ORIGINAL ARTICLE

Frequency of major congenital malformations in neonates born in three tertiary care hospitals of Pakistan.

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ABSTRACT... Objective: To assess the frequency of major congenital anomalies in newborns delivered in three different hospitals of Pakistan. **Study Design:** Cross Sectional study. **Setting:** Department of Pediatrics Hameed Latif Teaching Hospital, Lahore, Ittefaq Hospital Lahore and HITEC-IMS Taxilla. **Period:** Jan 2023 to November 2023. **Methods:** 205 neonates who met the inclusion criteria were included in the study from labour room of Hameed Latif Teaching Hospital Lahore, Ittefaq Hospital Lahore and HITEC-IMS Taxilla. Informed consent was obtained from parents. Demographic variables (name of mother, gender, birthweight and gestational age at birth) were recorded. Then neonates were admitted in NICU and evaluated for presence of congenital malformations including cardiovascular system defects, limb anomalies, genitourinary system, central nervous system and chromosomal anomalies. All data was entered in specially designed performa. **Results:** We found that overall gestational age was greater than 32 weeks with mean age of 37.190±2.00 weeks. Mean birth weight was 2.938±0.31 Kg. Frequency and percentage of patients according to gender was 60% males and 40% females. Major congenital malformation was seen in 2.9% patients. Among infants with major congenital malformations, cardiovascular system defects were seen in 33.3% infants, limb anomalies 16.7%, genitourinary system 16.7% and central nervous system defects were 33.3%. Parents of 67.7% of cases were cousins. It was observed that only 32.3% of the mothers had taken the recommended daily dose of folic acid in antenatal period. **Conclusion:** Central nervous system and cardiovascular system defects were the most prominent anomalies detected. Prenatal diagnosis may be helpful in decreasing mortality by offering early termination.

Key words: Congenital Malformation, Cardiovascular System Defects, Central Nervous System Defect, Newborn.

INTRODUCTION

A congenital anomaly or malformation is a birth defect which can be either structural or functional. It can adversely affect the appearance, structure or function of involved body part. The etiology of these defects is multifactorial depending upon genetic tendencies, environmental influences or a combination of both. The prevalence and specific types of congenital abnormalities is variable over different regions of the world. Central nervous system (CNS), cardiovascular system, and musculoskeletal system anomalies are most frequently observed.[1] According to WHO, every year approximately 240,000 neonatal deaths are caused due to congenital malformations. These disorders prove fatal for an additional 170,000 children aged 1 month to 5 years.[2] Classification of birth defects is based upon various factors such as severity, etiology and system affected.[3,4]

Due to scarcity of data on congenital malformations from developing countries, it is prudent to collect thorough and reliable information regarding the nature and prevalence of these malformations. Such data is helpful for monitoring purposes and planning of efficient public health strategies targeting prevention and treatment.[5]

In the research, 0.59% of newborns were found to have major congenital anomalies. Chromosomal abnormalities accounted for 8.7%, circulatory system issues were seen in 20.4%, genital system

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anomalies in 3.5%, urinary system defects in 4.1%, nervous system anomalies in 11.1%, and musculoskeletal issues in 15.5% of the cases.[6]

In a hospital based research in Kharian, Pakistan, the incidence of major congenital malformations among newborns was 7%.Further breakdown according to specific diagnosis was chromosomal abnormalities 6.19%, circulatory system issues in 13.27%, genitourinary system anomalies in 15.04%, nervous system complications in 20.35%, and musculoskeletal issues in 18.58% of the cases.[7]

In a different study conducted in Abbottabad, the prevalence of major congenital malformations among newborns was found to be 4.2%. chromosomal abnormalities accounted for 5%, cardiovascular system issues were observed in 16%, urogenital system anomalies in 6%, nervous system defects in 31%, and musculoskeletal problems in 9% of the cases.[8]

Evidence from literature suggests that major congenital malformations are not very common in newborns, yet there is variation in findings observed from different sources. This inconsistency incites the debate that whether neonates should receive specialized attention for screening of malformations. Additionally, there is scarcity of local research in this specific area. Hence, we undertook this study in three tertiary care hospitals of Rawalpindi and Lahore to ascertain the prevalence of congenital malformations among neonates. Our aim is to produce findings relevant for local contexts, paving the way for recommending routine screening of neonates for these malformations. Such an approach would promote timely interventions through antenatal screening of expectant mothers. This will help to reduce the adverse outcomes associated with congenital malformations in newborns.

