https://doi.org/10.22502/jlmc.v8i1.317 **Original Research Article**

"Overcoming the Defect": Congenital Anomalies in 6984 Consecutive Deliveries in a Tertiary Care Center in Nepal

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ABSTRACT

Introduction: Neonatal congenital anomalies are the 17th leading cause of global burden of disease. Congenital anomalies in low-income countries are high due to prevalence of nutritional deficiencies, intrauterine infections, teratogenic exposure and unsupervised self-medication. This study aimed to find various antenatal risk factors for birth defects and the delays in health seeking behaviors in cases of still births with birth defects. **Methods:** The records of mothers and neonates born with congenital anomalies during the period of 30 months were reviewed. Consanguinity, intrauterine infections, presence of anemia, and history of drug intake were noted. In cases of stillborn with anomalies; mode of delivery, labor complications and the delays leading to morbidities were also noted. **Results:** There were 65 congenital anomalies among 6984 consecutive deliveries during the study period with an incidence of 9.3 per 1000 births. Out of all diagnosed anomalies, 43 were seen in live births and 22 in still births. The common anomaly noted amongst live births was of musculoskeletal system (n=11, 25.5%) whereas defects in nervous system was common in the still born fetus (n=10, 45.4%). A notable finding in the present study was consanguinity which was present in 30 parents (46.1%) whose newborn had a congenital defect. Folic acid supplementation was taken by only 26 mothers (40%) in the present study. In cases of still births with anomalies, most deliveries were vaginal (97%) without any maternal complications. Delay in deciding to seek care was observed in most cases (n=14, 63.6%) in the present study. **Conclusion:** Non consanguineous marriage and folic acid supplementation is useful in preventing congenital anomalies. Encouraging early antenatal visits might help in early detection of anomalies.

Keywords: Birth defect, Folic acid, Risk factors, Still birth

INTRODUCTION:

Neonatal congenital anomalies are the 17th leading cause of global disease burden, and a traumatizing experience to a mother and an overwhelming encumber to the family.[1] Congenital anomalies in low-income countries are high due to prevalence of nutritional deficiencies, intrauterine infections, exposure to teratogen and unsupervised self-medications.[2]Recognizing the causative factors for still births and congenital anomalies play an important role in preventing these conditions. Identifying both clinical and socio-demographic factors and further educating on preconceptional and prenatal risks will help in reducing the rate of still births and birth defects. This study was aimed to find various antenatal risk factors for birth defects and to evaluate the delays in health seeking behaviors in cases of still births with birth defects.

METHODS:

This was a retrospective study carried outin the department of Obstetrics and Gynecology of Lumbini Medical College and Teaching Hospital,

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Nepal over a period of two months from February to March 2020. During this period,the records of neonates born with congenital anomalies during the period of thirty months (June 2016 to December 2019) were reviewed.

Neonatal records of all babies with congenital anomalies were studied and antenatal history reviewed. Consanguinity, intrauterine infections, presence of anemia, history of drug intake, alcohol consumption and smoking during pregnancy were noted. In cases of stillborns with anomalies, mode of delivery, labor complications and the three delays leading to morbidities namely, delay in seeking care, reaching care and receiving care were also noted. Still births were taken as birth of a baby with no signs of life after 22 weeks of gestation or weighing ≥500 grams, which included both antepartum and intrapartum still births. The present study included the congenital anomalies where the diagnosis was made by a pediatrician after thorough clinical evaluation. Different types of birth defects classified according to their International Classification of Diseases (ICD-10).

All the collected socio-demographic and clinical information were recorded in a structured proforma. The data were then entered and analyzed using Statistical Package for Social Sciences (SPSS™) software version 20. Results were expressed as frequencies, percentages, mean, and standard deviations.This study was approved from Institutional Review Committee (IRC-LMC 03-J/019) of Lumbini Medical College teaching Hospital.

RESULTS:

During the study period, there were 6984 total deliveries out of which 65 babies were born with congenital anomalies. The incidence was 9.3 per 1000 births. Twenty-two babies (33.85%) were still births and the remaining 43 (66.15%) were born alive. The mean age of the mothers was 24.33 ± 4.31 years.

Table 1 shows the different types of birth defects classified according to their International Classification of Diseases (ICD 10) codes. The most common birth defect was of the nervous system (24%) followed by musculoskeletal system (17%). Anencephaly and talipes equinovarus were the most frequent anomalies in these systems. The most common anomaly in still births was that of thenervous system (n=10, 45.4%) and in live births wasthat of the musculoskeletal system (n=11,25.5%). Birth defects were more common in female babies (n=34, 52.3%) than in males (n=31, 47.7%).

Out of all the babies born with birth defects, 39 (60%) were term pregnancies, 18(27.7%) were preterm and the rest were post term i.e. born after 42 weeks.

Antenatal risk factors for birth defects are shown in Table 2. Consanguinity was present in 30 (46.1%) of the cases and folic acid wastaken by 26(60%) mothers only.

