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Frequency of Various Congenital Anomalies among Neonates Born at a Tertiary Care Hospital of Karachi

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ABSTRACT

Objective: To determine the frequency of various congenital abnormalities in neonates born at tertiary care hospitals. **Methodology:** This cross sectional study was conducted at the Department of Obstetrics and Gynaecology at Jinnah Postgraduate Medical Centre (JPMC), Karachi from 11 July 2024 to 11 Jan 2025. Comprehensive data on each neonate were collected, including gender, maternal age, and maternal BMI, considering that both low and high BMI are risk factors for congenital anomalies. Additional data such as parity, booking status, residential status, maternal comorbidities (diabetes and hypertension), and gestational age were documented. **Results:** Most patients 66.4% had 1-5 children, 18.9% had nulliparous (0 children), and 14.7% had more than five children. The majority 78.3% was un-booked cases, and 21.7% were booked, according to the mode of admission. Hydrocephalus was the most common condition at 25.2%, followed by anencephaly at 16.8% and spina bifida at 14.0%. Meningocele was observed in 13.3% of patients, while microcephaly and cleft lip were less frequent, occurring in 10.5% and 8.4% of patients, respectively. **Conclusion:** The study found a high prevalence of congenital anomalies, with hydrocephalus being the most common. Maternal hypertension was linked to hydrocephalus, microcephaly, and cleft lip. Diabetes showed a strong association with meningocele and microcephaly. Consanguinity was a key factor in spina bifida, meningocele, and microcephaly.

INTRODUCTION

Human development is an intricate and elaborate process, from a single-cell zygote to a complex multicellular organism. Fortunate are those fetuses that navigate this journey without any obstacles. The birth of a malformed baby is a tragic event for any family and society as a whole[1]. The influence of teratogens, such as pathogens, extensive chemical exposure, environmental pollution, and the indiscriminate use of drugs by mothers in their daily lives, has led to an increased incidence of congenital abnormalities in newborn children. Congenital anomalies are significant causes of prenatal mortality and morbidity. Consequently, antenatal diagnosis and fetal therapy have become crucial in the field of human embryology, as noted by Dolk[2].

Congenital anomalies significantly impact infant mortality rates across various socio-economic backgrounds globally. Each year, approximately 7.9 million children, accounting for 6% of total births worldwide, are born with severe congenital anomalies of hereditary origin[3]. In the United States, these conditions are observed in 3% to 5%[3] of live births. In comparison, Europe reports a lower prevalence of 2.1%, according to data from EUROCAT, the European Network of Congenital Anomaly Registers[4]. In India, congenital anomalies contribute to 8% to 15% of perinatal deaths and 13% to 16%[5] of neonatal deaths. In Pakistan, they account for about 6% to 9% of perinatal deaths[6]. Among these congenital anomalies, approximately 40% to 60% are of unknown aetiology, 20% are attributed to a combination of hereditary and environmental factors, 7.5% to single-gene mutations, 6% to chromosomal abnormalities, and another 5% to maternal illnesses such as diabetes mellitus or infections[7]. Additionally, low socio-economic status and low literacy rates are

significant contributors to the prevalence of congenital anomalies in a population[8].

A study conducted in Peshawar 2015 aimed to determine the frequency of various congenital abnormalities in neonates. It found that out of 1,062 deliveries, 31 newborns (2.9%) exhibited congenital anomalies. The most common congenital anomalies identified were hydrocephalus (22.6%), anencephaly (12.9%), spina bifida (9.7%), and meningocele, microcephaly, and cleft lip, each at 6.5%[9]. Similarly, a study conducted in a tertiary care hospital in Abbottabad reported anencephaly as the most prevalent congenital anomaly, observed in 10% of neonates, followed by spina bifida at 8.4%, myelomeningocele at 5.7%, and hydrocephalus at 4.03%[10].

Our study aims to determine the frequency of congenital anomalies among neonates born in a tertiary care hospital. Past studies have shown varying trends in congenital abnormalities, which may be attributed to differences in population dynamics, pregnancy-related complications, and the utilization of antenatal services[11]. In recent years, congenital anomalies have become a significant concern for neonatologists and paediatricians. The findings of our study will not only assist policymakers in allocating resources for the management of common anomalies. Still, they will also provide insights for neonatologists and paediatricians to investigate further the factors associated with different congenital anomalies in neonates.

METHODOLOGY

This cross sectional study was conducted at the Department of Obstetrics and Gynaecology at Jinnah Postgraduate Medical Centre (JPMC), Karachi from 11th July 2024 to 11th January 2025. The sample size was determined using the OpenEpi sample size calculator based on a prevalence rate of anencephaly of 12.9%, with a margin of error of 5.5% and a confidence level of 95%. This resulted in a minimum required sample size of 143 neonates. A non-probability consecutive sampling technique was used for participant selection.

