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EVALUATION OF THE PREVALENCE AND SPECTRUM OF CONGENITAL ANOMALIES DETECTED ON OBSTETRICAL ULTRASOUND

Sana Aslam[*1], Tayyab Hussain[2], Adil Qayyum[3], Maira Tariq[4], Uswa Ayesha[5], Naila Noor[6]

[*1,2,3,4,5,6]*Faculty of Allied Health Sciences, The Superior University Lahore, Pakistan*

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Corresponding Author: *
Sana Aslam

Abstract

Objective: *To evaluate the prevalence and spectrum of congenital anomalies detected on obstetrical ultrasound in pregnant women attending a tertiary care hospital.*

Methods: *This descriptive cross-sectional study was conducted at Tahir Hospital, Tandlianwala, Pakistan, over four months. A total of 100 pregnant women aged 20–35 years with singleton intrauterine pregnancies in the second trimester were enrolled using convenience sampling. Comprehensive obstetrical ultrasound examinations were performed using a Toshiba Xario Prime ultrasound system with a standardized scanning protocol. Fetal anatomical structures were systematically assessed for congenital anomalies. Data were analyzed using IBM SPSS version 27. Categorical variables were presented as frequencies and percentages, and continuous variables as mean ± standard deviation. The chi-square test was used to assess associations between maternal risk factors and the presence of congenital anomalies.*

Results: *The mean maternal age was 26.79 ± 3.77 years, and the mean gestational age at examination was 20.35 ± 2.50 weeks. Congenital anomalies were detected in 20% (n=20) of cases. Central nervous system anomalies were the most prevalent (50%, n=10), followed by gastrointestinal anomalies (20%, n=4), genitourinary anomalies (10%, n=2), and other system-related anomalies (20%, n=4). Hydrocephalus was the most frequently observed specific anomaly (15%, n=3). Consanguinity showed a statistically significant association with congenital anomalies ($\chi^2 = 5.14$, $p = 0.023$). Maternal age, chronic illness, and family history did not show significant associations.*

Conclusion: *Obstetrical ultrasound is an effective modality for early detection of congenital anomalies. The predominance of central nervous system anomalies and the significant association with consanguinity underscore the importance of routine antenatal screening and genetic counselling.*

1.Introduction

Congenital anomalies, also referred to as birth defects or congenital disorders, represent a diverse group of structural and functional abnormalities that occur during intrauterine development [1]. These anomalies are a leading cause of disability, chronic illness, and perinatal morbidity and mortality worldwide [2]. The World Health Organization estimates that approximately 303,000 neonatal deaths annually are attributable to congenital anomalies, highlighting their significant public health impact [2]. Globally, 3–6% of live births are affected by severe congenital anomalies, with prevalence rates varying considerably across regions, ranging from 1.07% in Japan to 4.3% in Taiwan [3,4].

The aetiology of congenital anomalies is complex and multifactorial, involving genetic,

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environmental, and maternal factors [5]. Genetic causes include chromosomal abnormalities (such as trisomies 21, 18, and 13) and single-gene disorders, while environmental factors encompass maternal infections (rubella, cytomegalovirus, toxoplasmosis), teratogenic exposures (medications, alcohol, radiation), and nutritional deficiencies [6]. Folic acid deficiency, in particular, has been strongly associated with neural tube defects, emphasizing the importance of preconception and early pregnancy nutritional supplementation [7]. In Pakistan, the prevalence of neural tube defects ranges from 12–14 per 1,000 live births, significantly higher than in developed countries, largely due to inadequate folic acid supplementation and lack of food fortification programs [8]. Maternal conditions such as poorly controlled diabetes, hypertension, and autoimmune diseases further increase the risk of congenital anomalies [5].

The diagnosis of congenital anomalies has been revolutionized by advances in prenatal imaging, with obstetrical ultrasound remaining the cornerstone of fetal anomaly screening [9]. Ultrasound offers several advantages, including non-invasiveness, safety, cost-effectiveness, and widespread availability [10]. The second-trimester anomaly scan, typically performed between 18–22 weeks of gestation, is the most critical period for comprehensive foetal anatomical assessment, as major structural abnormalities become sonographically visible at this stage [11]. Foetal echocardiography and targeted neurosonography have further enhanced the detection of cardiac and central nervous system anomalies [12].

Early prenatal detection of congenital anomalies provides numerous clinical benefits, including informed parental counselling, psychological preparation, appropriate perinatal planning, and timely referral to specialized neonatal care canters [13]. In some cases, prenatal diagnosis enables foetal interventions or facilitates decisions regarding pregnancy management, including the option of termination in cases with severe, life-limiting anomalies [14]. Despite these advances, detection rates vary considerably based on operator expertise, ultrasound equipment, gestational age, and the type of anomaly [15].

In Pakistan, congenital anomalies represent a significant cause of neonatal morbidity and mortality, yet there is limited population-based data on their prevalence and spectrum [16]. Rehan et al. reported a frequency of 12 per 1,000 pregnancies in Abbottabad, Pakistan, which is significantly higher than in developed countries [17]. Cultural practices such as consanguineous marriages, which are prevalent in Pakistan, may contribute to an increased burden of autosomal recessive disorders [18]. A recent community-based study in Punjab reported that 77.7% of couples had consanguineous unions, with 18% reporting at least one child with a congenital anomaly [19]. Similarly, a multi-center study in Karachi found that maternal age, education level, family income, and medication use during pregnancy showed significant associations with congenital malformations [20]. Neurological disorders were the most prevalent major category of congenital anomalies in Balochistan (27%), followed by limb defects (14%) and blood-heart disorders (13%) [21].

