Original Research Article

FREQUENCY RISK FACTORS AND PATTERN OF CONGENITAL ANOMALIES IN NEONATES IN NAWABSHAH PAKISTAN

ABSTRACT

Objectives: The objectives of this study were to determine the frequency, risk factors and pattern of congenital anomalies in neonates admitted at a tertiary care hospital. **Methodology:** It was a descriptive cross-sectional study. Research was conducted at NICU Pediatric ward, PMCH Nawabshah. Duration of study was 6 months after the approval of synopsis from January 2020 to July 2020. This study involved 300 neonates in NICU which were screened for congenital anomalies. Age, gender and birth weight of the neonate was recorded. Mothers were also enquired about consanguinity and use of folic acid and smoking and diabetic status during pregnancy. Frequency and type of birth defect were noted and were compared across various subgroups based on various neonatal and maternal factors. **Results:** The mean age of the neonates was 10.5±7.4 days while the mean birth weight was 2.89±0.74 Kg. There were 174 (58.0%) male and 126 (42.0%) female neonates with a male to female ratio of 1.4:1. 77 (25.7%) neonates were low birth weight. Birth defect was noted in 35 (11.7%) neonates. 3 neonates had multiple anomalies. The frequency of birth defects was significantly higher in women with no use of folic acid during pregnancy (14.8% vs. 5.2%; p-value=0.015) and cousin marriage (16.6% vs. 2.8%; p-value<0.001). **Conclusion:** In the present study, 11.7% of neonates had birth defects comprising of congenital hydrocele, spina bifida, cleft lip/palate and club foot that were associated with lack of folic acid use during pregnancy and cousin marriages. Public awareness against cousin marriages and antenatal counseling of mothers for routine folic acid use during pregnancy should be assured for this.

Key words: Birth Defect, Consanguinity, Folic Acid, Nawabshah

INTRODUCTION

Birth defects or congenital anomalies (CA) are disorders of structure, function and behavior that occur during embryonic development and can be screened before delivery, at birth or later during early infancy and childhood[1]. These birth defects have a significant impact on families as well as health system, contributing specially to morbidity and mortality in neonates. Congenital anomalies may be categorized as major or minor defects. A minor defect is defined as structural abnormality which is present at birth and has insignificant effect on physiological function, but may affect cosmesis e.g. ear tag, while on the other hand a major defect has a substantial effect on physiological function or on public appropriateness e.g. a cleft lip and VSD (ventricular septal defect)[2].

Whatever the type of congenital anomaly is there, it remains stressful for the mother as well as whole family who have waited long enough to hold a normal child. Furthermore the nursing care of an abnormal child may itself be very stressful for the family like a child with cleft lip/palate. Sometimes, the abnormality becomes stressful with the passage of time like a child with ambiguous genitalia is a source for persistent frustration of the family. According to WHO, an estimated 303,000 newborns die within 4 weeks of birth every year, worldwide, due to birth defects; contributing a major effect on individuals as well as on families, healthcare systems and societies[3].

Global prevalence of congenital anomalies range from 3-6%[4], while in Pakistan various studies shows different prevalence, for example a study done at 3 hospitals of Punjab showed a prevalence of 7%[5], prevalence of birth defects was also variable for different birth defects. Regrettably, ≥90% of birth defects are encountered in low and middle income countries (LMICs)[6]. The frequency of birth defects differs from population to population. Also different ethnic backgrounds have variable prevalence of congenital anomalies. In Pakistan 6-9% of perinatal deaths can be attributed to different congenital anomalies[7].

