Original Article

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Incidence of Birth Defects among Live Born Neonates at Tertiary Level Maternity Hospital in Nepal

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Abstract

Introduction: Birth defects are structural and functional anomalies that present before, at birth, or later in life. This study aimed to find out total incidence, type of structural birth defects in live born babies in Nepal.

Methods: The study was conducted at a tertiary level maternity hospital in Nepal from 14 April 2018 to 13 April 2019. Data was collected on online Newborn Birth Defect data base developed by WHO South- East-Asia Regional Office. All live born babies with external and internal birth defects confirmed by radiographic, ultrasonography and echocardiography until seven days of life were included. Ethical approval was obtained from Institutional Review Committee of hospital.

Results: Total of 21,564 live babies were delivered during one-year study period. Out of these, 220 (1.02%) had one or more birth defects. Number of male babies {130 (59%)} were more than female {89 (40.9%)}. 176 (80%) babies with the malformation were born to mother within age group 20 to 35 years. Of the total 220 babies with birth defects, 197 (89.5%) had isolated malformations and remaining 23 (10.4%) had sequence malformations. The most frequent malformations involved cardiovascular system 125 (56.8%) followed by gastrointestinal system 37 (16.8%), musculoskeletal system 34 (15.4%) and central nervous system 18 (8.1%).

Conclusions: Incidence of overall birth defects in this study was found to be 1.02% in which cardiovascular system anomalies was the most common followed by gastrointestinal, musculoskeletal and central nervous system anomalies.

Introduction

Birth defects refer to structural or functional anomalies that present before or, at birth or later in life.[1] Birth defects can occur during any stage of pregnancy but most occur in the first trimester.[2] These defects can be caused by genetic abnormalities and / or environmental exposures, although the underlying etiology is often unknown. Birth defects can be isolated or present in a characteristic combination or pattern that may affect one or more organ systems.[1]

Structural birth defects are related to a problem with body parts. Range of such defects

includes neural tube defects (NTDs), cleft lip with or without cleft palate, heart defects, and abnormal limbs. Functional birth defects are related to a problem in working of a body part or system. These often lead to developmental disabilities and can include disorders such as: muscular dystrophy, phenylketonuria, loss or impairment of vision and hearing. Structural birth defects are clinically obvious at birth whereas functional defects may only be diagnosed later in life. For example, bleeding disorders like hemophilia is usually not clinically obvious until infancy or childhood. Birth defects not only plays a major role in fetal loss, but also contribute significantly to pre-term births, childhood and adult morbidity.[3] The treatment and rehabilitation of these children with birth defects is very costly with considerable repercussions on their families. Hence the need to identify causative and risk factors to prevent them early, where possible.[3]

Knowing the incidence of birth defects is important for planning and implementing programs to reduce morbidity and mortality resulting from it. Realizing the paucity of incidence of the birth defect studies, this study is being commenced. The aim was to estimate the total incidence, types of structural birth defects in live newborns delivered at the tertiary level maternity hospital of Nepal. This data can be used as baseline to analyze burden of the problem and prevent using existing opportunities.

Methods

The retrospective, institution based study was conducted in tertiary level maternity hospital of Nepal, from 14th April 2018 to 13th April 2019. All the newborn babies delivered in the institution during this period were examined by paediatricians. Thorough physical examination was done to identify any birth defects. All live born babies with external birth defects identified at birth or before discharge from the hospital were included. Those babies with suspected internal defects were sent for radiography, ultrasonography or echocardiography confirmation. Internal defects confirmed within seven days of life were included. Babies who were still-birth and elective termination of pregnancy for fetal anomaly (ETOPFA) were excluded in this study. Data is being collected on online system of World Health Organization- South East Asia Regional Office - New Born Birth Defect (WHO- SEARO-NBBD) data base since 2014. Each case meeting the inclusion criteria were filled in separate performa. The major birth defects were classified according to the International Classification of disease version- 20 (ICD-10) for all birth defects observed by using the Birth Defects atlas.[4] Ethical approval was obtained from Institutional Review Committee of the hospital.