Neonates delivered at JPMC, Karachi, from the start of the study until the required sample size was achieved were included, regardless of gender. Exclusion criteria included parents or guardians unwilling to participate and neonates transferred from other hospitals. The study commenced following approval of the synopsis by the Research Department of CPSP. Mothers of infants who met the eligibility criteria were included in the study after obtaining informed consent from parents or guardians.

Comprehensive data on each neonate were collected, including gender, maternal age, and maternal BMI, considering that low and high BMI are risk factors for congenital anomalies. Additional data such as parity, booking status, residential status, maternal comorbidities (diabetes and hypertension), and gestational age were documented using a specially designed proforma.

Patient data were entered and analyzed using SPSS version 26.0. Mean and SD were calculated for continuous variables such as maternal age, BMI, and gestational age. Frequencies and percentages were calculated for categorical variables, including the baby's gender, consanguinity, parity, booking status, residential status, maternal comorbidities (diabetes and hypertension), and congenital anomalies. The Chi-square was applied to see the association among variables. A p-value of ≤0.05 was considered statistically significant.

RESULTS

This study included one hundred forty-three patients who met the inclusion criteria. The average age of patients was 26.64±4.90 years, with an average gestational age of 38.28±2.45 weeks. The average height was 1.44±0.26 meters, weight was 65.41±5.16 kg, and BMI was 34.85±12.23 kg/m². Most patients had (66.4%) 1-5 children, 18.9% had nulliparous (0 children), and 14.7% had more than five children. The majority (78.3%) was un-booked cases, and 21.7% were booked, according to the mode of admission. Regarding residential status, 53.1% were from rural areas, and 46.9% were from urban areas. Regarding health conditions, 14.7% had maternal diabetes mellitus, and 34.3% had maternal hypertension. A high proportion (73.4%) reported consanguinity (blood relation between parents). The gender distribution of babies was nearly equal, with 47.6% male and 52.4% female. (Table. I).

Hydrocephalus was the most common condition at 25.2%, followed by anencephaly at 16.8% and spina bifida at 14.0%. Meningocele was observed in 13.3% of patients, while microcephaly and cleft lip were less frequent, occurring in 10.5% and 8.4% of patients, respectively. (Figure. I).

The association between congenital outcomes (meningocele, spina bifida, hydrocephalus, anencephaly, cleft lip, and microcephaly) and baseline factors were shown in the table. II. It was seen that most of the patients of meningocele (57.9%), spina bifida (55.0%), and hydrocephalus (75.0%) occurred in women with 1-5 children. Hypertension was associated with 38.9% of hydrocephalus patients, 40.0% of microcephaly patients, and 41.7% of cleft lip patients. Maternal diabetes was linked to 26.3% of meningocele patients and 20.0% of microcephaly patients. Consanguinity was prevalent in 90.0% of spina bifida patients, 81.3% of meningocele patients, and 66.7% of microcephaly patients. The chi-square test showed significant associations ($p < 0.01$) for all variables, suggesting these factors may influence the prevalence of these conditions. (Table. II).

Table I

Demographics and baseline profile

Variable	Mean±S.D	N (%)
Age (years)	26.64±4.90	
Gestational age (weeks)	38.28±2.45	
Height (m)	1.44±0.26	
Weight (kg)	65.41±5.16	
BMI (kg/m^2)	34.85±12.23	
Parity		
0		27 (18.9)
1-5		95 (66.4)
>5		21 (14.7)
Mode of admission		
Booked		31 (21.7)
Un-booked		112 (78.3)
Residential status		
Urban		67 (46.9)
Rural		76 (53.1)
Maternal diabetes mellitus		
Yes		21 (14.7)
No		122 (85.3)
Maternal hypertension		
Yes		49 (34.3)
No		94 (65.7)
Consanguinity		
Yes		105 (73.4)
No		38 (26.6)
Gender of baby		
Yes		68 (47.6)
No		75 (52.4)

