Research Article

iMedPub Journals
www.imedpub.com

Journal of Biology and Medical Research

2017
Vol.2 No.1:1

Pattern and Prevalence of Congenital Birth Defect Among Neonates Admitted to Special Newborn Care Unit (SNCU) Of Pokhara Academy of Health Science (PAHS), Nepal

Bastola R*, Gurung R, Bastola BS, Bastola SS and Bastola L

Department of Pediatrics, Pokhara Academy of Health Sciences, Pokhara, Nepal

***Corresponding author:** Bastola R

Dr_rc@yahoo.com

Department of Pediatrics, Pokhara Academy of Health Sciences, Pokhara, Nepal.

Tel: +61450922032

Citation: Bastola R, Gurung R, Bastola BS, Bastola SS, Bastola L (2017) 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. Vol.2 No.1:1

Abstract

Objective: To find out the pattern and prevalence of congenital birth defects among neonates in western regional hospital, Nepal.

Methods: This is a prospective hospital-based study conducted over a period of 12 months in the Special newborn care unit of Western Regional Hospital, Pokhara, Nepal from 30th March 2016 to 30th March 2017. Neonates (newborn up to 28 days of age) admitted or examined in the unit irrespective of their condition comprised the study population. They were first examined by the Medical officer at the time of admission and subsequently by a Pediatrician. Information of mode of delivery, gender, weight, gestational age, consanguinity, maternal age, antenatal visit record and family history were recorded on a predesigned Performa. After clinical examination, relevant investigations like ultrasonography, radiology, echocardiography, laboratory and genetic studies were done to confirm diagnosis if it was needed. Statistical data was analyzed by using Excel 2016.

Results: Out of 1144 total admissions, 96 (8.39%) neonates were having birth defect. Of them, 52 (54.10%) were male and 44 (45.90%) females. Anomalies related to the central nervous system were 12 (12.50%) musculoskeletal 4 (4.16%), genitourinary 12 (12.50%), cardiovascular system 12 (12.50%), ear, eye, face, neck 20 (20.83%), digestive system 16 (16.66%), syndromes and skin 20 (20.83%) each.

Conclusion: Congenital birth defects being one of the main cause of neonatal mortality, are very common in our country and most commonly affected system in our study was ear, eye, face and neck anomalies and with syndromes followed by digestive, genitourinary, musculoskeletal, cvs, and skin in descending order of frequency. So, healthcare providers must stress upon primary prevention like vaccination, nutrition and drugs to decrease preventable cause of congenital birth defect.

Keywords: Congenital birth defect; Neonates; Western regional hospital; Nepal; Pattern; Prevalence

Received: November 13, 2017; **Accepted:** November 21, 2017; **Published:** January 03, 2018

Introduction

Congenital birth defect means the defect or malformation that is present at birth. It is an abnormality of physical structure that is seen at birth or within few weeks after birth [1]. According WHO documents 1972, the term congenital birth defect (CBD) should

be confined to structural defects present at birth. CBD can be classified in to major and minor. Minor defects have structural abnormality present at birth with minimal effect on clinical function, but they may have a cosmetic effect e.g. preauricular tag in Goldenhar syndrome. But major malformation results on significant effect of function or on social acceptability e.g. ventricular septal defect (VSD) and cleft lip.

A syndrome is a pattern of abnormalities that occur together and are associated with a set number of signs and symptoms while dysmorphology is the study of abnormalities of human form and its mechanism that cause these abnormalities [2]. Almost 20-30% of infantile mortality and 30% to 50% post-neonatal deaths are due to CBD. First trimester is the crucial period for morphogenesis of organs, especially between the 3rd and 8th weeks of gestation where any insult in any form can cause congenital abnormality. So, this is the period for preventive intervention strategy especially for developing countries where prevalence of CBD is very high. In south east Nigeria prevalence of CBD is higher (29.4%) in low birth weight (LBW) babies than normal birth weight babies (35.3%) and in consanguineous than non-consanguineous marriages in India [3,4].

