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Original Article

Prevalence and Pattern of Birth Defects in a Tertiary Hospital in Lalitpur, Nepal

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ABSTRACT

Introduction: Birth defect is one of the least studied areas in most developing countries like Nepal where low birth weight, prematurity, sepsis, and perinatal asphyxia are still the leading causes of neonatal and infant mortality. However, studies have shown that the incidence of birth defects is increasing trend and has significant impacts on individuals, families, healthcare, and society. Awareness of birth defects is essential in both preventions as well as early intervention. The objective of this study was to find out the pattern and prevalence of birth defects presenting in newborns in KIST Medical College Teaching Hospital, Lalitpur, Nepal.

Materials and Methods: This is a hospital-based cross-sectional, descriptive study conducted in the NICU, Nursery, and postnatal wards of KIST Medical College Teaching Hospital, Lalitpur, Nepal from August 2018 to August 2020. The study population included all inborn newborns with birth defects, stillbirths, and IUFD of more than 22 weeks gestation with birth defects. Outborn babies admitted in Nursery/NICU and abortions of less than 22 weeks gestation were excluded from the study. The data was collected using WHO based surveillance system with a clinical review. Data were analyzed using SPSS version 23. Prevalence and pattern of birth defects were expressed in frequency and percentages.

Results: In a total delivery of 3360, the incidence of the birth defect was 3.75% (n=126). Of the total birth defects, 28.9% (n=112) were born alive and 11.1% (n=14) were stillborn. Among the live births, 2.4% (n=3) died during their hospital stay. The commonly occurring birth defects were cardiovascular (35.7%, n=45) followed by oro-facial defects (23%, n=29), genitourinary (15.9%, n=20), musculoskeletal (15%, n=19), central nervous system (15%, n=19), various syndromes (7.1%, n=9), gastrointestinal (4.7%, n=6) and respiratory (0.8%, n=1).

Conclusions: The true magnitude of birth defects in Nepal is unknown due to the lack of national birth defect surveillance. The lack of proper data regarding birth defects is undermining its impact on perinatal health. Awareness about birth defect and their prevalence can significantly affect the prevention strategy and the management plan in decreasing perinatal mortality and subsequent neonatal and infant mortality as well.

Keywords: Birth defects; Congenital anomalies; Newborns

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INTRODUCTION

A birth defect is the structural and functional anomalies that are present at birth and is broadly categorized as minor and major anomalies.[1] Minor anomalies bear less functional significance except for cosmetic purposes while major anomalies can be life-threatening or have a significant disability.[2] Birth defects remain one of the least focused areas of disease surveillance in

most developing countries like Nepal where sepsis, low birth weight, prematurity, and perinatal asphyxia are still the leading causes of neonatal and infant mortality. However, hospital-based studies published in the recent past have shown that birth defects are emerging as an important cause of perinatal and neonatal mortality.[4] Global estimate of birth defect prevalence with clinical significance is about 2-3%.[5] In 2004, WHO estimated that globally, approximately 7% of all neonatal deaths were caused by birth defects.[6] It is estimated that the prevalence rate of birth defects is 4.7% in developed countries, 5.6% in middle-income countries, and 6.4% in the low-income countries.[7]

Birth defects are the main causes of spontaneous abortion, stillbirth, perinatal death, infant death, and congenital disability, which may have significant impacts on individuals, families, healthcare systems, and society.[8,9] Birth defects are not prioritized in low-income countries as they are considered to be rare and with high mortality of affected infants.[12] Another reason for the under-prioritization of these conditions is the understanding that most birth defects are not preventable through low-cost primary care strategies. The true magnitude of birth defects in Nepal is unknown due to the lack of national birth defect surveillance. The need for data arises as some of these conditions can be prevented through primary care interventions in the preconception period like prenatal folic acid supplementation, vaccination against rubella, and genetic counseling. Also, early diagnosis and intervention can be done in the antenatal period with fetal scans and genetic testing.[10] Thus the awareness about the birth defect and their prevalence can significantly affect the prevention strategy and the management plan in decreasing perinatal mortality and subsequent neonatal and infant mortality as well.