Understanding the mechanisms behind the development of CA is vital for the prevention as well as genetic counseling that may help in eradication of the problem. Generally, the etiology of birth defects remains unclear but is thought to be multifactorial. There are some known genetic, environmental and other causes or risk factors associated with congenital anomalies but approximately 50% of all congenital anomalies cannot be linked to a specific cause[8]. Genetic predisposition or cousin marriages also play a major role in birth defects[9]. Infectious agents and infections seem to be the most imperative environmental factor in low and middle income countries. Inheritable congenital diseases, previous miscarriages and history of stillbirths are other important risk factors in the etiology of CAs. Major CAs occur in 2–3% of live births, 20–30% of still births and 14–28% of pediatric hospitalizations have been in the United States have been linked to CAs. Mortality is very high among major CAs in low and middle income countries rising to 25–80%. A substantial number of survivors also undergo life-long debilities, with birth defects responsible for 25.1 to 35.6 million disability-adjusted life years, worldwide[10].

Each year in United States, about 1,645 babies are born with spina bifida[11]. Globally the range for some congenital anomalies like cleft lip/palate is different in various socioeconomic groups, like in low socioeconomic regions its prevalence is 82/1000 live births and in high income regions its 39.7/1000 live births[12]. The prevalence of cleft lip is 1 in every 1,000 live births[12]. It is more common in male individuals (80%) than in female individuals, and its incidence rate increases slightly with maternal age where as the incidence of cleft palate is much lower than that of cleft lip at 1 in every 2,500 live births, and is more common in female individuals (67%) than in male individuals[13]. Encephalocele has an estimated prevalence of 0.8–5.0 per 10,000 live births[14]. Every year, around 100,000 babies worldwide are born with a clubfoot. Clubfoot is an inborn deformity of the foot, where either one or both feet are twisted inward, causing the child

to walk on his ankles. Left untreated, the condition causes severe lifelong disability. 80% of untreated clubfoot are found in developing countries[15]. Minor anomalies occur in approximately 15% of newborns but these structural abnormalities are not themselves detrimental to health but, in some cases, are associated with major defects. For example, infants with one minor anomaly have a 3% chance of having a major malformation; those with two minor anomalies have a 10% chance; and those with three or more minor anomalies have a 20% chance[16].

There has been an increasing drift towards the prevalence of birth defects owing to emergence of new threats like Zika virus[17,18]. China has recently introduced surveillance and monitoring system to detect birth defects while there is no such monitoring system in our country in spite of increasing need for such a system owing to alarmingly higher birth defects in this part of the world[19].

This study was performed with the rationale to determine the frequency and distinguish the most common types of birth defects and identify their risk factors if any in our study population so that the findings may help health experts to focus on the preventive tactics accordingly.

The objectives of this study were to determine the frequency, risk factors and pattern of congenital anomalies in neonates admitted at a tertiary care hospital.

MATERIALS AND METHODS

Study Design

It's a descriptive cross sectional study.

Setting

Research was conducted at NICU Pediatric ward PMCH Nawabshah.

Duration of Study

Duration of study was 6 months after the approval of synopsis from January 2020 to July 2020.

Sample Size

Sample size of 300 cases was calculated with 95% confidence level and 5% margin of error while taking expected frequency of birth defects to be 7.0% in Pakistan[4].

Formula: $\frac{Z_{1-a/2}^2 P(1-p))}{d^2}$

Sampling Technique

Patients were selected by Non-Probability, Consecutive Sampling.

Inclusion Criteria

All neonates (babies of age 0-28 days) of either gender admitted to NICU at Pediatric ward PMCH Nawabshah.

Exclusion Criteria

Neonates who had any physical/structural deformity that was secondary to birth trauma or other iatrogenic cause were not included.

Parents who did not gave consent were to be part of the study

Data Collection Procedure

These babies were evaluated for type of congenital anomaly and risk factors if found any. The required data was collected on predesigned proforma by the researcher himself and in his absence a suitable person was assigned to record data. All the expenditures of study if any were compensated by researcher himself.

Data Analysis Procedure

Data was entered and analyzed through SPSS version 20.

1. Numerical variables; age in days and birth weight have been presented by mean ±SD.
2. Categorical variables i-e gender, presence of congenital anomaly and type of congenital anomaly and risk factors has been presented by frequency and percentage.