The cause of CBDs may be either genetic or environmental or sometime even unknown. Among genetic causes, 6% are due to chromosomal abnormality, 25% of single-gene disorders, and 20% of multifactorial and in around 50% cases, the cause is unknown [4]. Folate supplementation during peri-conception period is the most popular and proven preventive measure for neural tube defects [5]. In developed countries of South America food fortification with folic acid significantly reduced incidence of neural tube defects [6]. Consanguinity being the tradition of some communities is very common in Nepal especially among Mongolian origin ethnic groups like Gurung and Rai people. Maternal age is also a risk factor for CBD. Other risk factors include teratogenic drug intake especially from anti-cancer drugs, professional hazard like radiation exposure, maternal illnesses like hypothyroidism, smoking and alcohol consumption [6]. Modern antenatal screening methods like ultrasonography, maternal serum markers, chorionic villus sampling, amniocentesis etc. can be used to detect congenital birth defect which can lead us for manual or therapeutic termination of pregnancy. These days in developed world in utero intervention is gaining popularity for CBDs like hydrocephalus, Posterior Urethral Valves (PUVs), cleft lip and hydro nephrosis [7].

Prevalence of CBD varies between 3% to 7% but depends on geographical, racial and ethnic parts of world [8,9]. Pattern of CBD is also not similar. Malformation related to brain has the highest incidence of CBD i.e. 10/1000 followed by heart 8/1000, kidney 4/1000, limb 1/1000 and miscellaneous 6/1000 live births [10]. Prevalence of CBD is higher in black children than in white. In Nepal, 13% of neonatal deaths are due to congenital anomalies [11]. In Pakistan also, there is high Morbidity and mortality among children with CBD [12].

Preventive measures like supplementation of folic acid, vaccination of mother prior to conception particularly against rubella and chickenpox can contribute to CBD prevention [13] Nepal government has just started to give rubella vaccine in national immunization protocol but so far, no vaccination against chickenpox is introduced. Secondary prevention is especially focused on early antenatal detection using modern methods like USG, maternal serum markers, chorionic villus sampling, Amniocentesis followed by medical or surgical termination of pregnancy, but it involves social, legal and religious issues in Nepal.

We conducted this study to find out the pattern and prevalence of CBD in neonates admitted to our special newborn care unit (SNCU), hoping the information available from this study will help the managers to make preventive strategy to decrease the prevalence of CBD in this western region or Pradesh of Nepal.

Materials and Methods

This was prospective hospital-based study conducted in the special new born care unit (SNCU)of western regional hospital, Nepal from 30th March 2016 to 30th March 2017. All the neonates admitted to the SNCU and delivered either through lower section cesarean section (LSCS) or vaginal in the hospital were included. The study includes gender, gestational age, weight, race, ethnicity and geographical distribution. Thorough neonatal examination and detection of any kind of CBD was done by the medical officer at the time of admission in SNCU or in the Pediatric OPD which than was followed by pediatrician. Neonates were done hematological, radiological and genetic investigations. USG (cranial and abdominal) and CXR (chest X-ray) were done by the radiologist to detect and rule out multiple congenital birth defects, where it was thought necessary. Echocardiography and genetic studies were requested to be done in central hospitals of Kathmandu and standard international lab center SRL. Variables were predesigned according to WHO Performa where gender, weight, gestational age, mode of delivery, consanguinity, maternal age, antenatal visit record and family history were recorded. Post-mortem investigation of neonates was not done due to religious and social factors. Oral consent from the parents for the involvement in the study was taken. The results then were analyzed using Excel 2016.

Results

Overall 1144 neonates were included in the study. Among them 96 (8.39%) were found to have different types of CBDs; 52 (54.10%) were males and 44 (45.83%) females. LBW neonates were 16 (16.66%) and further distribution in weight category revealed that 72 (75.01%) were between 2.5 kg to 4 kg and 8 (8.33%) >4 kg. Regarding gestational age, 8 (8.33%) were preterm, 84 (87.51%) full-term and 4 (4.16%) post-term. Besides this, 24 (24.48%) neonates were born via lower section Caesarean Section (LSCS) and 72 (75.52%) by spontaneous vaginal delivery (SVD) or vacuum assistant vaginal delivery. By doing analysis on maternal age it was found that 16 (16.66%) mothers were above 30 years, 20 (20.83%) 25-30 years, 12 (12.51%) 20-25 years, and 48 (50.00%) below 20 years. Family history of Congenital birth defect was present in 8(8.33%) cases **(Table 1)**. It was found that ear, eye, face, neck related congenital birth defect 19, 19.79%

Table 1 Study parameters (n=96).