In a meta-analysis done in India in 2018 from 52 hospitals, the prevalence of birth defects in 802,658 births was 184.48 per 10,000 births with anencephaly as the most commonly reported anomaly (21.1 per 10,000 births) followed by talipes (birth prevalence 17.9 per 10,000 births.[13] Both of these defects were either preventable with preconception folic acid supplementation or managed with minimal orthopedic intervention respectively. Another population-based study done in Utah in 2017, including 270878 total births had a prevalence of 2.03%. Spina bifida, hydrocephalus, and clubfoot were the most commonly reported at 26.9%. The definite cause was assigned in 20.2% (n=1114) of cases and 79.8% (n=4390) had unknown etiology.[14] This study clearly shows the gap in knowledge regarding birth defects and the need for further research.

WHO-SEARO started an online system of newborn-birth defects (SEAR-NBBD) database for Nepal in 2014, designed to support data management for newborn health, birth defects, and stillbirths. About 220 hospitals from 9 countries are a part of the NBBD Surveillance network and 170 hospitals from 7 countries are reporting data on birth defects since 2014. The aim was to establish a baseline assessment and monitor the occurrence of birth defects in the region so that appropriate measures can be set.[10]

These studies exclude stillbirths and spontaneous abortions, which shows that the current prevalence may just be the tip of the iceberg. Thus, awareness of the prevalence and types of birth defects is essential in both prevention as well as initiation of early intervention. The present study was undertaken to have the baseline data of birth defects among institutional births in a tertiary care hospital.

MATERIALS AND METHODS

This was a hospital-based cross-sectional, descriptive study conducted in the NICU, Nursery, and postnatal wards of KISTMCTH, Lalitpur, Nepal. The duration of the study was 2 years starting from August 2018 to August 2020. The study population included all inborn newborns during the study period, including stillbirths and IUFD of more than 22 weeks gestation. Outborn babies admitted in Nursery/NICU and abortions of less than 22 weeks gestation were excluded from the study. Non-probability convenience sampling was done using a performed WHO-SEARO-based surveillance system after a clinical review.[10]

Diagnosis of a birth defect was made by thorough clinical evaluation of the newborn by pediatricians and also after relevant investigation reports like echocardiography, ultrasonography, radiographs, karyotyping, etc. The diagnosis was based on the ICD-10.[20] The patterns of congenital anomalies or birth defects along with their systemic distribution were documented. The case reports were reviewed and audited by the maternal and child health review committee every month as well as periodic quality control occurring at the country level by SEARO-NBBD. [10] After the identification of birth defects, parents were informed about the study and maintenance of confidentiality. Informed consent was taken. Ethical approval was obtained from the Institutional Review board.

The demographic profile of all mothers enrolled in the study were age in completed years, parental consanguinity, and parity. Antenatal history included folic acid intake, total ANC visits, history of smoking or alcohol intake during pregnancy, and family history of birth defects in first-degree relatives, birth defects in previous pregnancies, and previous spontaneous abortions. Labor events included the mode of delivery and neonatal data included; gestational age, gender, birth weight, the plurality of birth, and neonatal outcome.

The data was collected using WHO based surveillance system with a clinical review.[10] Each defect was coded with ICD 10 coding system. Data were analyzed using SPSS version 23. Prevalence and pattern of birth defects were expressed in frequency and percentages.

RESULTS

A total of 126 birth defects were identified in a total delivery of 3360 taken between a period of August 2018 to August 2020. Maternal parameters are depicted in table 1. Most of the females were of 20-35 years age group (n=101; 80.2%). The incidence of birth defect thus calculated was 3.75%. Of the total birth defects, 28.9% (n=112) were born alive and 11.1% (n=14) were stillborn. Among the live births, 2.4% (n=3) died during the course of their hospital stay. In the gender distribution, 65% (n=82) were male, 33.3% (n=42) were female and 1.6% (n=2) were of indeterminate sex. In respect of gestational age, 24.6% (n=31) were preterm, 52.4% (n=66) were of term gestation and 23% (n=29) were postdated pregnancies. The majority of babies (56.7%, n=84) had normal birth weight, with 28.6% (n= 36) having low birth weight and 4.8% (n=6) being large for age. The distribution pattern of birth defects was as per shown in graph 1 and table 2.

