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Frequency and Spectrum of Congenital Abnormalities Diagnosed on Ultrasound Scan in a Tertiary Care Hospital

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ABSTRACT

Background: Congenital anomalies represent a major cause of perinatal morbidity and mortality worldwide. Timely detection during routine prenatal care allows for better counseling, management, and improved outcomes. Ultrasound serves as the primary screening modality, yet prevalence patterns vary across regions and populations. **Objective:** To determine the frequency and types of congenital anomalies diagnosed on ultrasound scan. **Study Design:** Cross-sectional study. **Duration and Place of Study:** The study was conducted from May 2024 to October 2024 at the Department of Obstetrics and Gynaecology, Lady Reading Hospital, Peshawar. **Methodology:** A total of 225 pregnant women, aged 18–50 years, with singleton pregnancies between 18 and 20+6 weeks of gestation were enrolled through consecutive sampling. Women with epilepsy or unwilling to participate were excluded. After informed consent, demographic details were recorded, followed by detailed history, examination, and targeted ultrasound by a consultant gynecologist. Congenital anomalies were categorized into central nervous system, urinary, skeletal, gastrointestinal, and cardiovascular groups based on established sonographic criteria. **Results:** Congenital anomalies were detected in 15 women (6.7%), while 210 (93.3%) had normal findings. Central nervous system anomalies were most frequent (33.3%), followed by urinary (20.0%), gastrointestinal (20.0%), skeletal (13.3%), and cardiovascular anomalies (13.3%). Maternal age showed no significant association (p=0.594), with anomalies present in 7.6% of mothers ≤30 years and 5.3% in mothers >30 years. Parity was also non-significant (p=0.381). Gestational age was significantly associated (p=0.007), with no anomalies detected ≤19 weeks, compared to 9.6% >19 weeks. **Conclusion:** Ultrasound screening in the second trimester is a valuable tool for identifying congenital anomalies, with the central nervous system most frequently affected.

INTRODUCTION

Congenital abnormalities, are structural or functional anomalies that occur during intrauterine development and are present at birth.[1] These abnormalities can affect any part of the body, including the heart, brain, limbs, or internal organs, and may vary in severity from minor defects with minimal clinical significance to major malformations that can impact survival or quality of life.[2] Causes of congenital abnormalities are multifactorial, encompassing genetic mutations, chromosomal abnormalities, environmental exposures, maternal infections, nutritional deficiencies, and certain medications taken during pregnancy.[3] Early identification of these anomalies is essential for appropriate prenatal counseling, management, and planning for postnatal care.[4] The epidemiology of congenital abnormalities encompasses the examination of their prevalence, nature, and spread within afflicted groups.[5] Some congenital anomalies are more frequent than others; e.g., cardiac defects, neural tube defects, cleft lip and palate, and limb malformations are frequently noted.[6] Epidemiological studies revealed that the occurrence of certain abnormalities has been found to differ with maternal age, genetics, environmental influences, and geographic location.[7] Identification of the most common congenital anomalies assists the clinician in tailoring screening agendas, planning preventive measures, and improving maternal-fetal outcomes.

Ultrasound scanning has also been the gold standard for prenatal identification of congenital anomalies based on its safety, availability, and real-time assessment abilities.[8] Routine prenatal ultrasonography, such as the first-trimester nuchal translucency screen and detailed mid-trimester anomaly ultrasonography, can be used to identify structural anomalies like cardiac malformations, neural tube defects, renal malformations, and skeletal dysplasias.[9] A high percentage of prevailing congenital anomalies, like ventriculomegaly, spina bifida, congenital

heart malformations, and cleft lip/palate, has been reliably identified through ultrasonography, based on studies. Early and precise identification makes it possible to conduct timely interventions, parental consultation, and educated decision-making on management of the fetus.[10]

In a study by Babu RS, et al. has shown that frequency of congenital anomalies was 3.8% diagnosed on ultrasound scan, further analysis of these anomalies was central nervous system 45.94%, urinary system 21.62%, skeletal system 8.10%, gastrointestinal tract 16.22% and cardiovascular system was 8.10%.[11]

Congenital anomalies are still an important cause of neonatal morbidity and mortality, though local information on their patterns and prevalence in Peshawar is scarce. This work will be valuable in giving evidence on the common congenital anomalies diagnosed through ultrasound in the area, enabling caregivers to enhance early detection, counseling, and management. The information can also inform public health policy, prenatal screening programs, and allocation of resources toward decreasing the impact of congenital disorders in Peshawar.

METHODOLOGY

This cross-sectional study was conducted from May 2024 to October 2024 in the Department of Obstetrics and Gynaecology at LRH Peshawar. Ethical approval for the study was obtained from the institutional review board prior to initiation. A total of 225 pregnant women were enrolled, with the sample size calculated using WHO sample size software, considering a 95% confidence level, a 2.5% margin of error, and an expected frequency of congenital anomalies of 3.8% detected on ultrasound.[11]

Women aged 18 to 50 years with a singleton pregnancy between 18 and 20+6 weeks of gestation, regardless of parity, who presented for routine antenatal care were included. Patients with a history of epilepsy or those who declined participation were excluded. Written informed consent was obtained from all participants after explaining the purpose of the study, ensuring confidentiality, and clarifying that the procedure posed no risk to mother or fetus. Demographic details including maternal age, gestational age, and parity were recorded for each participant.