Parameter		Frequency	Percentage
Gender			
Male		52	54.10 %
Female		44	45.90 %
Gestational Age			
Pre-Term		8	8.33 %
Term		84	87.51 %
Post-Term		4	4.16 %
Weight			
<2.5 KG		16	16.66 %
2.5-4 KG		72	75.01 %
>4 KG		8	8.33 %
Mode of Delivery			
LSCS (Lower section caesarean section)		24	24.48 %
Others		72	75.52 %
Consanguinity	Yes	8	8.33 %
	No	88	91.66 %
Parity	One	8	58.33 %
	Two or more	88	41.44 %
Family history	Yes	8	8.33 %
	No	88	91.66 %
Maternal age			
>30 years		16	16.66 %
25-30 years		20	20.83 %
20-25 years		12	12.51 %
<20 years		48	50.00 %

were the most common birth defect and syndromes 19, 19.79% followed by birth defect related to digestive system 14, 14.56% genitourinary 12, 12.52%, CNS 10, 10.41% musculoskeletal 9, 9.37% cardiovascular system (CVS) 8, 8.33% and skin 5, 5.21% **(Table 2)** respectively.

Discussion

In our study the prevalence rate of CBD was found to be of 8.39%. The occurrence of CBD depends upon many factors like geographical distribution, nature of sample, cultural influence, and socioeconomic status. Worldwide prevalence is 3% to 7% but varies from country to country [9]. Our prevalence rate is quite like this region like of India (7.5%) but was relatively low compared to 13% reported in Pakistan and it was high that of hospital neonatal unit based study (4.23%) of other countries like Nigeria that has 2.7%, in Taiwan 4.3%, Oman 2.46% and of Bahrain 2.7% [14-17]. In our study most common CBD were ear anomalies followed, in descending order of frequency, by face and neck anomalies and syndromes, digestive, genitourinary, musculoskeletal, CVS, and skin anomalies. Study from Saudi Arabia Asindi et al. [17] stated CNS as the most frequently affected system followed by Musculoskeletal and then Renal. Likewise, a study from Iran Abdi-Rad et al. [18] stated CNS, musculoskeletal, gastrointestinal, urogenital and chromosomal disorders in descending order of frequency. Similarly, Indian studies Gupta et al. [19,20] showed CNS followed by Musculoskeletal and then CVS related birth defect respectively in descending order of prevalence. But some studies from Iran Mosayebi et al. [21-24] and India Gupta et al. [25] showed Musculoskeletal anomalies as the commonest form of birth defects. Similarly, a study from Pakistan Rafi et al. [7,14] reported Gastrointestinal defects as the commonest but others Shamim et al. [13,15] supported CNS findings.

The differences in our study and the other maybe because we were lacking the set-up of sub-specialties like plastic surgery, pediatric surgery, neurosurgery, pediatric urology and

Table 2 Distribution of congenital birth defects (n=96)

System	Birth defect Type	Frequency	Percentage of Total Cases
CNS (Central Nervous System)		**10**	10.41 %
Meningomyelocele+/-hydrocephalus		4	
Microcephaly		1	
Encephalocele		1	
Dandy walker syndrome		2	
Anencephaly		2	
Musculoskeletal system		**9**	9.37 %
Congenital talus equine Varus		4	
Polydactyly		1	
Craniosynostosis		1	
Syndactyly		1	
Spinabifida		2	
Genitourinary system		**12**	12.52 %
Polycystic Kidney		1	
Hydronephrosis		3	
Posterior Urethral Valve		2	
Hypospadias		2	
Congenital Hydrocele		2	
Undescended testes		2	
Cardiovascular system		**8**	8.33 %
Acyanotic		6	
Cyanotic		1	
Complex		1	
Digestive system		**14**	14.58 %
TEF/ Esophageal atresia		3	
Diaphragmatic hernia		2	
Duodenal atresia		4	
Anorectal malformations		3	
Gastroschisis		1	
Exomphalocele		1	
Ear, Eye, Face, Neck		**19**	19.79 %
Abnormal pinna		1	
Buphthalmous		1	
Cleft lip and palate		13	
Micrognathia		4	
Syndromes		**19**	19.79 %
Down syndrome		4	
Arthrogryposis multiplex congenita		1	
Pier Robin syndrome		4	
Goldenhar Syndrome		1	
Skin		5	5.21 %
Preauricular tags		3	
Hemangioma		1	
Giant hairy nevus		1	
Total		96	100 %

neonatology and neonates after birth or in utero are referred from here for especial care to the tertiary care center of Kathmandu, Nepal. Male outstripped female (54.10%male and 45.90% female) in the prevalence CBDs in our study as of the study from Brazil Oliveira et al. [26] (55.5% male and 43.6% in females). It was also supported by the results of other studies, Shamim et al. [13,23,26] but there was one study where female outnumbered male (female 59.1%) [27].