METHODS

This study was aimed to assess the frequency of major congenital malformationsinnewborns delivered in three tertiary care hospitals of Pakistan.

This Cross sectional study was conducted at Department of Pediatrics Hameed Latif Teaching Hospital Lahore, Ittefaq Hospital Lahore and HITEC-IMS Taxilla from Jan, 2023 to Nov, 2023 after approval from institutional ethics committee (HLTH/ADMIN-23/304) (28.12.23).

Sample Size

A Sample size of 205 cases was calculated with 95% confidence level, 3.5% marginoferrorand taking expected percentage of major congenital malformations i.e 7%.[4]

Sampling Technique

Non-Probability consecutive sampling.

Inclusion Criteria

Neonates delivered at gestational age greater than 32 weeks of either gender delivered in labour room.

Exclusion Criteria

*   Neonates born after in-vitro fertilization.
*   Patients over six weeks of age.

Diagnostic Criteria

Congenital malformations were diagnosed by detailed physical examination and or radiological studies in suspected cases.

Data Collection Procedure

205 neonates who met the inclusion criteria were included in the study. Informed consent was obtained from attendants. The neonates were recruited from the Department of Obstetrics & Gynecology of Hameed Latif Teaching Hospital Lahore, Ittefaq Hospital Lahore and HITEC-IMS Taxilla. Demographic variables (name of mother, gender, birthweight and gestational age at birth) was also obtained. The neonates were admitted in NICU and evaluated for presence of congenital malformations including cardiovascular system defects, limb anomalies, genitourinary system, central nervous system, (as per operational definition). All the record was kept in pre designed structured performa.

Data Analysis

Data was entered and analyzed through SPSS

version 22. Quantitative variables like gestational age at birth and birthweight were presented as mean and SD. Qualitative variables like gender and major congenital malformations (cardiovascular system defects, limbs anomalies, genitourinary system, central nervous system) were represented as frequency and percentage. Data was stratified for gender, birth weight and gestational age at birth. Stratified groups were compared for major congenital malformations by using chi square test. P value <0.05 was considered as significant.

RESULTS

In this study the gestational age of the patients was greater than 32 weeks with mean age of 37.190±2.00 weeks and mean birth weight was 2.938±0.31 Kg as shown in Table-I.

Frequency and percentage of patients according to gender was 60% males and 40% females as shown in Table-II. Consanguinous marriage was present in parents of 67.7% of cases with various anomalies. Only 32.3% of the mothers had taken oral folic acid during pregnancy as shown in Table II. Major congenital malformation was seen in 2.9% patients as shown in Table-III. Among infants with major congenital malformations, cardiovascular system defects were seen in 33.3% infants, limb anomalies 16.7%, genitourinary system 16.7% and central nervous system defects were 33.3% as shown in Table-IV.

Demographics	Mean±SD
Gestational age at birth (weeks)	37.190±2.00
Birth weight (Kg)	2.938±0.31

Table-I. Mean±SD of patients according to gestational age and birth weight n=205

Gender	Frequency	Percentage
Male	123	60%
Female	82	40%
Consanguinity	139	67.7%
Folic acid & Multivitamins intake	66	32.3%

Table-II. Frequency and percentage of patients according to gender n=205

Major Congenital Malformation	Frequency	Percentage
Yes	6	2.9%
No	199	97.1%
Total	205	100%

Table-III. Frequency and percentage of patients according to major congenital malformation n=205

Type of Major Congenital Malformation	Frequency	Percentage
Cardiovascular System	2	33.3%
Limb Anomalies	1	16.7%
Genitourinary System	1	16.7%
Central Nervous System	2	33.3%
Total	6	100%

Table-IV. Frequency and percentage of patients according to types of major congenital malformation (n=6)

DISCUSSION

Congenital malformations are reported in approximately 3% of live births. The incidence varies in different regions.[9] These anomalies can be diagnosed during antenatal or post natal period depending upon their particular nature. Congenital malformations are important contributors to infant disease and death. The developed world has well organized monitoring and screening programs to determine the prevalence of congenital malformations and develop effective strategies for their prevention.[10]