Table 1: Maternal parameters (n=126)

Parameter			Frequency
Maternal age	< 19 years		18 (14.3%)
	20 – 35 years		101 (80.2%)
	> 35 years		7 (5.6%)
Parity	Primipara		75 (59.5%)
	Multipara		51 (40.5%)
Plurality	Single		122 (96.8%)
	Multiple		4 (3.2%)
Mode of delivery	Vaginal delivery		74 (58.7%)
	Elective LSCS		28 (22.2%)
	Emergency LSCS		24 (19%)
ANC visits	Yes (n=122; 96.8%)	4 visits	88 (72.1%)
		< 4 visits	34 (27.9%)
	No		4 (3.2%)
Folic acid/iron intake	Given		90 (71.4%)
	Not given		22 (17.5%)
	Unknown		14 (11.1%)
consanguinity	Yes		1 (0.8%)
	No		125 (99.2%)
Birth defects in previous pregnancies	Yes		3 (2.4%)
	No		123 (97.6%)
Spontaneous abortions	Yes		11 (8.7%)
	No		115 (91.3%)

Table 2: Distribution of birth defects (n=126)

CNS anomalies: 15%(n=19)
- Spina bifida occulta (1)
- Occipital encephalocele (1)
- Sacral myelomeningocele (1)
- Myeloschisis (1)
- Anencephaly (4)
- Congenital hydrocephalus (4)
- Microcephaly (1)
- Dolicocephaly (1)
- Alobar holoprosencephaly (1)
- Craniosynostosis (1)
- Choroid plexus cyst (2)
- Congenital arachnoid cyst (1)

Oro-facial anomalies: 29(23.01%)
- Cleft palate (3)
- Cleft lip (2)
- Cleft palate and lip (2)
- Preauricular sinus (7)
- Preauricular skin tag (6)
- Ankyloglossia (3)
- Congenital corneal opacity (1)
- Congenital cataract (1)
- Microtia (1)
- Bilateral aniridia (1)
- Congenital hypoplasia of depressor angularis oris (2)

GI anomalies: 6(4.76%)
- Imperforate anus (2)
- Rectovaginal fistula (1)
- Hirschsprung disease (2)
- Tracheoesophageal fistula (1)

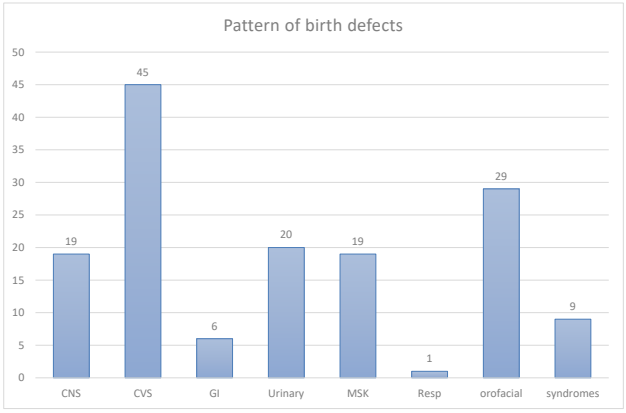
Respiratory anomalies: 1 (0.8%)
- Congenital adenomatous malformation of lungs (1)

Musuloskeletal anomalies: 19(15.07%)
- Talipes equinovarus (7)
- Polydactyly upper limbs (5)
- Polydactyly lower limbs (2)
- Syndactyly (2)
- Hypoplasia of 3rd toe (1)
- Hypoplasia of 2nd to 4th fingers (1)
- Arthogryposis (1)

Syndromes: 9(7.1%)
- Down's syndrome (2)
- Arnold Chiari malformation (1)
- Dandy Walker malformation (2)
- VATER association (1)
- Pierre Robin's sequence (1)
- Hereditary ectodermal dysplasia (anhidrotic) (1)
- Situs inversus (1)

CVS anomalies: 45(35.7%)
- Single umbilical artery (6)
- ASD (21)
- PDA (10)
- Severe Pulmonary stenosis (1)
- Tetralogy of Fallot (1)
- Single atrium (1)
- Severe MR (1)
- Complex cyanotic congenital heart defect (1)
- Hydrops fetalis (2)
- Hemangioma right cheek (1)

Genitourinary anomalies: 20 (15.9%)
- Dilated renal pelvis (2)
- Congenital hydronephrosis (5)
- Bilateral hypoplastic kidneys (1)
- Hypospadias (6)
- Congenital hydrocele (1)
- Cryptorchidism (1)
- Micropenis (2)
- Ambiguous genitalia (2)



Graph 1: Distribution of birth defects. (n=126)