In term babies compared to the preterm and post term ones, the incidence of CBDs was found to be significantly higher in our study. But the study done in Brazil Oliveira et al. [26] shows preterm babies with more incidence of CBD (67% pre-term and 33% term). Another study done in India also shows similar results [26]. The study done in Pakistan, our regional allay supports our study Shamim et al. [13] with the tendency of anomalies more common in term neonates.

It is considered that maternal age is an important factor for the association of CBD. Our study revealed that mothers below 20 years of age had high incidence of producing malformed babies (50.00%). The study done in Pakistan Gul et al. [28] has showed the highest (80.6%) incidence between the age group of 20-40 years but in our study, it was found to be 50.00%.

There was high prevalence of CBD among prime than gravid 2 or more in our study which shows that the early marriages induced pregnancy may have the high prevalence of CBD.

Consanguinity though considered a controversial association with CBDs is very common among Mongolian communities of Nepal. As in our study only 8.33% of CBDs had consanguinity history and it is common in certain community only. Increased prevalence of CBD in consanguineous couples is due to homozygous expression of recessive genes inherited from common ancestors [16]. Family history of CBDs was present in 8.33% of our cases. Similar history of CBD was found in different studies with the prevalence being 8.6%, 9% and 17% reported earlier Taksande et al. [27-30]. There is no strong explanation for these differences in different studies except that cousin marriages are very common in our study as well as in Egypt where 17% was found.

There were some limitations in our study, as it was based in a Special Newborn Care Unit of Western Regional Hospital which is not the tertiary center. Also, our hospital did not have Pediatric cardiology department so there were the chance of missing cases too. Also, we didn't have Pediatric Surgeons for which we did refer many surgical cases for the diagnosis and management purpose. Moreover, the hospital didn't have genetic study facilities.

Conclusion

CBDs are very common in western region of Nepal. Birth defects related to ear, eye, face and neck were the most commonly affected organ in our study. Similarly, different syndromes are also not rare. Term, LBW, male gender, consanguinity, early maternal age and family history of CBDs were associated risk factors for CBDs in neonates. Facts of prevalence and pattern of CBDs are vital to make strategy for preventive measures in both community and center level by the health care managers.

References

1 Hudgins I, Cassidy SB (2008) Congenital malformations among live births at Arvand Hospital Ahwaz, Iran. Pak J Med Sci. 24: 33-37.

2 Genetic Evaluation of Congenital Hearing Loss Expert Panel (2002) Evaluation of infant with single or multiple congenital anomalies. Guide-lines American College of Medical Genetics. PMC 4: 162–171.

3 Obu HA, Chinawa JM, Uleanya ND, Adimora GN, Obi IE (2012) Congenital malformations among newborns admitted in the neonatal unit of a tertiary hospital in Enugu, South East Nigeria: A retrospective study. BMC Res Notes. 5: 177.

4 Rajangam S, Devi R (2007) Consanguinity and chromosomal abnormality in mental retardation and or multiple congenital anomalies. J Anat Soc India 56: 30-33.

5 Castilla EE, Orioli IM, Lopez-camelo JS, Dutra MG (2003) Preliminary data on changes in neural tube defect prevalence rates after folic acid fortification in South America. Am J Med Genet 123: 123-128.

6 Parveen F, Tayyub S (2007) Frequency and pattern of congenital anomalies in newborn and associated maternal risk factors. J Coll Physicians Surg Pak 17: 340-343.

7 Rafi M, Iqbal Z, Saleem M, Waseem M, Anwar J, et al. (2011) Pattern of congenital malformations and their outcome at Sheikh Zayed Medical College/Hospital Rahim Yar Khan. Pak J Med Health Sci 5: 94-96.