3. Data has been stratified for age, gender and risk factors to address effect modifiers. Post-stratification chi-square test has been applied taking $p \leq 0.05$ as significant.

RESULTS

The age of the neonates ranged from 1 to 28 days with a mean of 10.5±7.4 days while the birth weight ranged from 1.5 Kg to 4.3 Kg with a mean of 2.89±0.74 Kg. Majority (n=190, 63.3%) of the neonates were aged between 1-14 days of life followed by 110 (36.7%) neonates aged between 15-28 days of life as shown in Figure 1.

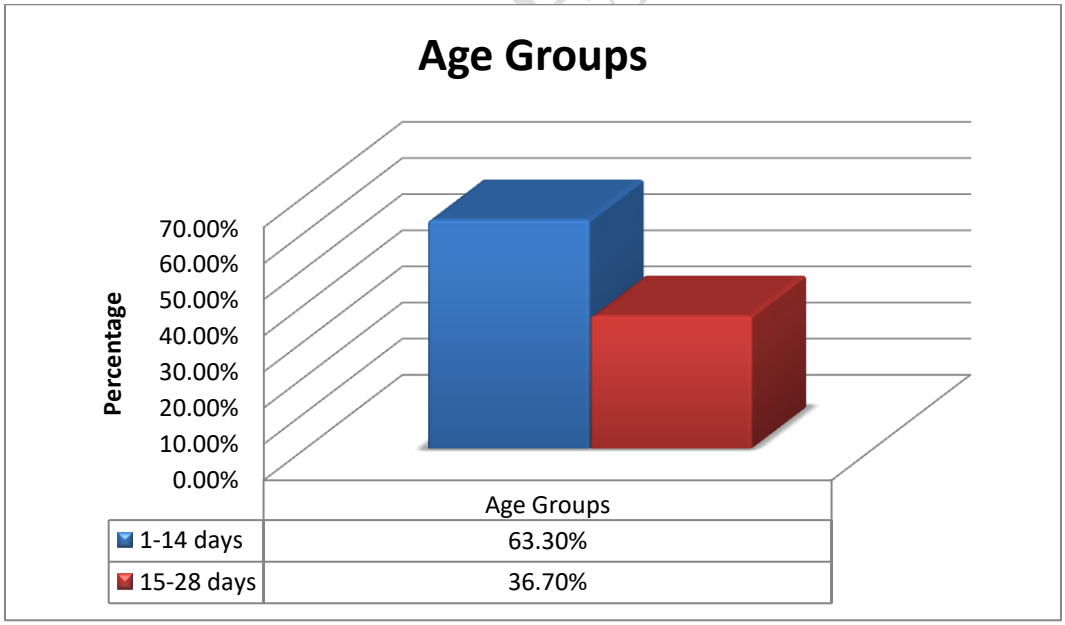

Fig. 1. Distribution of various age groups in the studied sample

There were 174 (58.0%) male and 126 (42.0%) female neonates with a male to female ratio of 1.4:1 as shown in Figure 2.