The frequency of congenital anomalies observed in our study was 2.9% i.e. 29/1000. Another study from Kohat concluded that frequency of malformations was 0.97%.[11] Where as a study in Karachi observed a frequency of 11.4 per 1000 live births.[12] A study in India found that the frequency of congenital anomalies was 1.91%.[13] Our study results are comparable to a study from Iran which reported that the frequency of congenital anomalies was 29.4 per 1000 live births.[14] Our results also relate to a Canadian research which reported the frequency to be 36.18 per 1000.[15]

In our study, we found that majority of defects were related to the central nervous system and cardiovascular system (33.3%). CNS anomalies included spinal dysraphism, anencephaly and hydrocephalus. Some cases of hydrocephalus were associated with spina bifida. These results are consistent with a Turkish study which demonstrated that congenital anomalies of central nervous system were frequently observed.[16] Another research from Egypt also showed that CNS malformations were most commonly reported in both live and still births.[17]

In our study male to female gender ratio was 1.2:1.Lisi reported that the variation in gender distribution depends on whether the malformation is isolated, associated with another anomaly or syndromic.[18] It is hence inferred that gender distribution should be studied in relation to every anomaly separately rather than collectively for all malformations.

Marriages between cousins are prevalent in various ethnic and religious populations.[19] Pakistan is considered to have the highest global rate of inter cousin marriages (61%) involving first and second cousins.[20] In our study cousin marriage was found in 67.7% of cases with various congenital anomalies (Table-II). A similar research in Iran concluded that occurrence of birth defects was 3.5 times more frequent in consanguineous parents as compared to others.[21]

It was observed in our study that only 32.3% females had taken preventive daily oral dose of folic acid during gestation (Table-II). This indicates that dietary non compliance is a major contributory factor for birth defects. To decrease the incidence of neural tube defects, it is advised that all expectant mothers should daily take 400 mcg of folic acid.[22]

Research indicates that advanced maternal age is associated with a rise in chromosomal disorders, making it a potential determinant of Down's syndrome.[23] In our study mean age of mothers was 30±8 years. Only 19.4% of mothers were above 40 years of age. Our results are similar to a study done in Iran which quoted maternal age of approximately 25 years and about 9% mothers were more than 35 years.[19] Apart from maternal age, increased parity has a higher incidence of congenital anomalies.[24] In our study about 70% mothers were having multiple children. This observation in comparable to another study by Qazi et all which concluded that most of the birth defects were reported in neonates of multiparous females.[25] Contrasting results were quoted in a study by Perveen et all that demonstrated more congenital anomalies in first born babies.[16] Old age of mothers is a significant risk factor associated with congenital malformations. Due to this concern, women over the age of 30 should undergo more thorough evaluations, as they are more prone to have babies with congenital malformations.

One of the limitations of our study is that there is a proven correlation between folic acid deficiency and neural tube defects. We were not able to find the serum and blood folate levels due to financial constraints. Moreover, the exact diagnosis of chromosomal abnormalities was not established due to unavailability of relevant tests. Since it is a cross-sectional descriptive study, the results should not be considered a representation of whole population. However this study highlights a significant public health concern, which is a harbinger of further well organized studies.

Birth defects are a significant contributor to fetal deaths, therefore it is essential to ascertain the frequency and prevalence of these defects in the society. This study found that birth defects were commonly observed in young mothers. Family marriages were reported as a major association. The most common defects were associated with central nervous system. Diagnosis in early gestation is crucial in reducing death in perinatal period, as it provides the possibility of timely abortion when necessary. This study sheds light on the prevalence of birth defects and observed risk factors associated with them. Additional research is suggested to devise preventive strategies for lowering the occurrence of congenital malformations.

CONCLUSION
Central nervous system and cardiovascular system defects constituted the major part of anomalies observed. Mortality can be reduced by diagnosis in early pregnancy and offering termination if considered appropriate. Marriages among cousins and lack of folic acid intake were reported two frequent associations of congenital malformations. It is possible to eliminate these risk factors by avoiding inter cousin marriages and emphasizing the intake of folic acid in first trimester of pregnancy.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

SOURCE OF FUNDING

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2	Madiha Fayyaz	Introduction and Discussion writing.	
3	Somayya Siddiqa	Data analysis.	
4	Amna Siddique	Conclusion, References.	
5	Nazir Ahmed Malik	Proof reading.	
6	Haroon Rashid	Proof reading.	