A detailed clinical history and examination were performed, followed by a comprehensive ultrasound assessment conducted by a consultant gynecologist with more than three years of post-fellowship experience. Congenital abnormalities were identified and documented according to predefined criteria. Central nervous system anomalies were considered present if there was significant dilatation of all cerebral ventricles (transverse diameter of the third ventricle ≥7 mm) or disproportionate widening of basal and Sylvian CSF fissures. Urinary system anomalies included absent kidney, absent ipsilateral renal artery, or compensatory hypertrophy of the contralateral kidney. Skeletal abnormalities were defined by severe or moderate shortening of bones, limb bowing, focal spinal hypomineralization, cloverleaf skull with micromelia, small thorax, rib fractures with bell-shaped thorax, or polydactyly associated with skeletal dysplasia. Gastrointestinal tract anomalies were noted in cases of polyhydramnios, reduced intraluminal fluid, non-visualization of the stomach bubble, or inability to pass a feeding tube into the stomach. Cardiovascular defects were considered present if ventricular septal defects or transposition of the great arteries were identified on standard views. The prevalence of congenital abnormalities and their distribution across these systems were recorded on a structured proforma.

Data were analyzed using IBM SPSS version 26. Continuous variables such as maternal age, gestational age, and parity were presented as mean ± standard deviation. Frequencies and percentages were calculated for categorical variables, including the overall presence of congenital abnormalities and their classification into central nervous system, urinary system, skeletal system, gastrointestinal tract, and cardiovascular anomalies. Stratification for maternal age, gestational age, and parity was performed, and post-stratification chi-square tests were applied, with a p-value of ≤0.05 considered statistically significant.

RESULTS

The study included 225 participants with a mean age of 29.99±3.15 years, mean gestational age of 20.00±1.09 weeks, and mean parity of 2.06±0.90 (as shown in Table 1).

Table 1

Patient Demographics

Demographics	Mean ± SD
Age (years)	29.99±3.15
Gestational Age (weeks)	20.00±1.09
Parity	2.06±0.90

Overall, congenital anomalies were detected in 15 patients (6.70%) while 210 patients (93.30%) had no anomalies detected (as shown in Table-II). Among the 15 patients with congenital anomalies, the central nervous system was the most commonly affected system accounting for 5 cases (33.30%), followed by urinary system and gastrointestinal tract each with 3 cases (20.00% each), while skeletal system and cardiovascular system were each affected in 2 cases (13.30% each) (as shown in Table 2).

Table 2

Frequency and Spectrum of Congenital Abnormalities Diagnosed on Ultrasound Scan

Congenital Anomalies	Frequency	Percentage
Overall Presence		
Yes	15	6.70%
No	210	93.30%
Total	225	100.00%
Type of Anomalies (n=15)		
Central Nervous System	5	33.30%
Urinary System	3	20.00%
Gastrointestinal Tract	3	20.00%
Skeletal System	2	13.30%
Cardiovascular System	2	13.30%
Subtotal	15	100.00%

When examining the association between congenital anomalies and demographic factors, maternal age demonstrated no significant association, with 10 cases (7.6%) occurring in mothers ≤30 years compared to 121

cases (92.4%) without anomalies in the same age group, while 5 cases (5.3%) occurred in mothers >30 years versus 89 cases (94.7%) without anomalies (p=0.594) (as shown in Table-III). Gestational age showed a statistically significant association, with no cases (0.0%) detected at ≤19 weeks compared to 68 cases (100.0%) without anomalies, while 15 cases (9.6%) were detected at >19 weeks versus 142 cases (90.4%) without anomalies (p=0.007) (as shown in Table-III). Parity demonstrated no significant association, with 15 cases (7.2%) occurring in mothers with parity ≤3 compared to 192 cases (92.8%) without anomalies, while no cases (0.0%) occurred in mothers with parity >3 versus 18 cases (100.0%) without anomalies (p=0.381) (as shown in Table 3).

Table 3

Association of Congenital Anomalies with Demographic Factors

Demographic Factors		Congenital Anomalies		p-value
		Yes n(%)	No n(%)	
Age Group (years)	≤30	10 (7.6%)	121 (92.4%)	0.594*
	>30	5 (5.3%)	89 (94.7%)	
Gestational Age Group (weeks)	≤19	0 (0.0%)	68 (100.0%)	0.007*
	>19	15 (9.6%)	142 (90.4%)	
Parity Group	≤3	15 (7.2%)	192 (92.8%)	0.381*
	>3	0 (0.0%)	18 (100.0%)	

