Original Article

Surveillance of Congenital Malformations and Their Possible Risk Factors

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Abstract

Objective:To determine the frequency, distribution, and patterns of congenital malformation (CMF) among babies born in a teaching hospital in Punjab, while also investigating factors contributing to their occurrence.
Methodology: This prospective study was conducted at Tertiary care Hospital Nishtar 2, Multan from January 2023 to December 2023. CMF was operationally defined as structural abnormalities detected either at birth or within the first week of life, with major anomalies necessitating surgical or medical intervention due to serious structural, cosmetic, or functional impairment. The diagnosis of CMF relied on prenatal ultrasound or clinical assessment by experienced neonatologists.
Results: Folic acid intake and consanguinity was the most common in congenital malformations as 30.9% and 26.5%, respectively. Out of 68 malformations, 45.6% were CNS anomalies followed by hydrocephalus 19.1% infants. Musculoskeletal anomalies were found in 17.6% patients, followed by talipeseqinovarus 7.4% infants. Gastrointestinal anomalies were found in 19.1% infants, followed by fetal ascites 7.4% infants. Renal anomalies were found in 17.6% infants, followed by multicystic/dysplastic kidneys 10.3% infants.
Conclusion: The study highlights consanguineous marriages and insufficient folic acid intake as primary risk factors, advocating for increased awareness on folic acid intake during conception and early pregnancy, avoidance of cousin marriages, and universal antenatal care to aid in prevention, timely diagnosis, and management of congenital anomalies.
Keywords: Congenital malformation, Risk factors, Surveillance, Genetic disorder, Prevalence

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Introduction

Congenital malformations of the fetus represent disruptions in early fetal development and are emotionally challenging for both expectant mothers and obstetricians alike.[1] With improvements in medical care and nursing, infectious diseases and malnutrition have decreased, shifting the spotlight to congenital malformations, which now account for nearly 2 out of every 1000[2] infant deaths in the United States according to statistics. Incidence rates vary significantly across different ethnicities and geographical regions.[3]

In Asia, numerous population and hospital-based studies reveal a consistent prevalence of approximately 2.5%[4] of newborns presenting congenital birth defects, making it the third leading cause of perinatal mortality in the country. These anomalies, ranging from minor to major, can stem from genetic predispositions, environmental factors, or a combination thereof, posing a significant challenge to obstetricians in terms of prediction and prevention.[5]

Surveillance systems also collect information on potential risk factors associated with congenital malformations.[6] These risk factors may include maternal age, exposure to teratogenic agents (such as certain medications, chemicals, or infections) during pregnancy, genetic factors, lifestyle factors, and socioeconomic status.[7],[8] Preventing congenital anomalies involves several key interventions: maintaining a healthy diet and weight, ensuring

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sufficient intake of vitamins and minerals like folic acid, abstaining from harmful substances such as tobacco[9], avoiding infections linked to congenital anomalies, minimizing exposure to environmental hazards like heavy metals and pesticides, and limiting exposure to certain medications and radiation.[10]

This study focused on the surveillance of congenital malformations and their possible risk factors holds significant importance for public health, medical practice, and research. By providing valuable data into prevalence rates, risk factors, and potential interventions, this study will play an important role in improving the health and well-being of individuals and populations affected by congenital malformations.

Methodology

This prospective study was conducted at Tertiary care Hospital Nishtar 2, Multan from January 2023 to December 2023, and a prominent referral center in Punjab catering to patients from both government and private hospitals in the region. The study aimed to determine the incidence of Congenital Malformation of Fetuses (CMF) among 800 consecutive deliveries at the institute. CMF was operationally defined as structural abnormalities detected either at birth or within the first week of life, with major anomalies necessitating surgical or medical intervention due to serious structural, cosmetic, or functional impairment. The diagnosis of CMF relied on prenatal ultrasound or clinical assessment by experienced neonatologists. Prior to inclusion in the study, informed consent was obtained from all participating patients; ensuring ethical standards were upheld throughout the research process.