Figure I

Prevalence of outcome among study patients

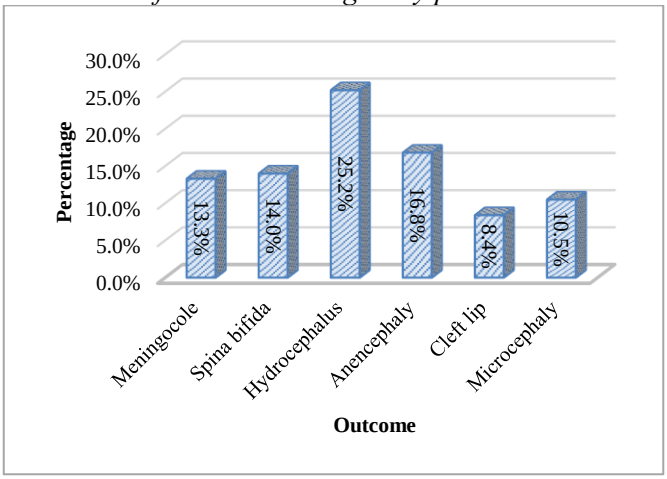

Table II

Association between outcomes and baseline profile

Variable	Outcomes						p-value
	Meningocole n=19	Spina bifida n=20	Hydrocephalus n=36	Anencephaly n=24	Cleft lip n=12	Microcephaly n=15	
Parity							
0	5 (26.3)	7 (35.0)	6 (16.7)	6 (25.0)	2 (16.7)	6 (40.0)	<0.001*
1-5	11 (57.9)	11 (55.0)	27 (75.0)	13 (54.2)	8 (66.7)	6 (40.0)	
>5	3 (15.8)	2 (10.0)	3 (8.3)	5 (20.8)	2 (16.7)	3 (20.0)	
Hypertension							
Yes	6 (31.6)	6 (30.0)	14 (38.9)	7 (29.2)	5 (41.7)	6 (40.0)	<0.001*
No	13 (68.4)	14 (70.0)	22 (61.1)	17 (70.8)	7 (58.3)	9 (60.0)	
Diabetes							
Yes	5 (26.3)	2 (10.0)	6 (16.7)	4 (16.7)	1 (8.3)	3 (20.0)	<0.001*
No	14 (73.7)	18 (90.0)	30 (83.3)	20 (83.3)	11 (91.7)	12 (80.0)	
Consanguinity							
Yes	13 (81.3)	18 (90.0)	22 (61.1)	15 (62.5)	6 (50.0)	10 (66.7)	<0.001*
No	6 (18.8)	2 (10.0)	14 (38.9)	9 (37.5)	6 (50.0)	5 (33.3)	

N (%), chi-square test for association was applied. *Significant p-value

DISCUSSION

The study revealed that the overall prevalence of congenital anomalies was comparable to previous reports from similar tertiary care settings. The most commonly observed anomalies were neural tube defects, congenital heart defects, musculoskeletal abnormalities, and gastrointestinal malformations. These findings align with studies conducted in other developing countries, where such anomalies are frequently reported due to genetic predisposition, environmental exposures, and nutritional deficiencies[12].

Neural tube defects have been strongly associated with maternal folate deficiency, which remains a significant concern in low-resource settings[13]. Similarly, congenital heart defects are among the most prevalent structural anomalies, often linked to multifactorial causes, including maternal infections and exposure to teratogens[14].

Hydrocephalus was this study's most common congenital condition, affecting 25.2% of cases. Similarly, a study conducted by Khan et al[9] in Peshawar reported hydrocephalus in 22.6% of newborns with congenital anomalies, indicating a consistent prevalence in different regions. Another study conducted by Sial et al[15] reported 33.3% anomalies in the central nervous system.

Similarly, findings showed anencephaly in 16.8% and spina bifida in 14.0% of cases. An Egyptian study by Shawky et al[16] reported slightly lower figures, with anencephaly at 12.9% and spina bifida at 9.7%. These differences might be attributed to regional variations or sample sizes.

The occurrence of cleft lip in this study was 8.4%. Another survey from interior Punjab conducted by Langah et al[17] found cleft palate to be the most common anomaly, followed by cleft lip. This suggests that orofacial clefts are among the region's most prevalent congenital anomalies, about 6%.This study identified maternal hypertension in 34.3% of cases, with significant associations with hydrocephalus (38.9%), microcephaly (40.0%), and cleft lip (41.7%). This aligns with global research conducted by Bellizzi et al[18], indicating that chronic hypertension during pregnancy increases the risk of congenital malformations, including renal, limb, and orofacial defects, by up to 4.3%.

In our study, most women were multipara and multigravida, which aligns with findings by Qazi et al[19], who reported that two out of three congenital malformations in newborns were associated with maternal multigravidity. However, this contrasts with the study by Perveen et al[20], which demonstrated a higher incidence of congenital anomalies in primigravida mothers.

CONCLUSION

The study found a high prevalence of congenital anomalies, with hydrocephalus being the most common. Maternal hypertension was linked to hydrocephalus, microcephaly, and cleft lip. Diabetes showed a strong association with meningocele and microcephaly. Consanguinity was a key factor in spina bifida, meningocele, and microcephaly.

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