account for 9% of the surgical burden of diseases and are significant causes of morbidity and mortality [26]. About half of the birth defects are correctable with surgical interventions [25]. However, "The Global Burden of Disease study 2013" identified birth defects among the top ten causes of mortality in children less-than-five- years of age [27]. In this study, mortality rate from birth defects was 7.06%. Although, birth defects were 30.72% of all admissions, mortalities from birth defects were 53.24% of all deaths. Mortalities from other diseases were only 2.75%. Some diseases such as, TEF/OA, gastroschisis, intestinal atresia, omphalocele, and CDH had extremely high mortality rates than other centers in the world. These are mostly neonatal surgical emergencies and the dismal outcomes can be attributed to the lack of total parenteral nutrition (TPN), inadequate neonatal care, inexperience in the management of surgical neonates in the neonatal intensive care unit (NICU), lack of trained neonatal anesthesiologist and single lung ventilation, absence of extracorporeal membrane oxygenation, delayed presentation, and deficiencies in surgical skills. WHO, along with the concerned countries, has been taking several actions to improve the situation and child mortality rate fell by 51% over the last three decades. But this decline has been slower in neonates than in older children and one in five neonatal mortalities was attributed to birth defects [28,29]. Establishment of separate neonatal surgical department with proper NICU and TPN facilities might improve the scenario.

This study has several limitations. It has all the inherent limitations of a retrospective study. Since individual patient's files could not be checked, there were scopes for mistakes in entries in the disease diagnosis. Moreover, some diseases or conditions, which were associated

with other abnormalities, were not always recorded in the final diagnoses because there is a tendency to record only the primary problem in the final diagnosis during primary data entry. This is why, some defects might seem to be under-represented in this series, especially associated abnormalities of musculoskeletal and neurosurgical conditions.

## 5. CONCLUSION

Birth defect is a major disease burden for a pediatric surgical facility and a major concern for child health situation. The prevalence was about 31% of all pediatric surgical admissions and mortality rate was 53.25% of all pediatric surgical mortalities. Most common system involved was GI followed by GU system. Most common diseases were inguinal hernia, hypospadias and ARM. Conditions such as, TOF/OA, gastroschisis, intestinal atresia, omphalocele, and CDH had very high mortality rates. Improvement in the outcome of these defects needs to be prioritized.

## CONSENT

It is not applicable.

## ETHICAL APPROVAL

This retrospective study did not involve any patient-contact, exposure of patients' identity or photograph. Permission for publication was taken from the Head of the department and the Head of the institute.

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## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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