8 Kucik JE, Alverson CJ, Gilboa SM, Correa A (2012) Racial/ethnic variations in the prevalence of selected major birth defects, 1994-2005. Public Health Rep 17: 52-61.

9 Park K (2005) Congenital malformations. In: Park K (ed). Park's Text book of Preventive and Social Medicine. (15th edn). Banarsidas Bhanot Publishers. pp. 379-380.

10 Kumar V (2010) Congenital abnormality of brain. In: Kumar V, Abbas A, Fausto N (eds). Robbins and Cotran pathological basis of disease. Seattle Washington: W B Sanders, USA. p. 470.

11 UNICEF/WHO/The World Bank/UN Pop Div. Level and Trends in Child Mortality. Report 2014.

12 Ambe JP, Madzica AG, Akpede GO, Mava Y (2010) Pattern and outcome of congenital malformations in newborn babies in Nigerian teaching hospital. West Afr J Med 29: 24-29.

13 Shamim S, Chohan N, Qmar S (2010) Pattern of congenital malformations and their neonatal outcome. J Surgery 15: 34-7.

14 Gillani S, Kazmi NHS, Najeeb S, Hussain S, Raza A (2011) Frequency of congenital anomalies among newborns admitted in nursery of Ayub teaching hospital Abbottabad, Pakistan. J Ayub Med Coll Abbottabad 23: 117-121.

15 Tayebi N, Yazdani K, Naghshin N (2010) Prevalence of congenital malformations and its correlation with consanguineous marriages. Oman Med J 25: 37-40.

16 Singh A, Gupta RK (2009) Pattern of congenital anomalies in newborn: A hospital based prospective study. J K Science 11: 34-36.

17 Asindi AA, Al Hifzi I, Bassuni WA (1997) Major congenital malformations among Saudi infants admitted to Asir central hospital. Ann Saudi Med 17: 250-253.

18 Abdi-Rad I, Khoshkalam M, Farrokh-Islamlou HR (2008) The prevalence at birth of overt congenital anomalies in Urmia Northwest Iran. Arch Iran Med 11: 148-145.

19 Gupta S, Gupta P, Soni JS (2012) A study on incidence of various systemic congenital malformations and their association with maternal factors. Natl J Med Research 2: 19-21.

20 Parmar A, Rathod SP, Patel SV, Patel SM (2010) A study of congenital anomalies in newborn. Natl J Integr Res Med 1: 13-17.

21 Mosayebi Z, Movahedian AH (2007) Pattern of congenital malformations in consanguineous versus non-consanguineous marriages in Kashan, Islamic Republic of Iran. East Mediterr Health J 13: 868-875.

22 Ali A, Shafikhani Z, Abdulahi M (2008) Congenital malformations among live births at Arvand Hospital Ahwaz, Iran. A prospective study. Pak J Med Sci 24: 33-37.

23 Khatemi F, Mamorri GA (2005) Survey of congenital major malformations in 10,000 newborns. Iranian J Pediatr 5: 315-320.

24 Golalipour MJ, Ahmadpour-Kacho M, Vakili MA (2005) Congenital malformations at a referral hospital in Gorgan, Islamic Republic of Iran. East Mediterr Health J 11: 707-15.

25 Gupta RK, Gupta CR, Singh D (2003) Incidence of congenital malformations of the musculoskeletal system in live newborns in Jammu. JK Science 5: 157-60.

26 Oliveira CI, Costa AR, Ferrarese VCC, Mozvaz DC, Fett-conte AC (2011) Birth defects in newborns and stillborns: An example of the Brazilian reality. BMC Notes 4: 343.

27 Taksande A, Vilhekar K, Chaturvedi P, Jain M (2010) Congenital malformations at birth in central India, A rural medical college hospital based data. Indian J Hum Genet 16: 159-63.

28 Gul F, Jabeen M, Khan AS (2012) Frequency of congenital malformations and associated risk factors at Liaqat Memorial Hospital Kohat. Khyber Med Univ J 4: 119-24.

29 Ahmed AM, Elkader SA, Kader SA, Hamid AA, Gaafar HM (2011) Assessment of risk factors for fetal congenital anomalies among pregnant women at Cairo university hospitals. J Am Sci 7: 899-908.

30 Sridhar K (2009) A community based survey of visible congenital anomalies in rural Tamil Nadu. Indian J Plast Surg 42: 184-191.