A comprehensive analysis was undertaken, incorporating an extensive array of data encompassing various factors influencing maternal and neonatal health. These factors included maternal age, parity, obstetric history, pregnancy complications, and neonatal attributes such as sex, gestational age, birth weight, and Apgar score, alongside any clinically evident malformations. Furthermore, the presence or absence of numerous risk factors, such as extremes of maternal age (≤20 or ≥35), history of congenital malformations in offspring or family, recurrent spontaneous abortions, consanguinity, alcoholism, smoking, low socioeconomic status, maternal illnesses during pregnancy, chronic conditions necessitating prolonged medication, oligohydramnios, polyhydramnios, intrauterine growth restriction, uncontrolled periconceptional diabetes mellitus, and exposure to infections or teratogens like rubella or CMV, were meticulously documented for further analysis.

Exposure histories to radiation, chemicals, and fever were documented, with anomalies classified according to the International Classification of Diseases (ICD-10) codes. Karyotyping wasn't feasible due to its unavailability and costliness, particularly challenging for patients of low socioeconomic status. Socioeconomic status was evaluated via Kuppuswami's criteria. Autopsy examinations were forgone due to lack of parental consent. Data analysis utilized SPSS 15, with incidences of total and system-specific malformations calculated. The strength of associations between factors was determined through chi-square methods and odds ratios, with significance set at $p < 0.05$.

Results

Among total 800 deliveries, congenital malformations were found in 68 (8.5%) infants. The demographic characteristics of mother and infant were almost identical in congenital malformation and non-congenital malformation, (p>0.050). (Table. I).

Association of congenital malformation and demographic characteristics of mother and infant was shown in table. II.

Table I: Association of congenital malformation and demographics of the mother & infant.

Demographics	Congenital Malformation		p-value
	Yes n (%)	No n (%)	
Mother			
	Mean±SD		
Age (years)	29.09±6.15	28.85±5.28	0.727
Area of residence			
Urban	32 (45.6)	279 (38.1)	0.226
Rural	37 (54.4)	453 (61.9)	
Antenatal care	33 (48.5)	368 (50.3)	0.783
Gender of Infant			
Male	28 (41.2)	343 (46.9)	0.369
Female	40 (58.8)	389 (53.1)	
Birth weight			
<2.5 kg	8 (11.8)	118 (16.1)	0.346
≥25 kg	60 (88.2)	614 (83.9)	
Period of gestation			
<14 weeks	8 (11.8)	68 (9.3)	0.098
14-28 weeks	32 (47.1)	406 (55.1)	
29-36 weeks	14 (20.6)	81 (11.1)	
≥37 weeks	14 (20.6)	180 (24.6)	

Folic acid intake and consanguinity was the most common in congenital malformations as 21 (30.9%) and 18 (26.5%), respectively (Table. II). Out of 68 malformations, 31 (45.6%) were CNS anomalies followed by hydrocephalus 13 (19.1%) infants. Musculoskeletal anomalies were found in 12 (17.6%) patients, followed by talipeseqinovarus 5 (7.4%) infants. Gastrointestinal anomalies were found in 13 (19.1%) infants, followed by fetal ascites 5 (7.4%) infants. Renal anomalies were found in 12 (17.6%) infants, followed by multicystic/dysplastic kidneys 7 (10.3%) infants. (Table. III).

Table II: Risk factors of congenital malformation.

Risk factor	Congenital Malformation		p-value
	Yes n(%)	No n(%)	
Consanguinity	18 (26.5)	177 (24.2)	0.674
Folic acid intake	21 (30.9)	236 (32.2)	0.819
History of miscarriages	8 (11.8)	86 (11.7)	0.997
Maternal medical illness	5 (7.4)	60 (8.2)	0.808
History of drug intake	11 (16.2)	90 (12.3)	0.357
History of congenital anomalies	14 (20.6)	121 (16.5)	0.393
History stillbirth/neonatal death	17 (25.0)	204 (27.9)	0.613
History Radiation exposure	11 (16.2)	128 (17.5)	0.785

Table III: Anomaly types.