DISCUSSION

The global estimate of birth defect prevalence with clinical significance is 2-3%. [5] The WHO and March of Dimes have reported the incidence of 7% neonatal death and 3.3 million under-five mortality due to congenital anomalies.[10] The prevalence of birth defects in the present study is 3.75% which is higher than the studies conducted in the tertiary center with a higher referral rate for obstetrics cases.[16, 18] These findings may be due to the inclusion of minor defects as well as birth defects of clinical significance. The findings were compared with the meta-analysis done in India in 2018 for a national estimate of birth defect prevalence of 184.48 per 10,000 births.[13] A similar study done by Tomoyuki et al in Japan showed a prevalence of 18.9/1000 births.[19] A cohort study done in 12 hospitals in Nepal in 2021, showed a prevalence of 5.8 per 1000 live births. [21] Different studies done in Nepal showed a prevalence of 8.39% done in Pokhara in 2017(n=1144) including both live and stillbirths[15]andof 1.1% among the live birth in Dhulikhel hospital.[16] This was significantly higher than the previous two studies done in maternity hospitals (0.36%) and Western regional hospitals (0.42%) in Nepal. [17, 18]

The commonly occurring birth defects were cardiovascular (35.7%, n=45) followed by oro-facial defects (23%, n=29), genitourinary (15.9%, n=20), musculoskeletal (15%, n=19), central nervous system (15%, n=19), various syndromes (7.1%, n=9), gastrointestinal(4.7%, n=6) and respiratory (0.8%, n=1). Atrial septal defects, hypospadias, and talipes equinovarus were among the commonest congenital defects. A study done in Pokhara in 2017, showed the prevalence of anomalies related to the central nervous system (12.50%) musculoskeletal (4.16%), genitourinary (12.50%), cardiovascular system (12.50%), oro-facial (20.83%), digestive system (16.66%), syndromes and skin (20.83%) each.[15] Similarly, a study done in Dhulikhel hospital

from 2015 to 2017 showed the prevalence of cardiovascular systems(26.8%) followed by the musculoskeletal system (15.7%) and face (13.8%).[16]In a large study done in 12 hospitals of Nepal in 2021, the commonly occurring birth defects were anencephaly (3.95%), cleft lip (2.77%), cleft lip and palate (6.13%), clubfeet (3.95%), eye abnormalities (3.95%) and meningomyelocele (3.36%).[21] In a meta-analysis and systemic review done in India in 2018, anencephaly was the most commonly reported anomaly with a birth prevalence of 21.1 per 10,000 births followed by talipes with a birth prevalence of 17.9 per 10,000 births.[13] The predominance of cardiovascular defects in our center may be attributed to the availability of pediatric cardiologists and regular screening for high-risk newborns. This is similar to the prevalence shown in Dhulikhel hospital.[16] This also highlights the need for regular echo screening in high-risk newborns for early detection and management of congenital heart disease which generally presents later in infancy. The lower incidence of central nervous system defects in our center can be due to the detection of these anomalies in the antenatal anomaly scans which often leads to medical termination. As our study does not include stillbirths or abortions before 22 weeks of gestation, many such anomalies would have been excluded, which shows that the current prevalence may just be the tip of the iceberg. This is shown by a study done in Biratnagar in 2019, including ante-natal diagnosed congenital anomalies showing involvement of the central nervous system (37%) predominantly anencephaly, and musculoskeletal (13%).[22] This was further clarified in a study comprising 19,244 pregnant women in Hokkaido, Japan from 2003 through 2012 which stated that approximately one-tenth of patients with birth defects delivered between 12 and 21 weeks of gestation and 39% of CNS defects delivered before 22 weeks of gestation. [19]

The true magnitude of birth defects in Nepal is unknown due to the lack of national birth defect surveillance. The lack of proper data regarding birth defects is undermining its impact on perinatal health. Awareness about birth defect and their prevalence can significantly affect the prevention strategy and the management plan in decreasing perinatal mortality and subsequent neonatal and infant mortality as well.

LIMITATIONS

There are several limitations to this study. This study only represents a single tertiary care center and the results may not represent the actual prevalence. A population-based study rather than a hospital-based study is required for the projection of the national birth defect prevalence. The unavailability of advanced diagnostics like genetic studies, karyotyping, metabolic screening, and easy accessibility to echocardiography may also have affected the prevalence. Further, abortions and medical termination due to congenital defects and defects diagnosed during the initial pregnancy were not included in the study which might also have attributed to the lower prevalence. Birth defects may also have been underreported as all congenital defects are not apparent in the early newborn period.

CONCLUSIONS

Congenital birth defects are one of the emerging causes of infant mortality and morbidity. It has a significant impact on individuals, families, healthcare systems, and society as well. The true magnitude of birth defects in Nepal is unknown due to the lack of national birth defect surveillance. Awareness about birth defect and their prevalence can significantly affect the prevention strategy and the management plan in decreasing perinatal mortality and subsequent neonatal and infant mortality as well.

