Clinical Study on Congenital Birth Defects in a Tertiary Hospital, Western Nepal

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ABSTRACT

Introduction: Congenital Birth Defect (CBD) is one of the commonly encountered problems in neonatal period and major birth defects contribute significantly to neonatal morbidity and mortality. Minor anomalies are frequently seen in newborn which are not detrimental to health but may be associated with major defects serving as a clue for diagnosing more serious underlying defect. Our purpose of study is to determine burden of CBDs, clinical profile and outcome of those neonates born with CBDs admitted in Neonatal Intensive care Unit (NICU) and Obstetric Unit of Devdaha Medical College.

Methods: A hospital based prospective study was performed from 1st Feb 2023 to 30th Sept 2023. All live newborns were screened for birth defect and clinical examination was done every day till discharge. All newborn with CBDs fulfilling the inclusion criteria were taken for study. Maternal characteristics like age, parity, mode of delivery, comorbidities and risk factors were noted. Newborn characteristic like sex, week of gestation, birth weight along with co morbidities (like neonatal sepsis, birth asphyxia) and outcome were noted. Data obtained were coded and entered in SPSS (version 24) for analysis.

Results: The incidence of CBDs was 6.53% with incidence of major defect being 2.5% and minor defect4%. Among all defects, skin anomaly was the commonest defect (30.6%) observed. Among the major defects, musculoskeletal system was the commonest. More than 50% of such babies were born via caesarean section and 1/3rd had low birth weight and nearly 80% were term babies. Among the babies with CBDs, 40% babies had various comorbidities like Neonatal sepsis, birth asphyxia requiring NICU admission and 8% of babies with CBD were referred for surgical correction and 1 (2%) expired.

Conclusion: Congenital malformation is one of the commonly encountered cases (6.53%) among which minor anomaly is more predominant (61.22%) with more than 80% having good neonatal outcome.

Keywords: Congenital Birth Defects, Neonatal Intensive Care Unit, Newborn.

Introduction

Congenital means present at birth and the combination terminologies like congenital anomaly (CA), congenital defect or congenital abnormality imply the structural problem present at birth without specifying the etiology.[1] Congenital birth defects (CBDs), termed by WHO, is defined as structural or functional anomalies that occur during intrauterine life which develop prenatally and may be identified before or at birth, or later in life.[2] Both CA and CBDs can be used interchangeably as they mean the same.[1] Globally 7.9 million babies are born with CBD, out of which more than 3 million die by the age of 5 years and more than 3 million babies live with disability.[3] Of this global data, low and middle income countries carry more than 90% burden because of sharp differences in maternal health, financial constraints, greater percentage of consanguineous marriages and other risk factors.[2]

Congenital anomaly (CA) is classified into major and minor anomalies. Major structural anomalies are those with medical and social consequences bearing the greater percentage of consanguineous marriages and other risk factors.[2]

Congenital anomaly (CA) is classified into major and minor anomalies. Major structural anomalies are those with medical and social consequences bearing the highest incidences among abortions and stillbirths.[1] Prevalence of major CA is 2-3% as shown by various studies.[4,5] Some of these major malformations escape early detection and are found by 5 years of age making the equal number of 2-3%.[6] Minor anomalies are those

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structural alterations without having medical and social burden. Presence of two or more minor anomalies is an indication that a major defect may be present.[1]

Prevalence of CBD depends on geographical, racial and ethnic parts of the world because of underlying genetic susceptibilities, exposure to risk factors, variability in case diagnosis, ascertainment and reporting system.[7] Studies from other countries have shown the prevalence of CA varying between 1 to 4%.[7,8,9] Though true magnitude of birth defects is not known in Nepal, various studies conducted at regional level have found the prevalence lying between 2 to 9%.[10,11,12] Verbal autopsy findings of newborn deaths in the 2006 Nepal Demographic and Health Survey showed that 8% of neonatal death is due to congenital anomalies.[13]

Pattern of CBD is not similar as shown by various studies. According to March of Dime report, three common serious birth defects are Congenital heart defects, Neural tube defects and hemoglobin disorders.[3] Other studies in India and Nepal shown Musculoskeletal and CNS anomalies were the commonest defects respectively.[10,14]