Anomaly type	N	%
CNS Anomalies	31	45.6
Hydrocephalus	13	19.1
Anencephalus	6	8.8
Meningomyelocele	5	7.4
Encephalocele	3	4.4
Hydroceph+ Meningomyelocele	4	5.9
Microcephaly	0	0.0
Musculoskeletal Anomalies	12	17.6
Talipeseqinovarus	5	7.4
Polydactyly	3	4.4
Achondroplasia	4	5.9
Gastrointestinal Anomalies	13	19.1
Omphalocoele	4	5.9
Gastroschisis	4	5.9
Fetal ascites	5	7.4
Renal Anomalies	12	17.6
Polycystic kidneys	5	7.4
Multicystic/Dysplastic kidneys	7	10.3

Discussion

In developed countries, special programs have been designed to estimate the prevalence of congenital anomalies, which occur in 2-3% of births, and to prevent them through timely diagnosis and intervention. These programs aim to detect anomalies early, either shortly after birth or later, to ensure prompt medical attention and intervention when necessary.[11]

The prevalence of congenital anomalies at our hospital was found to be 8.5%, per 800 total births, which aligns closely with findings from other studies. For instance, Qadir M et al[12] reported 1.23% congenital anomalies in Mardan medical complex, while Madi et al[13] reported an incidence of 1.25% in their study conducted in Kuwait.

Similarly, researchers in Iran documented a rate of 1.9%, and a study in Iran reported a prevalence of 1.4%.[14] These findings collectively underscore the importance of monitoring and understanding the occurrence of congenital anomalies across diverse populations and healthcare settings.

In our study, a notable pattern emerged wherein the predominant anomalies observed were related to the central nervous system (45.6%), with musculoskeletal anomalies following closely behind (16.6%). This observation aligns with findings reported by Gul F et al[15] in their research conducted in Kohat, Khyber Pakhtoonkhwa. Moreover, this order of affected systems was similarly noted by Karim et al[16] in their respective studies, thereby providing further evidence for the consistency and robustness of these findings across different research endeavors.

The findings of our study reveal that lack of folic acid intake (30.9%), followed by consanguinity (26.5%) emerges as the foremost risk factor contributing to the occurrence of congenital anomalies. Notably, our country exhibits alarmingly high rates, with consanguineous marriages accounting for a staggering 61% of unions. This figure surpasses those reported by Nasreen et al[17], who documented a consanguinity rate of 46% in their study. Furthermore, research conducted by Sheridan et al[18] in Bradford underscores the severity of this issue, demonstrating a doubling of the risk of congenital anomalies associated with consanguineous marriages. These findings underscore the critical importance of addressing consanguinity as a significant factor in mitigating the incidence of congenital anomalies within our population.

Maternal age is also a contributing factor as in our study mean age of patients was 29.09±6.15 years. Tootoonchi et al[19] reported an average age of 25.69 years with a standard deviation of 5.54 years in their study. Additionally, Tomatyr AG et al[20] found that 8.7% of their subjects were over 35 years old.

In our study, it was observed that 11.8% of babies had a birth weight of less than 2.5kg. This finding is consistent with two local studies conducted in Abbottabad[21] and Kohat[15], which reported similar results. It is well-established that there is an association between congenital anomalies and intrauterine growth restriction (IUGR), with low birth weight often being indicative of this condition. Furthermore, IUGR can potentially act as both a cause and an effect of congenital anomalies, or they may co-exist concurrently. This underscores the intricate relationship between fetal development, intrauterine conditions, and the occurrence of congenital abnormalities, highlighting the importance of further research and clinical vigilance in this area.[21]

Conclusion

The study highlights consanguineous marriages and insufficient folic acid intake as primary risk factors, advocating for increased awareness on folic acid intake during conception and early pregnancy, avoidance of cousin marriages, and universal antenatal care to aid in prevention, timely diagnosis, and management of congenital anomalies.

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