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REFERENCES

1. Rosano A, Botto LD, Botting B, Mastroiacovo P. Infant mortality and congenital anomalies from 1950 to 1994: an international perspective. J Epidemiol Community Health. 2000 Sep;54(9):660-6. Crossref
2. CDC birth defect surveillance Toolkit. Assessed on 2nd October. Available from: Website
3. Slavonev A. Dysmorphology. In: Kliegman, Stanton, St, editors. Nelson Textbook of Pediatrics. Vol 1.1 South Asia Edition. Elsevier; 899-909pp. Crossref
4. Chen et al. Epidemiology of birth defects based on surveillance data from 2011–2015 in Guangxi, China: comparison across five major ethnic groups. BMC Public Health (2018) 18:1008 Crossref
5. Dolk H, Loane M, Garne E. The prevalence of congenital anomalies in Europe. Adv Exp Med Biol. 2010;686:349-64. Crossref
6. The global burden of disease: 2004 Update. In. Edited by organization WH. Geneva; 2008: 1-160. Website
7. Castillo Taucher S. March or dimes global report on birth defects. Revistamedica de Chile. 2007;135(6):806-13. Crossref
8. Kar A. Birth defects in India: magnitude, public health impact and prevention. JKIMSU. 2014;3(2):7-16. Website
9. Christianson A, Howson PC, Modell B. Global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Foundation; 2006. Accessed 13 Oct 2022. Website
10. World Health Organization, Regional Office for South-East Asia. Prevention and control of birth defects in South-East Asia region Strategic framework (2013-2017). New Delhi, 2013. Accessed 13 Oct 2022. Website
11. Park K (2005) Congenital malformations. In: Park K (ed). Park's Text book of Preventive and Social Medicine. (15th edn). BanarsidasBhanot Publishers. pp. 379-80. Crossref
12. Christianson A, Modell B. Medical genetics in developing countries. Annu Rev Genomics Hum Genet. 2004;5:219 -65. Crossref
13. Bhide and Kar A national estimate of the birth prevalence of congenital anomalies in India: systematic review and meta-analysis. BMC Pediatrics (2018) 18:175. Crossref
14. Marcia L Feldkamp, John C Carey, Janice L B Byrne, Sergey Krikov, Lorenzo D Botto Etiology and clinical presentation of birth defects: population based study. BMJ 2017;357:j2249 Crossref
15. Bastola R, Gurung R, Bastola BS, Bastola SS, Bastola L. Pattern and Prevalence of Congenital Birth Defect Among Neonates Admitted to Special Newborn Care Unit (SNCU) Of Pokhara Academy of Health Science (PAHS), Nepal. J Biol Med Res. 2017;2(1):9

16. Dongol SS, Sradanandha S, Shrestha RPB, Bahadur R,Joshi A, Shrestha A, Pattern and Risk Factor Associated with Congenital Anomalies Among Young Infants Admitted in Dhulikhel Hospital. BJHS 2018;3(3)7:548-53. Crossref

17. Malla BK. One year review study of congenital anatomical malformation at birth in Maternity Hospital (Prasutigriha), Thapathali, Kathmandu. Kathmandu Univ Med J (KUMJ). 2007;5(4):557-60. Website

18. Sharma I, Rijal BT, Thapa SB, PoudelI. Congenital anatomical malformaon at birth in Western Regional Hospital, Pokhara, Nepal. Journal of Universal College of Medical Sciences 2013; 1(4):37- 40. Crossref

19. Tomoyuki Hanaoka , Naomi Tamura. Prevalence and Risk of Birth Defects Observed in a Prospective Cohort Study: The Hokkaido Study on Environment and Children's Health. J Epidemiol 2018;28(3):125-32. Crossref

20. International Statistical Classification of Diseases and Related Health Problems. Accessed 13 Oct 2022. Website

21. Paudel P, Sunny AK, Gurung R, Gurung A, Malla H, Rana NB, Kc N, Chaudhary RN, Kc A. Burden and consequence of birth defects in Nepal-evidence from prospective cohort study. BMC Pediatr. 2021;21(1):81. Crossref

22. Yadav M, Subedi S, Prevalence and pattern of Birth Defects in a Tertiary Referral Center, JoNMC. 10:1(2021)20-24. Crossref