



Frequency of Congenital Anomalies in Newborns of Women having Age above 35 Years of Age

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ABSTRACT

Objective: To determine the frequency of various congenital abnormalities in newborns born to mothers having age above 35 years of age. **Methodology:** This cross-sectional study was conducted at the Department of Obstetrics and Gynaecology at Jinnah Post graduate Medical College, Karachi from 5th of August 2023 to 31st January 2024. 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:** Among patients with no previous births, 24.6% had congenital heart defects, 11.1% had Down syndrome, 15.2% had hydrocephalus, 17.2% had anencephaly, and 21.2% had cleft lip and cleft palate. In those with 1-5 births, congenital heart defects were observed in 59.4%, Down syndrome in 77.8%, hydrocephalus in 66.7%, anencephaly in 62.1%, and cleft lip and cleft palate in 61.6%. Patients with more than five births had lower frequencies of these anomalies. **Conclusion:** Congenital heart defects were the most commonly observed anomalies in 81.2% of newborns followed by cleft lip 58.2% born to mothers aged over 35 years. Early prenatal diagnosis of these abnormalities plays an important role in reducing perinatal mortality by enabling timely medical intervention, parental counseling, and, when necessary, the option of early termination of pregnancy to prevent complications associated with severe congenital defects.

INTRODUCTION

Congenital anomalies are structural or functional defects present at birth, contributing to neonatal morbidity and mortality. Advanced maternal age ≥ 35 years is a significant risk factor, increasing the likelihood of chromosomal abnormalities e.g., Down syndrome, congenital heart defects, and neural tube defects. Additionally, maternal conditions such as hypertension, gestational diabetes, and placental abnormalities further elevate fetal risks^{1,2}.

Globally, congenital anomalies affect approximately 3–6%³ of live births, with higher prevalence in low-resource settings due to limited prenatal care. The incidence of Down syndrome rises sharply with maternal age, from 1 in 1,250 at age 25 to 1 in 100 at age 40^{4,5}. Given the global trend of delayed childbearing, early screening, prevention, and management are essential for improving neonatal outcomes.

Congenital anomalies significantly impact infant mortality rates across various socio-economic backgrounds globally. In Pakistan, they account for about 6% to 9% of perinatal deaths⁶. 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 infections⁷. Additionally, low socio-economic status and low literacy rates are significant contributors to the prevalence of congenital anomalies in a population⁸.

Previous literature reported 2.9% exhibited congenital anomalies hydrocephalus 22.6%, anencephaly 12.9%, spina bifida 9.7%, and meningocele, microcephaly, and cleft lip, each at 6.5%⁹. Similarly, another study 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 newborns born to mothers having age above 35 years. 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 services¹¹. In recent years, congenital anomalies have become a

significant concern for neonatologists and pediatricians. 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 pediatricians 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 Post graduate Medical College, Karachi from 5th of August 2023 to 31st January 2023. The sample size was determined using the WHO sample size calculator based on a prevalence rate of Down Syndrome of 7.61%, with a margin of error of 4% and a confidence level of 95%. This resulted in a minimum required sample size of 170 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.

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

The study included 170 patients, with an average age of 47.11 ± 5.58 years and a mean gestational age of 38.26 ± 2.48 weeks. Most patients had a BMI of 34.36 ± 12.38 kg/m² and were predominantly multiparous 106 (62.4%). The majority 132 (77.6%) were un-booked admissions, and 115 (67.6%) had vaginal deliveries. Rural residents made up 104 (61.2%) of the sample, and 113 (66.5%) presented through OPD. Diabetes mellitus

was present in 52 (30.6%) of the patients. Regarding education, 75 (44.1%) had primary-level education, while 39 (22.9%) were illiterate. Economic status distribution showed 32 (18.8) in the upper class, 56 (32.9%) in the lower class, and 82 (48.2%) in the middle class. (Table 1).

Table 1

Demographics and Baseline Profile

Variable	Mean \pm S.D	N (%)
Age (years)	47.11 \pm 5.58	
Gestational age (weeks)	38.26 \pm 2.48	
Height (m)	1.45 \pm 0.26	
Weight (kg)	65.42 \pm 5.06	
BMI (kg/m ²)	34.36 \pm 12.38	
Parity		
0		38 (22.4)
1-5		106 (62.4)
>5		26 (15.3)
Mode of admission		
Booked		38 (22.4)
Un-booked		132 (77.6)
Mode of delivery		
Vaginal		115 (67.6)
C-section		55 (32.4)
Residential status		
Urban		66 (38.8)
Rural		104 (61.2)
Patient presenting		
Emergency		57 (33.5)
OPD		113 (66.5)
Maternal diabetes mellitus		
Yes		52 (30.6)
No		118 (69.4)
Educational Status		
Illiterate		39 (22.9)
Primary		75 (44.1)
Secondary		25 (14.7)
Matric or above		31 (18.2)
Economic Status		
Lower		56 (32.9)
Middle		82 (48.2)
Upper		32 (18.8)

The prevalence of congenital anomalies among the study patients showed that congenital heart defects were the most common, affecting 81.2% of cases. Cleft lip and cleft palate were observed in 58.2% of patients, while hydrocephalus was present in 19.4%. Anencephaly was found in 17.1% of cases, and Down syndrome had the lowest prevalence at 5.3%. (Figure 1).

