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Original Article

Prevalence and Pattern of Birth Defects in a Tertiary Referral Center

Munjal Yadav*, Shanti Subedi

Department of Obstetrics and Gynaecology, Nobel Medical College Teaching Hospital, Biratnagar, Nepal

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Abstract

Background

Congenital disorders (birth defects) are structural or functional abnormalities, which are present from birth, whether recognized at birth or later and constitute a major health problem worldwide. Congenital anomalies occur in 3 to 5 % of all pregnancies and 2 to 3 % of all births. The exact magnitude of birth defect related events is still unrevealed. Prevalence is high in Nepal but true magnitude is still unknown.

Materials and Methods

This is a prospective cross sectional study of all antenatal women who had an ultrasound revealing congenital anomalies as well who delivered with a fetal congenital malformation at department of Obstetrics and Gynecology, Nobel Medical College Teaching Hospital. Maternal variables like age, parity, period of gestation at detection, medical history, mode of delivery and complications were recorded. Fetal outcomes and details of anomalies were analyzed. Consanguinity and history of use of folic acid were also inquired.

Results

Our institutional prevalence of congenital anomalies was 1.25%. The mean age of the mother is 25.88 years. Anencephaly was seen in five live birth among the mothers of consanguineous marriage. The predominant system involved was central nervous system 37 (37%) followed by Musculoskeletal system 13 (13%). Most of them 38/56 (68%) were pre diagnosed by antenatal ultrasound only in the third trimester. Majority of them did not give history of the use of folic acid. Vaginal delivery was the preferred choice of the termination of pregnancy.

Conclusion

Congenital anomaly rate was 1.25% and Central Nervous System was the predominant system involved with Anencephaly being common in consanguineous marriage.

Keywords: *Anencephaly, Birth Defects, Consanguinity, Folic acid*

***Corresponding Author:**
Dr. Munjal Yadav
Lecturer
Email: moonzal13688@gmail.com
ORCID: https://orcid.org/0000-0003-1254-4585

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Introduction

Congenital disorders (birth defects) are structural or functional abnormalities, which are present from birth, whether recognized at birth or later and constitute a major health problem worldwide [1]. Congenital anomalies occur in 3 to 5 % of all pregnancies [2] and 2 to 3 % of all births [3].

Such defects involve a single organ or a system or it may present with multiple organs as well as multiple system involvement with various etiological factors [4]. Some common defects are anencephaly, hydrocephalus, neural tube defect (NTD), Down syndrome, cleft lip and palate, congenital heart disorders, clubfoot, etc[5].

The actual estimation of prevalence and distribution of congenital anomalies in a developing country like ours is underestimated because of the lack of proper and timely diagnostic facilities as well as a lack of proper data on this topic [6]. Neural tube defect (NTD) is a serious defect that can be prevented with folic acid supplementation to the pregnant mother before and during the pregnancy [7-9].

The exact magnitude of birth defect related events is still unrevealed. Prevalence is high in Nepal but true magnitude is still unknown. Current study aim is to establish the prevalence and pattern of congenital anomalies through obstetric ultrasonography in unselected population visiting Nobel Medical College Teaching Hospital.

Materials and Methods

This Cross sectional study was conducted at the department of Obstetrics and Gynecology, Nobel Medical College Teaching Hospital from May 2019 to May 2020. Institutional ethical committee clearance was obtained with reference number 264/2019. Written informed consent was taken from the parents of the included cases. All antenatal women who had an ultrasound revealing congenital anomalies as well who delivered with a fetal congenital malformation were included. Medico legal cases were excluded. The sample size was calculated to be 73. However, a total of 100 patients were enrolled. The sample size (n) was calculated as follows, $n = Z^2 x\ p\ x\ q/d^2 = (1.96)^2\ x\ 0.05\ x\ 0.95/\ (0.05)^2 = 73$, Where, Z= 1.96 for 95% confidence interval p= 5% [2] q= 1-p, d= margin of error = 5%. Sampling technique was purposive consecutive sampling.

Maternal variables like age, parity, period of gestation at detection, significant medical history, mode of delivery and complications were recorded. Feta outcome, predominant system involved and significant morphological findings were noted. Consanguinity and history of use of folic acid was inquired through a predesigned semi-structured questionaire. The classification of the malformation was done based on the anatomical system.