Delivery was by vaginal route in 21 (94.5%) cases of still births with birth defects. Delay in recognizing need of care was the most common

Table 1. Type of birth defects in live and still births (N=65).

Type of birth defect	ICD codes	Live birth n=43	Still birth n=22	Total N (%)
Congenital malformations of the nervous system	Q00-Q07	9	10	19 (29.2)
Congenital malformations of eye, ear, face and neck	Q10-Q18	4	1	5 (7.6)
Congenital malformations of the circulatory system	Q20-Q28	6	1	7 (10.7)
Congenital malformations of the respiratory system	Q30-Q34	3	1	4 (6.1)
Cleft lip and palate	Q35-Q37	4	1	5(7.6)
Other congenital malformations of the digestive system	Q38-Q45	3	0	3(4.6)
Congenital malformations of the urinary system	Q60-Q64	1	0	1 (1.5)
Congenital malformations and deformations of the musculoskeletal system	Q65-Q79	11	6	17 (26.1)
Other congenital malformations	Q80-Q89	1	1	2(3.0)
Chromosomal abnormalities, not elsewhere classified	Q90-Q99	1	1	2(3.0)

Table 2. Antenatal risk factors in birth defect cases.

Risk factors	Live births (N=43) n (%)	Still births (N=22) n (%)	Total (N=65) n(%)
Antenatal visits ≤4	22(51.5)	9 (40.9)	31 (52.3)
Consanguinity	19(44.2)	11 (50)	30 (46.1)
Folic acid not taken	28(65.1)	11 (50)	39 (60.0)
Alcohol during pregnancy	4(9.3)	3(13.6)	7 (10.7)
Maternal smoking during pregnancy	1 (2.3)	3(13.6)	4 (6.1)
Intrauterine infections	1 (2.3)	2(9.1)	3 (4.6)
Anemia	5(11.6)	8(36.4)	13 (20.0)
History of drug intake	2(4.7)	0 (0.0)	2 (3.0)

delay seen in 63.6% women (Table 3). The causes for delayed care seeking were no knowledge of danger signs, family pressure to deliver at home, husband living abroad thus no family support, previous negative experience in health facility. Causes for delay in reaching care were financial constraints and bad roads, unavailability of vehicles during monsoons and delayed referral from primary health centers.

Table 3. Still births with birth defects and labor characteristics. (N=22).

Variables		Frequency (%)
Mode of delivery	Normal	20 (90.9)
	Vaginal breech	1 (4.5)
	Cesarean section	1 (4.5)
Pregnancy complications	None	16 (72.75)
	Intrauterine growth restriction	3 (13.6)
	Hypertensive disease of pregnancy	1 (4.5%)
	Polyhydramnios	2 (9.1%)
Delays causing morbidities	Delay in recognizing care	14 (63.6%)
	Delay in seeking care	8 (36.4)
	Delay in receiving care	0 (0)

DISCUSSION:

Congenital anomalies are reported to account for 2.1–33.3% of stillbirths in low- and middle-income countries.[2]The frequency of anomalies in this study was 9.3 per 1000 births. The March of Dimes data on birth defects in South East Asian Region shows Nepal has the rate of 59.9 per 1000 live births.[3] In a prospective study in China, the prevalence of birth defects was 25.24 per 1000 perinatal infants with an uptrend over the past five years.[4]

The frequency of congenital anomaly reported in other studies are 3.46% in Bangladesh[5] and 0.81% and 0.3% in other centers in Nepal.[6,7] In a community household survey in Nepal, prevalence of congenital anomalies was 52 per 10,000.[8]

The commonly reported anomalies are cleft lip and palate,[6] anomalies central nervous system[7] and genitourinary system.[9] Our study too revealed common involvement of central nervous system and musculoskeletal system which was similar to a study conducted in Nigeria.[10]

Frequency of birth defects in Nepal is low compared to other countries. Under reporting of cases could be a major cause. Most of the congenital anomalies are reported from hospital data and anomalies in newborns delivered at home are missed. Nepal still has 57% of home deliveries.[11]

On the other hand, the incidence of birth defects in a rural setting should have been high as they are diagnosed late in pregnancy and the chances of early termination before the period of viability of 22 weeks is decreased. In a study done by S Shrestha et al, among 99 anomalies, 29 (29.29%) were detected before the period of viability.[12]

In the institute where this study is based, antenatal screening tests which are done routinely and have the potential to diagnose risk factors include blood sugar, venereal disease and ultrasonography. Screening tests like thyroid function tests or tests for infections like TORCH and for Down syndrome

like Triple test are still not affordable to all patients. Two newborns with anomalies suspected to have chromosomal anomalies in this study could probably have been diagnosed with Downs syndrome if these tests were made available. Women coming to our institute also do not opt for these tests even when advised because of financial constraints. Fetal echo has a prominent role in diagnosis of cardiovascular anomalies but again this is not accessible to women in this hilly region of the country so these anomalies are not detected in the antenatal period and maybe missed in the neonatal period if they are asymptomatic till late infancy. Neonatal screening for inborn errors of metabolism is not available and few cases of suspected anomalies in the neonatal period refuse to undergo confirmatory tests and leave against medical advice. This is a discouraging issue for service providers as the chance of correct diagnosis and counseling for future pregnancy is missed.