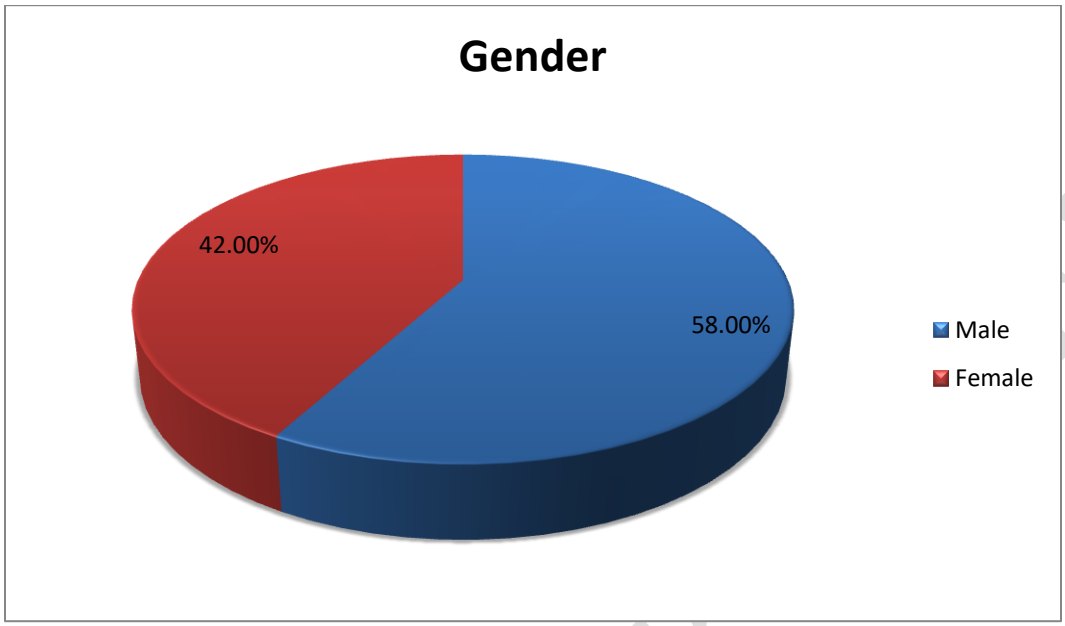

Fig. 2. Distribution of various genders in the studied sample

Upon inquiry from the mothers, a substantial proportion of women (67.7%) didn't take folic acid during pregnancy while history of consanguinity, smoking and diabetes was positive in 193 (64.3%), 56 (18.7%) and 37 (12.3%) mothers as shown in Table 1.

Table 1. Frequency of various risk factors in the studied population

Risk Factor	Frequency (n)	Percent (%)
Low Birth Weight	77	25.7%
Consanguinity	193	64.3%
No use of Folic Acid	203	67.7%
Smoking	56	18.7%

Diabetes	37	12.3%

There was no statistically significant difference in the frequency of birth defects across various subgroups based on age (p-value=0.950), gender (p-value=0.913), low birth weight (p-value=0.676), birth order (p-value=0.947) and smoking (p-value=0.255) and diabetic (p-value=0.709) status of the mother. However, the frequency of birth defects was significantly higher in women with no use of folic acid during pregnancy (14.8% vs. 5.2%; p-value=0.015) and cousin marriage (16.6% vs. 2.8%; p-value<0.001) as shown in Table 2.

Table 2. Comparison of Birth Defects across various Subgroups

n=300

Characteristics	**n**	**Birth Defect n (%)**	**P-value**
Age			
• **1-14 days**	190	22 (11.6%)	0.950
• **15-28 days**	110	13 (11.8%)	
Gender			
• **Male**	174	20 (11.5%)	0.913
• **Female**	126	15 (11.9%)	
Low Birth Wight			
• **Yes**	77	10 (13.0%)	0.676
• **No**	223	25 (11.2%)	
Birth Order			
• **1**	63	7 (11.1%)	
• **1-4**	133	15 (11.3%)	0.947
• **≥5**	104	13 (12.5%)	
Folic Acid Use			
• **Yes**	97	5 (5.2%)	0.015*
• **No**	203	30 (14.8%)	
Smoking			
• **Yes**	56	9 (16.1%)	0.255
• **No**	244	26 (10.7%)	

Diabetes			
• **Yes**	37	5 (13.5%)	0.709
• **No**	263	30 (11.4%)	
Cousin Marriage			
• **Yes**	193	32 (16.6%)	<0.001*
• **No**	107	3 (2.8%)	

** The difference was statistically significant on chi-square test*

DISCUSSION

Once leading cause of perinatal mortality, perinatal infections have been controlled by the advent and availability of antibiotics. Birth defects have now surfaced as leading cause of intrauterine death and early neonatal death and are frequently associated with decreased productivity among the survivors[1]. Repeated fruitless surgery, tiring rehabilitation and occupational therapy all have failed to improve the outlook of problem. The focus therefore changed to timely identification of abnormal fetus and termination of pregnancy to reduce the burden of problem[2]. Still to date 8.14 million children are born every year with some sort of congenital anomaly which is alarming and requires ongoing research and measures to counteract this problem[1].