Data collected were entered in MS Excel and SPSS 23.0 version was used for descriptive analysis in percentage and mean.

Results

A total of 100 newborns with congenital defects were studied over the year. The incidence of congenital anomalies at our institute was 1.25%. The mean age of the mother is 25.88 years with a minimum age of 19 years old and maximum of 40 years old. Sixty percent of the pregnant females were in the range of 20-30 years of age group. Among total cases, 12 were muslim by religion and all of them had a consanguineous marriage. There were five cases of anencephaly. Other congenital anomalies found among themare listed in table 1.

Table 1: Congenital disordersin consanguineous marriage (12/100)

Congenital disorders in consanguinity	Frequency N (%)
Anencephaly	5 (41.66)
Phocomelia	2 (16.6)
Diaphragmatic hernia	1 (8.33)
Hydrocephalus	1 (8.33)
Hydrops fetalis	1 (8.33)
Club foot	1 (8.33)
Undescended testis	1 (8.33)
Total	**12 (100)**

Most of the newborns were male (n=56), 40 were female and 4 cases of ambiguous genitalia.

A total of 44cases were pre-diagnosed by Obstetric Ultrasound at our institute. 44 cases of congenital anomalies were diagnosed only after delivery during routine neonatal examination whereas 12 cases were referred with pre diagnosed cases from nearby health centers. Only 13 patients had regular antenatal visits at this institute.

Table 2: Showing major system involved

Major System involved	Frequency (%)
CNS	37 (37)
MSK	13 (13)
Digestive	10 (10)
Hydropsfetalis	9 (9)
Renal	9 (9)
Genitourinary	7 (7)
Cleft lip/palate	6 (6)
Respiratory	4 (4)
Lymphatic	3 (3)
Others	2 (2)
Total	100 (100)

Among the total congenital anomalies, the predominant system involved was the central nervous system 37(37%) followed by the musculoskeletal system 13(13%) and digestive 10(10%) as mentioned in table 2. Here others belong to two rare multisystem anomalies. One was Body stalk anomaly and the other was Isolated Ectopiacordis with anencephaly.

Table 3: Showing significantfindings (morphological and Ultrasonography detected)(N=98)

Significant Findings	Frequency (%)	Significant Findings	Frequency (%)
Anencephaly	17(17%)	Hydrops Fetalis	9(9%)
Hydrocephalus	13(13%)	Cleft lip/ cleft palate	6(6%)
Meningomyelocele	2(2%)	Congenital cystic adenomatoid malformation	2(2%)
Spina bifida	2(2%)	Congenital cystic adenomatoid malformation	2(2%)
Aqueductal agenesis	1(1%)	Cyclops	1(1%)
Corpus callosum agenesis	2(2%)	Cystic Hygroma	2(2%)
Club foot	6(6%)	Hydrocele	1(1%)
Poldactyly	2(2%)	Ambiguous Genitalia	4(4%)
Phocomelia	2(2%)	Renal Agenesis	1(1%)
Skeletal Dysplasia	2(2%)	Bilateral Hydronephrosis	5(5%)
KlippelFeil Syndrome	1(1%)	Hypospadias	1(1%)
Omphalocele	6(6%)	Undescended testis	1(1%)
Diaphragmatic Hernia	2(2%)	PUV with UB diverticulum	2(2%)
Anogenital atresia	2(2%)	UB diverticulum with HNS	1(1%)
Body Stalk Anomaly	1(1%)	Isolated Ectopiacordis with anencephaly	1(1%)

Anencephaly (17), hydrocephalus (13) and hydropsfetalis (9) were highly prevalent anomalies seen in our study. Among the total cases (56) diagnosed by antenatal ultrasound, majority of the cases 38(68%) were diagnosed only during the third trimester followed by second trimester 18(32%). Almost more than half (n=58) of the pregnant female did not use folic acid. Among congenital anomalies related to the central nervous system (37), 13(35.13%) did not have a history of use of folic acid. Regarding Medical history 4 were overt diabetic with Neural tube defects, one case of VDRL positive with Hydrops fetalis and only 2 cases had history of hypertensive disorders of pregnancy that delivered newborn with hydrocele and polydacty-lyeach. There were 89 cases of cephalic presentation, 10 cases of breech presentation and one case of face presentation. Pregnancy was terminated through vaginal delivery in 78 of them whereas 19 had to go through cesarean section for non-progress of labour. Two had vaginal birth after caesarean section. One even had to go through a Caesarean hysterectomy for atonic Postpartum haemorrhage.