Results

During study period, a total of 21,564 live infants were delivered. Of these newborns, 220 (1.02%) were found to have one or more birth defects. i.e the prevalence of birth defects on this study was 10 per 1000 total live birth.

As shown in table 1, the study showed that there were more male babies (59%) with birth defects than female (40.9%). Among the birth defect newborn babies, the majority (176 ie 80%) of the cases were born to mothers age group 20 to 35 years (Table 2). The incidence of anomaly was found higher among the full term live births than preterm deliveries. Of the total 220 babies with birth defects, 197 (89.5%) had isolated malformations and the remaining 23 (10.4%) had sequence malformations. The most frequent malformation involved was cardiovascular system 125 (56.8%) followed by gastrointestinal system 37 (16.8%) musculoskeletal system 34 (15.4%) and central nervous system 17 (8.1%). Distribution of birth defects in the study group shown in Table 3.

Table 1. Birth defects distributed according to sex and mode of delivery

Total live birth	21564
Total live birth defects	(1%) 220
Sex	
Male	(59%) 130
Female	(40.9%) 90
Mode of delivery	
Normal delivery	(51.3%) 113
Cesarean delivery	(48.1%) 106
Instrumental delivery	(0.45%) 1

Table 2. Frequency of birth defects classified according to maternal age and week of gestation

Maternal age	Number of cases
years 20 >	30 (13.6%)
years 35 - 20	176 (80%)
years 35 <	14 (6.3%)
Week of gestation (WOG)	
< 37 WOG	77 (35%)
> 37 WOG	143 (65%)

Table 3. Birth defects classified according to anatomical system

System	Frequency (220)
Cardiovascular	125 (56.8%)
Central Nervous	18 (8.1%)
Gastrointestinal	37 (16.8%)
Genitourinary	17 (7.7%)
Musculoskeletal	34 (15.4%)
Others	18 (8.1%)

Discussion

The incidence and types of birth defects differ from one country to another and even in same country from one region to another. Globally if we look at it, as per March of Dimes estimates, every

year eight million children worldwide are born with a serious birth defects due to genetic or environmental causes.[5] EUROCAT recorded a total prevalence of major birth defects of 23.9 per 1000, birth for 2003-2007, in which 8% were live birth.[6] The World Health Statistics annual birth data of 2010 reports the estimate would be 9.78 million children.[7]

In Nepal, children born with birth defects annually is 59.9 prevalence per 1000 live births.[8] In our study incidence of birth defects in neonate (1.02%) was found significantly higher than previous studies done at Maternity Hospital (0.36%),[9] Western Regional Hospital (0.42%),[10] Patan Hospital (0.81%),[11] combined 12 hospitals of Nepal (0.58%),[12] but similar to Dhulikhel Hospital (1.1%)[13] and Sarlahi (1.1%).[14] The discrepancy could have been resulted because of increased awareness among hospital staff about birth defects so that they examine the baby to look for anomalies and also for the increased facilities for investigations to detect internal defects. However, while comparing congenital malformation with neighbor countries like India (1.53%), Egypt (2%), Brazil (2.2%), Ethiopia (1.25%), these countries have slightly higher prevalence than our county.[15-18] This may be due to their cohort studies allows the collection of more information which are manifested later in life than document-based retrospective studies.[19]

In the present study, male infants were significantly higher with congenital anomalies than females. This observation agrees with the findings of studies conducted in Maternity Hospital,[9] Western Regional Hospital,[10] Dhulikhel Hospital[13] and at South India.[20] This could be due to X-linked recessive factor or Y-linked genetic basis.[20]

According to several literatures, extremes of ages were found to be associated with birth defects. Young age < 20 years[21] and / or advanced maternal age > 35 years.[22-24] However, results of our studies was not consistent with all of these findings. The incidence of malformation was higher in mother age group 20 to 35 years, which is similar to studies done at Patan Hospital,[11] South India [20] and in Bradford study.[25] This could be because this range of age is the common child bearing age group in our country. Also differences in study design, case definition, and potential confounders may play a role in this discrepancies.[26]

Birth defects encompass a wide array of structural and functional abnormalities that can occur in isolation (i.e. single defect) or as a group of defects (i.e.multiple defects). It is generally estimated that around 14% of babies are born with a single minor defects, around 2 to 3% have a single major defects and 1% of neonates have multiple defects.[6] Those babies with cleft lip and palates, Down's syndrome or babies with meningocele with hydrocephalus are included in isolated defect and with multiple birth defects are included in multiple defects in our study.