Among patients with no previous births, 24.6% had congenital heart defects, 11.1% had Down syndrome, 15.2% had hydrocephalus, 17.2% had anencephaly, and 21.2% had cleft lip and cleft palate. In those with 1-5 births, congenital heart defects were observed in 59.4%, Down syndrome in 77.8%, hydrocephalus in 66.7%, anencephaly in 62.1%, and cleft lip and cleft palate in 61.6%. Patients with more than five births had lower frequencies of these anomalies. Vaginal delivery was associated with 65.9% of congenital heart defect cases, 66.7% of Down syndrome cases, 66.7% of hydrocephalus cases, 62.1% of anencephaly cases, and

68.7% of cleft lip and cleft palate cases. Cesarean delivery had lower percentages across all anomalies. Among diabetic patients' congenital heart defect was present in 70.3% of patients, down syndrome in 55.6%, hydrocephalus in 72.7%, anencephaly in 69% and cleft lip and cleft palate in 69.7% patients, while non-diabetic patients had lower frequencies of all anomalies. All associations were statistically significant with a p-value of <0.001. (Table 2).

Figure 1
Prevalence of Outcome among Study Patients

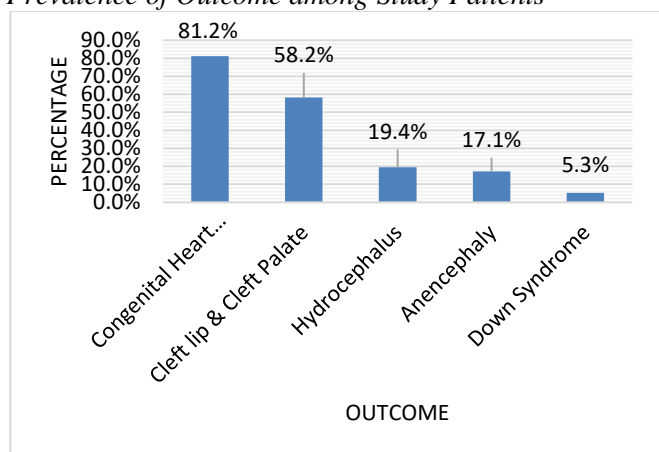


Table 2
Association between Outcomes and Baseline Profile

Variable	Outcomes					p-value
	Congenital Heart Defect	Down Syndrome	Hydrocephalus	Anencephaly	Cleft lip & Cleft Palate	
Parity						
0	34 (24.6)	1 (11.1)	5 (15.2)	5 (17.2)	21 (21.2)	
1-5	82 (59.4)	7 (77.8)	22 (66.7)	18 (62.1)	61 (61.6)	<0.001*
>5	22 (15.9)	1 (11.1)	6 (18.2)	6 (20.7)	17 (17.2)	
Mode of delivery						
Vaginal	91 (65.9)	6 (66.7)	22 (66.7)	18 (62.1)	68 (68.7)	<0.001*
c-section	47 (34.1)	3 (33.3)	11 (33.3)	11 (37.9)	31 (31.3)	
Diabetes						
Yes	97 (70.3)	5 (55.6)	24 (72.7)	20 (69.0)	69 (69.7)	<0.001*
No	41 (29.7)	4 (44.4)	9 (27.3)	9 (31.0)	30 (30.3)	

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

DISCUSSION

The prevalence of congenital anomalies among the study patients indicated that congenital heart defects were the most commonly observed anomaly, affecting 81.2% of the cases. Cleft lip and cleft palate were also frequently noted, with a prevalence of 58.2% among the patients.

Additionally, hydrocephalus was identified in 19.4% of the cases, while anencephaly was present in 17.1% of the study population. When comparing our findings with other studies, a study conducted by Perveen et al¹³ in Kohat reported that the overall frequency of congenital malformations was 0.97%. In contrast, research conducted in Karachi by Taksande et al¹⁴ observed a higher prevalence, reporting a frequency of 11.4 per 1000 live births. Similarly, a study from India conducted by Movafagh et al¹⁵ estimated the frequency of congenital anomalies at 1.91%, highlighting some regional variations in prevalence rates.

Our study results align closely with a study from Iran conducted by Lowry et al¹⁶, which documented a congenital anomaly frequency of 29.4 per 1000 live births. Furthermore, our findings are also comparable to research conducted by Tomatir et al¹⁷ in Canada, which reported a slightly higher frequency of 36.18 per 1000 live births. These comparisons emphasize the variations in congenital anomaly prevalence across different populations and geographic regions while also highlighting the similarities between our study and international research.

Congenital Heart defect was this study's most common congenital condition, affecting 81.2% of cases. Hydrocephalus was found in 19.4%. Similarly, a study conducted by Khan et al⁹ 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 al¹⁸ 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 al¹⁹ 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. In this study anencephaly was observed in 17.1% of newborns.

The occurrence of cleft lip in this study was 58.2%. Another survey from interior Punjab conducted by Langah et al²⁰ 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 al²¹, 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 al²², 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 al¹³, which demonstrated a higher incidence of congenital anomalies in primigravida mothers.

CONCLUSION

Congenital heart defects were the most commonly observed anomalies in 81.2% of newborns followed by

cleft lip 58.2% born to mothers aged over 35 years. Early prenatal diagnosis of these abnormalities plays an important role in reducing perinatal mortality by enabling timely medical intervention, parental counseling, and, when necessary, the option of early termination of pregnancy to prevent complications associated with severe congenital defects.

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