Another major hurdle in the accurate diagnosis and cause of birth defect is the unavailability of fetal autopsy. This can be done only for medico-legal purposes and even if clinical autopsy maybe possible; patients do no give consent for the procedure due to cultural reasons.

Maternal diabetes mellitus is a well documented risk factor for birth defects.[13] This study did not see any cases of diabetes as the incidence is low in this part of the country and also because women present late in pregnancy and the diagnosis is missed.

Consanguinity is an established risk factor for birth defects. The location of this center in Western Nepal has more people with indigenous groups where consanguineous marriages are acceptable. It was present in 46.1% of cases in this study. In the study by Rittler M et al., a significant association of consanguinity was observed for three congenital anomalies namely, hydrocephalus, hand polydactyly, and bilateral cleft lip or cleft palate.[14] In another study in North-Eastern France, consanguineous mating was present in 1.21% in parents of babies with birth defects whereas control group had consanguinity in only 0.27% ($p < 0.001$). Birth defects were 10.3 times more frequent in first degree consanguinity.[15] Another study done in Norway also showed that the relative risk of birth defects among children born with first degree consanguinity was about two.[16] Our study did not evaluate the association of consanguinity with different types of birth defects. A larger study with evaluation of each anomaly with consanguinity could be a scope for future studies.

Folic acid supplementation helps in prevention of neural tube defects.[17] In this study only 40% women had taken folic acid. Since all pregnancies are not planned, and even if they are, first antenatal visits are usually in the second trimester, this supplementation is not possible in our context. Peri-conceptional use of folic acid is recommended after large randomized trials[18] but in the context of rural Nepal, this is still a far-fetched goal due to lack of awareness. In a study by Paudel P et al., only 5% women knew that folic acid is to be taken pre-pregnancy.[19]

A large meta-analysis showed that folic acid fortification reduces the incidence of spina bifida. [20] Food fortification with folate has not been as successful as pregnancy supplementation coverage in Nepal.[21] Therefore this aspect has to be strengthened.

History of cigarette smoking and alcohol during pregnancy was present in only 10.7% mothers. This could be one reason for low incidence of birth defects. Finnel R et al. states that approximately 3% to 5% of children born in the United States have birth defects out of which 2% and 3% are teratogen-induced malformations resulting from environmental or iatrogenic exposures during pregnancy.[22] This study showed low rate of birth defects and even lower rate of teratogenic exposure. This could mean that women in this low-income country are currently facing issues of lack of awareness in consanguinity and inadequate antenatal and nutritional care but if they face greater environmental exposure to teratogens, the rate is likely to increase in the near future.

The three delays model (delay in recognizing need of care, reaching care and receiving care) has been used in assessing causes of maternal and neonatal morbidities and mortalities. [23,24] This study used the same model for understanding causes of still births with birth defects. Delay in seeking care was the most common delay. This was the cause of inadequate peri-conceptional and antenatal care and delayed diagnosis. Women in rural Nepal, do not get optimal care due to unavailability of basic needs like family support or transportation facility. 63.6%

of women with still births with anomalies who otherwise if had come to the facility in time could probably have had live births. If they had adequate knowledge on antenatal care, their babies could have been prevented from having birth defects or could have been timely diagnosed.

Primary health centers in Nepal have been strengthened over time and most women coming for antenatal visits are suggested to go to a tertiary center for anomaly scan at 20 weeks. This increases the rate of early detection of anomalies. But those women coming late for antenatal check up cannot be helped much in preventing birth defects but proper counseling for present and future pregnancy is still an opportunity not lost. Awareness regarding consanguineous marriage and folic acid intake before conception is necessary and can be done only if peri-conceptional visit to a heath center is reinforced.

Working on tackling the issue of delays in seeking care is seen as necessary so that birth defects are diagnosed in time. Either early termination of pregnancy can be advised or screening tests can be planned or at least they can be referred to a center for rehabilitation for babies with deformities. In the context of limited availability of diagnostic tests and treatment, preconceptional use of folic acid and timely diagnosis during antenatal period is a possible way to decrease birth defects and its morbidities and to help mothers with a less traumatic birth experience.

This study has a few limitations. This was a hospital based observational study. Congenital anomalies need to be assessed in a larger study in patients with or without established risk factors. This study does not consider environmental and dietary factors which are major contributors to birth defects. Degree of consanguinity can also be assessed in future studies as it is related to severity and involvement of different systems.

CONCLUSION:

Birth defects are common in this part of the country. Factors like consanguinity and intake of folic acid are associated modifiable factors which can be useful in preventing congenital anomalies. Encouraging early antenatal visits might help in early detection of anomalies. This will provide an opportunity for timely counseling so that it is less traumatic to the mother and the family.

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