The most prevalent birth defect in the current study was cardiovascular system. This may be because of availability of echocardiography facility in the hospital for last one year where all newborn with suspected congenital heart disease (CHD) undergo echocardiography examination early so that they are diagnosed within seven days of life. CHD is most prevalent global trend of birth defect. Since past 15 years, 1.35 million yearly newborns are recorded with CHD. Asia reported the highest CHD birth prevalence, with 9.3 per 1,000 live births.[27] Another multiethnic birth cohort study done in Brandford, UK also reported CHD as the most common anomaly in newborn in UK.[25]

There are several limitations in this study. Since, this is a retrospective study, getting complete information and avoiding biases were challenging. In our setting due to lack of karyotyping and metabolic screening were not able to include chromosomal and metabolic abnormalities which may have lead to under representation of disorders. The sample size is less and only represents one tertiary center.

Conclusions

Incidence of overall birth defects in this study was found to be 1.02% in which cardiovascular system anomalies was the most common followed by gastrointestinal, musculoskeletal and central nervous system anomalies. The incidence and types of congenital malformation can differ from one country to another and even in the same country from one region to another.

References

1. World Health Organization: WHO Facts sheet on Congenital Anomalies. In. Geneva; 2016.
2. Centers for Disease Control and Prevention. https://www.cdc.gov/ncbddd/birthdefects/index.html
3. Grover N: Congenital malformations in Shimla. India J Paediatr. 2000, 67: 249-51. 10.1007/BF02758158. DOI: https://doi.org/10.1007/BF02758158
4. World Health Organization, Centers for Disease Control and Prevention (U.S.) and International Clearinghouse for Birth Defects Monitoring Systems. (2014). Birth defects surveillance: atlas of selected congenital anomalies. World Health Organization. https://apps.who.int/iris/handle/10665/127941
5. March of Dimes. Global report on birth defects: the hidden toll of dying and disabled children. New York, 2006. http:// www.marchofdimes.com/ downloads/ Birth_Defects_Report-PF.pdf - accessed 19 February 2013.
6. EUROCAT [online]. Special report. A review of Environmental Risk factor for congenital Anomalies. 2004 [cited 2009 September 15) Available at: http:// www.eurocat-network.eu2
7. World Health Organization. Global Health Observatory. Available at http:// www.who.int/ gho/child_health/en/index.html - accessed November 2012.
8. Prevention and control of Birth Defects in South-East Asia Region. Strategic framework (2013-2017). https://media.tghn.org/medialibrary/2020/02/
9. 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):557-560.

10. Sharma I, Rijal B, Thapa S, Poudel I. Congenital anatomical malformation at birth in Western Regional Hospital, Pokhara, Nepal. J Univ Coll Med Sci. 2013;1(4):3740. DOI: https://doi.org/10.3126/jucms.v1i4.9572

11. Ansari I, Rajbhandari R, Chilise S, Shalh G, Maskey P, Maharjan R, et al. Congenital malformations at birth in 7,922 consecutive deliveries at Patan Hospital, Nepal. Patan Acad Health Sci , 1(2), 4–7. DOI: https://doi.org/10.3126/jpahs.v1i2.16636

12. Paddle P, Sunny AK, Gurung R, Gurung A, Malla H, Rana NB, et al. Burden and consequence of birth defects in Nepal-evidence from prospective cohort study. BMC Pediatr 21, 81 (2021). DOI: https://doi.org/10.1186/s12887-021-02525-2

13. 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. Birat J. Health Sci 2018;3(3)7: 548-553 DOI: https://doi.org/10.3126/bjhs.v3i3.22173

14. Peto H, Labrique A, Khatry SK, Shahabuddin M, Christian P, Ali H, Klemm R, et al. Implementation of community birth defects surveillance systems in rural Nepal (1994- 1997) and Bangladesh (2001-2007), 2012 (unpublished paper).