"Prevention is better than cure" is a well-known saying and fits very well in this scenario where preventing a birth defect would simply avoid the need for prenatal screening, termination of pregnancy and wastage of resources on rehabilitation of a child with birth defect. It would also relieve the social stigma associated with a disabled child[6]. In the light of this concept, a number of studies have assessed relationship between various maternal and environmental factors and birth defects and have reported varying results[1]. The main issue among the studies has been selection bias where birth defect definition and diagnostic criteria is quite variable among studies. Also the racial and geographic differences play a critical role[6,7].

In the present study, the mean age of the neonates was 10.5±7.4 days while the mean birth weight was 2.89±0.74 Kg. The birth weight of most of the neonates was in the ranged of 2.5-4 Kg (n=194, 64.7%) followed by <2.5 kg in 77 (25.7%) neonates and >4.0 Kg in 29 (9.6%)

neonates. Our observation is in line with that of Islam et al.[20] (2013) who reported similar mean age of 10.2±9.8 years and mean birth weight of 2.3±0.6 Kg among Bangladeshi neonates diagnosed of birth defect.

We observed that there was a slight male predominance with a male to female ratio of 1.4:1. Our observation is in line with that of Hussain et al.[5] (2014) who reported a similar male predominance with male to female ratio of 1.4:1 among such neonates at Combined Military Hospital, Kharian.

We also noted that the frequency of birth defects was significantly higher in women with no use of folic acid during pregnancy (14.8% vs. 5.2%; p-value=0.015) and cousin marriage (16.6% vs. 2.8%; p-value<0.001). Thus lack of folic acid use during pregnancy and cousin marriages appear potential risk factors of birth defects. Cousin marriage is a dilemma of Asian countries where a lot has been said and endeavored, yet the rate of cousin marriages remains alarmingly higher. Even in the present study, we noted that 64.3% of the mothers had cousin marriage. Moreover, folic acid which is a cheap and easily available medicine over the counter as well as free of cost in the pharmacies of many public hospitals was not taken by the pregnant mothers. In spite of routine antenatal visits and counseling by the attending gynecologist, this failure appears to be related with educational level of the mother and her partner as well as myths and phobias in the society. There is need for public awareness campaigns against the practice of cousin marriages as well as to improve the awareness of general public about the easy availability and importance of folic acid for the prevention of these birth defects.

The present study adds to the already published research evidence on the topic. In the present study, 11.7% of neonates had birth defects comprising of congenital hydrocele, spina bifida, cleft lip/palate and club foot that were associated with lack of folic acid use during

pregnancy and cousin marriages which advocates public awareness against cousin marriages and antenatal counseling of mothers for routine folic acid use during pregnancy to decrease the likelihood of birth defects and improve the perinatal outcome. The strengths of the present study were its large sample size of 300 cases. Standard guidelines were followed to diagnose and label a case of birth defect. We also stratified that data for various affect modifiers. However a very strong limitation to the present study was that it was a cohort study. A case-control study would have enabled estimation of risk associated with each contributing factors which would have been more helpful. Such a study is highly recommended in future research.

CONCLUSION

In the present study, 11.7% of neonates had birth defects comprising of congenital hydrocele, spina bifida, cleft lip/palate and club foot that were associated with lack of folic acid use during pregnancy and cousin marriages thus thesis need for public awareness against cousin marriages and antenatal counseling of mothers for routine folic acid use during pregnancy in order to decrease the likelihood of birth defects and improve the perinatal outcome.

Ethical Approval:

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

Consent

As per international standard, parental written consent has been collected and preserved by the author(s).

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