Though in majority of CA babies, causes are unknown but factors like genetic, environment and teratogens like maternal alcohol use, diabetes, infections, drug use and exposure to radioactive substances are shown to contribute to CA.[15] Some congenital disorders can be prevented and anomalies like Neural Tube Defects (NTDs), clefts, congenital heart diseases can be addressed by surgical care.[2,16] Thus prenatal diagnosis with initial surgical treatment has significant influence on outcome imposing the need of treatment to be carried out in centers with expertise and multidisciplinary team.[17]

Methods

This was a hospital based prospective study carried out in Newborn Intensive Care Unit (NICU) and Obstetric ward of Devdaha Medical College from 1st February 2023 to 30th September 2023. Ethical clearance was obtained from Institutional Review Committee (IRC) (Approval number 05/2023).Written informed consent was taken from the parents of the participants who were included in the study. As per WHO definition, any anatomical or functional abnormality detected after birth is defined as Congenital birth defect.[2] All live newborns were screened for CA at birth and systematic examination was done every day till discharge. CBDs were classified system wise. Major CBDs are those with significant effect on function requiring medical attention (eg Congenital heart disease, club foot) and minor CBDs are those with minimal effect on function but having cosmetic effect (hemangioma, nevus).[18] Out born babies, stillbirth and parents who did not consent for the study were excluded. A preformed proforma was developed and detailed characteristics of newborn with CBD (like birth weight, sex, gestational age) and maternal details (like age, parity, consanguinity) were noted. For the specific CBD, relevant investigations likechest X ray, Echocardiography and abdominal ultrasound were done. Outcome of each CBD was noted in terms of discharge, expiry, referral and leave against medical advice (LAMA). Echocardiography and abdominal ultrasound were done. Outcome of each CBD was noted in terms of discharge, expiry, referral and leave against medical advice (LAMA).

Live born babies with birth defects were the variable of interest of our study. Low birth weight (LBW) was defined as birth weight less than 2500gm. Birth Asphyxia was defined as APGAR score less than 7 at 1 minute. Neonatal sepsis was defined as clinical sign of sepsis with or without supportive laboratory values. Respiratory distress syndrome (RDS) is defined as respiratory distress evidenced by tachypnea, grunting and cyanosis requiring any form of respiratory support seen in preterm babies born before 37 completed weeks of gestation.

Sample size was calculated using $n=Z^2xpq/d^2$ where Z=z score for 95% confidence interval (1.96) p=prevalence, d=acceptance error. Taking prevalence of CBD as 2%[19] with 95% confidence interval and standard error of 4% the total number of newborn with birth defect would be 48. We screened total 759 of live newborn for birth defects during the study period. Data was entered into SPSS version 26.0 and frequency tables and charts were created.

Results

In our study period, total 759 newborn were delivered out of which 3 were stillbirth and 6 were intrauterine fetal death (IUFD) making a total of 750 live newborn. Thus we screened total 750 live newborn and found that 49 babies were born with CBD making an incidence of 6.53% (49/750). The incidence of babies born with major defect was 2.5% (19/750) and that of minor defect was 4% (30/744). Male and female ratio was almost equal to 0.9.

Table 1 shows the characteristics of newborn with CBD. Out of 49 babies, 16 (32.6%) had low birth weight (weight less than 2.5kg), most were term babies (75.51%) with mean birth weight of 2.73±0.5 kg. Regarding the mode of delivery, 22 (44.89%) had

normal vaginal delivery, 26 (53.06%) underwent caesarean section and one (2.04%) had instrumental delivery.

Table 2 showed the characteristics of mother who delivered babies with CBDs. Mothers who delivered the babies with CBD were of 19-38 years with mean age being 26.41±4.32 years and among them 28 (57.1%) were primiparous.

Table 3 depicts distribution of CA on the basis of system involvement. Out of 49 neonates majority had single system defect (93.8%) whereas only 3(6.1%) had more than one system defect. Among the total CA cases, minor defect carries the most burden of 61.22%. Among the major CBDs, musculoskeletal (MSK) anomalies were the most common one (47.33%). Among the single

Table 1 Neonatal characteristics (n=49)

Variables		No.	%
Sex	Male	24	48.98
	Female	25	51.02
Birth weight (kg)	1.5-2.49	16	32.65
	>2.5	33	67.35
Gestation	Term	38	77.55
	Preterm	11	22.45
Comorbidities	RDS	2	4.08
	Birth asphyxia	2	4.08
	Seizure	2	4.08
	Systemic Sepsis	13	26.53
Interventions required	Respiratory support	36	12.24
	Antibiotics	19	38.78