15. Gupta S, Gupta P, Soni JS. A study on incidence of various systemic congenital malformations and their association with maternal factors. NJMR. 2010;2(1): 19-21. Corpus ID: 73888336

16. Shawky RM, Sadik DI. Congenital malformations prevalent among Egyptian children and associated risk factors. EJMHG 2011;12(1):69-78. DOI:https://doi.org/10.1016/j.ejmhg.2011.02.016.

17. Porto R, Reis F, Oliveira CC da, de Melo E, Aragão J. Prevalence and Factors Associated at Presence of Central Nervous System Congenital Malformations. JAMR, 2015;6(10):956-64. DOI: https://doi.org/10.9734/BJMMR/2015/14642

18. Taye M, Afework M, Fantaye W, Diro E, Worku A. Magnitude of birth defects in central and northwest Ethiopia from 2010-2014: a descriptive retrospective study. PloS One. 2016;11(10): e0161998. DOI: https://doi.org/10.1371/journal.pone.0161998

19. González AJ, Morales JJ, Luna L, Arteaga J, Mutchinck OM. Beginner's guide to genetics: congenital malformations. BMJ 2004;12(SupplS6):437-80. DOI: https://doi.org/10.1136/sbmj.0412444

20. Neelambari YC, Das P, Sadagopan S, Uma A. Prevalence, pattern and outcome of congenital malformations in a tertiary care centre in South India. IJCP, [S.l.], v. 5, n. 3, p. 1044-1048, apr. 2018. ISSN 2349-3291. DOI : http://dx.doi.org/10.18203/2349-3291.ijcp20181539.

21. Croen LA, Shaw GM. Young maternal age and congenital malformations: a population-based study. Am J Public Health.1995;85(5):710-3. DOI: https://doi.org/10.2105/AJPH.85.5.710

22. Cleary-Goldman J, Malone FD, Vidaver J, Ball RH, Nyberg DA, Comstock CH, et al. Impact of maternal age on obstetric outcome. Obstet Gynecol. 2005 May;105(5 Pt 1):983-90. DOI: https://doi.org/10.1097/01.AOG.0000158118.75532.51

23. Hollier LM, Leveno KJ, Kelly MA, MCIntire DD, Cunningham FG. Maternal age and malformations in singleton births. Obstet Gynecol. 2000;96 (5 Pt 1):701. DOI: https://doi.org/10.1097/00006250-200011000-00011

24. Reefhuis J, Honein MA. Maternal age and non-chromosomal birth defects, Atlanta–1968-2000: teenager or thirty-something, who is at risk? Birth Defects Res A Clin Mol Teratol. 2004 Sep;70(9):572-9. DOI: https://doi.org/10.1002/bdra.20065

25. Sheridan E, Wright J, Small N, Corry CP, Oddie S, Whibley C, et al. Risk factors for congenital anomaly in mulethnic birth cohort: an analysis of the Born in Bradford study. Lancet 2013; 383:1350-59. DOI: https://doi.org/10.1016/S0140-6736(13)61132-0

26. Gill SK, Broussard C, Devine O, Green RF, Rasmussen SA, Reefhuis J, et al. Association between maternal age and birth defects of unknown etiology: United States, 1997-2007. Birth Defects Res A Clin Mol Teratol. 2012 Dec;94(12):1010-8. DOI: https://doi.org/10.1002/bdra.23049

27. Van der Linde D, Konings EE, SlagerMA, Witsenburg M, Helbing WA, Takkenberg JJ, et al. Birth Prevalence of congenital heart disease worldwide: a systemic review and meta- analysis. J Am Coll Cardiol. 2011;59(21):2241-7. DOI: https://doi.org/10.1016/j.jacc.2011.08.025