*Fischer Exact Test

DISCUSSION

Our results indicated an overall prevalence of 6.70% for congenital anomalies, within the normal expectation for population-based studies. The central nervous system proved the single most common system affected, accounting for one-third of all detected anomalies, in accordance with the critical period of development during which neural tube formation takes place and the extreme sensitivity of ultrasound in the detection of neural tube defects like spina bifida and anencephaly. The equal representation of urinary system and gastrointestinal tract anomalies at 20% each bears testimony to the multifarious embryological development of such systems during organogenesis, where interference in morphogenetic processes results in structural malformations. The analysis of gestational age indicated statistically significant association, with all anomalies being detected subsequently to 19 weeks of gestation, scientifically logical in view of the fact that this period corresponds with the culminating of complete organogenesis and the best period for detailed anatomical examination where fetal structures are adequately formed and amenable for precise ultrasonic examination. Lack of notable association with maternal age and also with parity points towards the possibility that advancing maternal age need be no longer such an overwhelming factor in the causation of structural anomalies, specifically considering they are much more rare compared to chromosomal abnormalities, while parity does little hold sway over occurrence of demonstrable structural malformations identifiable upon routine prenatal check-ups.

Our study results demonstrated an overall prevalence of congenital anomalies of 6.7% (15/225 cases) among participants undergoing ultrasound screening, which is higher than several comparable studies but falls within the range reported in the literature. This prevalence is substantially higher than the 3.6% reported by Hamid et al. [12] in their study of 1000 live births in Peshawar, Pakistan, and the 3% found by Onyambu CK et al. [13] in 500 low-risk pregnancies at Kenyatta National Hospital. However, our findings are more consistent with Tiwari et al. [14] who reported a 4.1% prevalence in 2000 pregnant women during routine second-trimester ultrasounds, and fall within the range of detection capabilities noted by Sylejmani et al. [15] who indicated that ultrasound can detect approximately 74% of major birth defects. The higher prevalence in our study may be attributed to differences in study population characteristics, gestational age at screening, or the sensitivity of ultrasound equipment and operator expertise.

Regarding the spectrum of anomalies detected, our study found central nervous system anomalies to be the most prevalent (33.3% of affected cases), followed by urinary system and gastrointestinal tract anomalies each occurring in 20.0% of cases, with skeletal system and cardiovascular system anomalies each representing 13.3% of detected cases. This pattern shows both similarities and differences compared to existing literature. Our finding of central nervous system predominance aligns with Mahdi et al. [16] who reported central nervous system anomalies as the most common (29.2%) in their study of 5142 pregnant women in Baghdad, and with Tiwari et al. [14] who found central nervous system involvement in 31.7% of affected cases. Similarly, Khalid et al. [17] reported central nervous system anomalies as the third most common (20%) after cardiac and musculoskeletal anomalies. However, our results differ from Hamid et al. [12] who found musculoskeletal anomalies to be most prevalent (22.22%), followed by central nervous system and genitourinary anomalies (both 19.44%). The variation in organ system involvement across studies likely reflects differences in screening protocols, gestational age at examination, and population-specific genetic and environmental risk factors.

The significant association between gestational age and congenital anomaly detection (p=0.007) in our study, where no anomalies were detected in pregnancies ≤19 weeks compared to 9.6% detection in pregnancies >19 weeks, underscores the critical importance of timing in prenatal diagnosis. This finding is consistent with the mean gestational age of 20.04±4.90 weeks reported by Ahmed et al. [18] for optimal detection of urinary tract anomalies and supports the recommendations by Tiwari et al. [14] for routine second-trimester ultrasound screening. The absence of significant associations with maternal age (p=0.594) and parity (p=0.381) in our study contrasts with some previous findings, particularly Mahdi et al. [16] who identified maternal age >35 years as a risk factor (12.5%), and Khalid et al. [17] who found a significant correlation between maternal diabetes mellitus and fetal congenital anomalies (p=0.006). The lack of age-related association in our study may be explained by our study population having limited representation of advanced maternal age pregnancies, thereby reducing the power to detect age-related effects that become more pronounced in women over 35 years.

There are several limitations of this study that need to be

taken into consideration during interpretation of findings. As it is a single-center study from one institution, the results may not be perfectly generalizable to other individuals or healthcare environments with varying demographic profiles, risk factor distributions, or clinical practices. The comparatively small sample size of 225 individuals may also have reduced statistical power for identifying associations with some of the demographic variables, especially with regard to rarer anomalies or in subgroup analyses. The cross-sectional nature of the study gives us a point prevalence of anomalies at the point of ultrasound examination but does not incorporate the dynamic process of fetal development or the fact that anomalies may emerge at various gestational ages. Using ultrasound alone as the diagnostic tool may also have led to underreporting of certain anomalies that are more effectively identified on the basis of alternative techniques of imaging or examination after birth, and long-term neonatal follow-up or determination of accuracy of prenatal diagnoses and emergence of anomalies after birth were not covered in the study.

CONCLUSION

It has been concluded in the present study that congenital anomalies are an important issue of medical trouble, and the frequency of involvement has been maximum in the central nervous system. Although maternal age and parity had no correlation with the identification of anomalies, gestational age had been significant, and it highlights the value of conducting ultrasound scans at the right period of pregnancy in maximizing the accuracy of medical diagnostics.

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