Table 2 Maternal characteristics of babies with CBD (n=49)

Variables	Variables	NO.	%
Maternal age	<19 years	1	2.04
	20-35 years	47	95.92
	>35 years	1	2.04
Parity	Primipara	28	57.15
	Multipara	21	42.85
Pleurality	Single	47	95.92
	Multiple	2	4.08
Mode of delivery	Vaginal delivery	22	44.90
	LSCS	26	53.06
	Instrumentation	1	2.04
Risk Factors	Adequate ANC visits	48	97.94
	Folic acid intake in first trimester	34	69.38
	Not sure of intake of folic acid	7	14.28
	Iron intake in second and third trimester	43	87.75
	Consanguinity in parents	1	2.04
	Birth defect in previous pregnancies	1	2.04
	Spontaneous abortion	6	12.24
	Eclampsia	1	2.04
	Meconium stained liquor	7	14.29
	Antepartum hemorrhage	1	2.04
	Previous termination of pregnancy for birth defect	1	2.04

Figure 1: Outcome of neonates having birth defects

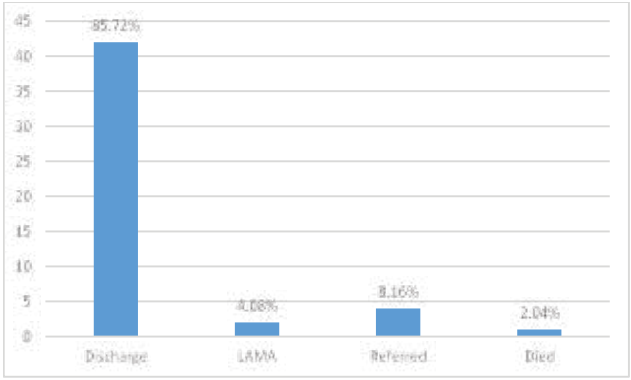

Table3. Distribution of congenital birth defects (n=49)

SINGLE DEFECT	
Haemangioma	6
Non melanocytic nevi	5
Accessory nipple	2
Skin tag right hand	1
Port wine stain	1
B. **Craniofacial abnormality (n=12)**	
Preauricular sinus	3
Low set ear	2
Accessory auricle	1
Abnormally shaped ear	1
Preauricular skin tag with eyelid coloboma	1
Helical skin tag	1
Excessive growth of antihelix	1
Pterygium colli	1
Right helix Pit	1
C. **Musculoskeletal System (n=10)**	
Polydactyly	3
Bilateral club foot	1
Congenital Left Patella dislocation	1
Left Genu recurvatum with patella dislocation	1
Left Talipuscalcaneovarus	1
Pes Planus With Talipus Valgus Right Foot	1
Supernumerary Fingers	1
Sacral Sinus	1
D. **Genitourinary System (n=7)**	
Paraphimosis	2
Congenital hydrocele	1
Hooded prepuce	1
Cryptorchidism with micropenis	1
Mucosal lining between rectum and vagina	1
Hypospadias	1
E. Circulatory System (n=6)	
Complex congenital heart disease	2
Atrial Septal defect (ASD)	1

Ventricular Septal Defect (VSD)	1
Hydrops fetalis	1
Single umbilical artery	1
F. **Gastrointestinal System (n=3)**	
Hirschsprung disease	1
Mesenteric cyst	1
Tracheosophageal fistula	1
MULTIPLE DEFECTS	
Craniofacial+ Skin	2
MSK +Skin	2

defects, anomaly involving skin was the most common 15(30.6%) fo llowed by craniofacial 12 (24.48%) and MSK system 10 (20.4%) respectively.

Figure 1 shows the outcome of neonates having birth defects. Out of 49 babies with birth defects, 20 (40.8%) required NICU admission. Regarding outcome, 41 (83.67%) babies were discharged, 01(2%) case of hydrops fetalis required resuscitation and ventilator support died within 24 hour and 4 (8.16%) cases were referred which included all cases of GI system and one MSK system as they required surgical intervention.