Discussion

Congenital anomalies have become an important cause of perinatal mortality in developed countries as well as developing countries. Most of the developing countries lack the proper estimation of the congenital anomalies. In our study, the institutional prevalence of birth defect was 1.25%. These findings are comparable to similar studies from India, which reported an incidence of 2.73% and 1.8% [10,11]. Our hospital is tertiary referral center, usually gets referred complicated cases and hence the prevalence in the hospital cannot be projected into the total population. Male predominance amongst congenital malformed babies was seen in our study which is similar to the study done by Lei Z in china [12]. The majority of the mothers were primi belonged to the age group 20 to 30 years and none were above 40 years, which is different from the study done in India reporting higher incidence in multiparous and older age [13]. Twelve (12%) cases had consanguineous marriage with high frequencies of anencephaly 5/12 (41.6%). Consanguineous marriage, which is still a common practice in the country, has been found out to have a significant role similar to study done by Kannan ZM in 2008[14].

The commonest congenital anomalies were central nervous system 37(37%) followed by musculoskeletal system13 (13%) and digestive system 10 (10%). However according to the study done by Khanal GP et al in 2019 in mid western part of Nepal, most commonly involved body system was central nervous system 51 (48.1%), followed by cardiovascular system 12 (11.3%), musculoskeletal system 10 (9.4%), gastrointestinal system 5 (4.7%), respiratory system 5 (4.7%), genitourinary system 4 (3.8%). During this period, 106 (1.02%) pregnant women presented with different congenital anomalies which is again similar to ours [15]. The study done by Shrestha S. et al in 2018 congenital anomaly rate was (0.37%) which is lower than ours is. Central nervous system had the highest common anomalies detected 40 (32%) followed by gastrointestinal anomalies 33 (26.4%) [16]. In our study 38(68%) of anomalies were detected only during the third trimester followed by second trimester 18(32%), however report by N kashyap 52.1% detected after 20 weeks [17]. The USG should be done as early as 11 weeks to detect the anencephaly so that the pregnancy can be terminated earlier within the legal framework of

the country [18].

Facial defect like cleft lip/palate 6(6%) were detected only after delivery during neonatal examination at delivery. Also study done by Shrestha S. et al in 2018 has shown low prediction and only 2 (15.3 %) of 13 cases were detected on USG, comparable to the study done by Clementi M et al [19], however, recent study shows, it can be detected in up to 65% [20]. Once the anomalous fetus is detected, the preferred method of delivery is through vaginal route without compromising the health of the mother. 78 (78%) of them delivered through vaginal route, 19 cases underwent cesarean section for non progress of labour. Two cases underwent vaginal birth after cesarean section and only one case underwent caesarean hysterectomy for intractable atonic postpartum haemorrahage. Despite of various governmental program for antenatal checkup, the detection of congenital anomalies was 68% in third trimester and the rate of congenital anomaly is still very high in eastern part of Nepal most probably due to illiteracy, poverty and lack of awareness regarding use of folic acid. Consanguineous marriage still being our culture and tradition commonly practised in muslim and some mongolian tribe adds the genetic burden might contribute to its higher prevalence.

Limitation of our study was multiple pregnancies as well as chromosomal anomalies. The low detection of cardiac anomalies in our study could be due to lack of fetal echocardiography service at our institute.

Conclusion

Congenital anomaly rate was 1.25% from our study. Central Nervous System was the predominant system involved with Anencephaly being common in consanguineous marriage. Majority of the pregnant women did not take folic acid during antenatal visits. Obstetric Ultrasound detected the anomalies mostly during the third trimester. Vaginal delivery was the preferred method for termination of pregnancy.

Recommendation

Taking care of the anomalous child is a burden to the family and the society in the developing countries, both mentally and economically. Maternal education, regular antenatal checkup, proper vaccination and supplementation of folic acid, avoidance of unnecessary drugs, toxins, and availability of early detection and termination of the congenital anomalous child will reduce the burden of congenital anomalies in society.

Conflicts of interests: None

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