Discussion

The incidence of overall birth defects (including major and minor) in our study was 6.5% which is much higher than incidence of 2-3% reported from Europe and Eastern India.[19,20] This incidence is even much higher than the incidence reported from Nepal (0.58%).[10] This discrepancy is explained by the fact that those studies were multi centered with large number of samples and included stillbirth and IUFD as well. Our incidence is similar to study done in Pokhara and Nigeria as these studies are single centered and share nearly similar study population size.[12,21] We observed the incidence of major CBDs to be 2.5% which is similar to other studies.[19,22]

Regarding the pattern of CBDs, we observed sharp difference from other studies. Among CBDs, skin anomaly was the most common (30.6%) followed by craniofacial anomalies (24.48%) which is in contrast to other studies.[11,14,20,21,22] It could be because in our study among the total CBDs, minor anomalies carried the major percentage. Paudel et al classified 76% of total CBDs as 'Others' for those other than commonly seen defects which might include skin and other minor anomalies.[10] Interestingly we did not observe any case of central nervous system (CNS) defect which is in contrast to other studies where CNS anomaly was one of the frequently occurring birth defects.[10,11,15] This could be either babies with CNS defect were not born during such short study period or 10 cases of stillbirth/IUFD were not included in our study as CNS anomaly is commonly associated with stillbirth or abortion.[23] This is further clarified by the study done in Japan during 10 year study period on more than 19000 pregnant mother with the result that 2 out of 5 CNS birth defects were delivered before 22 week of gestation.[24] In our study among the major CBDs, MSK defect was the commonest. This similar pattern was observed by other Indian studies as well.[14,25]

We did not observe any significant association between maternal age and CBDs because more than 90% of babies were born to mother 20-35 years of age with mean age of 26 years which represented the usual birth cohort of pregnant mother of our hospital. Study from Western region of Nepal showed that 50% of babies with CBDs were born to mother age less than 20 years ofmaternal age and CBDs because more than 90% of babies were born to mother 20-35 years of age with mean age of 26 years which represented the usual birth cohort of pregnant mother of our hospital. Study from Western region of Nepal showed that 50% of babies with CBDs were born to mother age less than 20 years of age.[12]A multi centered study from Nepal showed mothers less than 20 years of age are 1.64 times likely to give birth to babies with CBDs.[10] Our study showed more than 2/3rd babies with CBDs were born at term which is in contrast to a Brazilian study in which nearly 86% babies with CBDs were preterm. However other studies done in Pakistan and other region of Nepal supported our result.[12,27]

We did not find any gender preponderance for CBDs as shown by many studies however we observed that female outnumbered male neonates in CBDs involving cardio-vascular system (5 out 6 babies were female) and male predominance in CBDs involving genitourinary system (6 out of 7 babies were male) although we need larger sample to support this result.

It is an established fact that any case of CBD is significantly associated with various perinatal outcomes like higher rates of caesarean deliveries, perinatal asphyxia and NICU admission for various reasons.28In our study, more than half of babies with CBDs were born via caesarean/instrumental delivery and nearly 40% babies with CBDs needed NICU admission for various other comorbidities like neonatal sepsis, perinatal asphyxia requiring treatment as shown in Table1.This shows the need for early identification of such defects and efforts must be made to deliver such babies in center with facility. This finding is supported by another multi centered study from Nepal.[10]

The risk factors for CBDs are not new and are known for decades. We admit that for the correlation of the maternal risk factors with birth defects control group was not taken into account so it would be inappropriate to establish association between the two however maternal risk factors we observed were history of spontaneous abortion (12.24%), no intake of folic acid (16.3%), meconium stained liquor (14.29%) and others as shown in Table 2. These risk factors were identified by other studies as well.[10,14]According to WHO report 2019, 30% of neonatal death is attributed to congenital anomalies and this percentage has increased over the years because overall neonatal mortality rate has declined as other causes like neonatal sepsis and birth asphyxia are taken care of.[2] Other studies from Nepal and India also showed mortality rate varying from 20 to 30%.[10,14] However we observed mortality due to CBDs of only 2% which is explained by the fact that we had major percentage of minor anomalies and neonates with major anomalies involving GI system were referred for surgical correction so immediate outcome could not be assessed.

Limitations

There were several limitations to this study. As compared to other studies our sample size was small. As this study was done in tertiary Hospital result cannot be extrapolated to general population. Being a resource limited setting, genetic work up could not be done. Socio demographic and risk factors were not studied in detail as control group was not taken into account. Stillbirth and abortion were excluded which affected the percentage of incidence. Those defects requiring surgical correction were referred affecting the outcome result.

Conclusion

The incidence of major birth defect is 2.5% and overall defect is 6.5%. All newborns should be screened for congenital anomaly and various comorbidities are associated with such defects requiring the need for proper antenatal screening and easy access for high facility tertiary center for delivery of such babies.

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