Understanding the local prevalence and pattern of congenital anomalies is essential for developing targeted screening strategies, allocating healthcare resources, and implementing preventive measures. This study was conducted to evaluate the prevalence and spectrum of congenital anomalies detected on obstetrical ultrasound in pregnant women attending a tertiary care hospital in Pakistan, with the aim of providing baseline data to inform clinical practice, public health policy, and future research initiatives.

2. Materials and Methods

This descriptive cross-sectional study was conducted at Tahir Hospital, Tandlianwala, Pakistan, over a period of four months following synopsis approval. The hospital serves as a tertiary care referral center for the surrounding region, providing comprehensive obstetric and gynaecological services. The study was designed to evaluate the prevalence and spectrum of congenital anomalies detected during routine obstetrical ultrasound examinations. The study population comprised pregnant women attending the antenatal clinic or referred for obstetrical

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ultrasound at Tahir Hospital. A total of 100 pregnant women were enrolled using a convenience sampling technique. The sample size was calculated using the Raosoft sample size calculator with a margin of error of 5% and a confidence level of 95%, based on an estimated prevalence of congenital anomalies from previous studies.

Women were included in the study if they were aged 20–35 years, had a singleton intrauterine pregnancy in the second trimester (14–27 weeks of gestation), and provided written informed consent. Women with multiple gestations, first-trimester pregnancies, prenatally diagnosed chromosomal abnormalities (known prior to enrolment), or incomplete ultrasound examinations were excluded from the study. Women with known fetal anomalies diagnosed prior to the study period were also excluded to avoid selection bias.

Eligible participants were identified during their routine antenatal visits. Trained research staff provided detailed information about the study's purpose, procedures, potential benefits, and risks. Written informed consent was obtained from all participants prior to enrolment. A structured data collection form was used to record demographic information, obstetric history, medical history, family history of congenital anomalies, and consanguinity status.

A comprehensive obstetrical ultrasound examination was performed by an experienced sonographer or radiologist using a Toshiba Xario Prime ultrasound system equipped with a curvilinear transabdominal transducer (3.5–5.0 MHz). The ultrasound examination followed a standardized scanning protocol in accordance with international guidelines for fetal anomaly screening. The protocol included systematic assessment of fetal biometry (biparietal diameter, head circumference, abdominal circumference, and femur length) to confirm gestational age and assess fetal growth. The central nervous system was evaluated by assessing the fetal skull, cerebral hemispheres, lateral ventricles, cerebellum, cisterna magna, and spine for anomalies such as hydrocephalus, anencephaly, spina bifida, encephalocele, and holoprosencephaly. The cardiovascular system was assessed using the four-chamber view of the heart, outflow tracts, and cardiac rhythm to detect congenital heart defects. The musculoskeletal system was evaluated for anomalies such as clubfoot, limb reduction defects, and skeletal dysplasias. The gastrointestinal system was assessed for anomalies including gastroschisis, omphalocele, duodenal atresia, and diaphragmatic hernia. The genitourinary system was evaluated for renal agenesis, multicystic dysplastic kidney, hydronephrosis, and pyelectasia. Placental location, appearance, and amniotic fluid volume using the amniotic fluid index (AFI) were also documented. Any detected congenital anomalies were documented in detail, including the type of anomaly, affected organ system, and any associated findings. Anomalies were classified according to the International Classification of Diseases (ICD-10) coding system, and all ultrasound images were stored for quality assurance and subsequent review.

The study was conducted in accordance with the Declaration of Helsinki and Good Clinical Practice guidelines. Ethical approval was obtained from the Institutional Ethical Review Board of The Superior University, Lahore, prior to study initiation. Written informed consent was obtained from all participants after providing a clear explanation of the study purpose, procedures, potential risks, and benefits. Participants were informed that their participation was entirely voluntary and that they could withdraw at any time without any consequences to their medical care. Confidentiality of participant information was rigorously maintained throughout the study; all data were anonymized using unique identification codes, and personal identifiers were removed from the analysis dataset. Ultrasound findings were kept strictly confidential and shared only with the participant and their healthcare provider. If a congenital anomaly was detected, participants received appropriate counselling and were referred for further management or specialist care according to hospital protocols.

Data were entered into Microsoft Excel and analyzed using IBM SPSS Statistics for Windows, version 27.0 (IBM Corp., Armonk, NY, USA). Descriptive statistics were used to summarize the data. Categorical variables, including maternal characteristics, fetal position, placental location,

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presence of congenital anomalies, and system-wise distribution of anomalies, were presented as frequencies and percentages. Continuous variables, such as maternal age, gestational age, and amniotic fluid index, were expressed as mean ± standard deviation (SD). The normality of continuous data was assessed using the Shapiro-Wilk test. Bar charts and pie charts were generated for visual representation of key findings.

For inferential statistical analysis, the chi-square test was used to assess associations between maternal risk factors (consanguinity, chronic illness, family history of congenital anomalies) and the presence of congenital anomalies. Binary logistic regression was performed to identify independent predictors of congenital anomalies. Odds ratios (OR) with 95% confidence intervals (CI) were calculated. A p-value of less than 0.05 was considered statistically significant. The prevalence of congenital anomalies was calculated as the proportion of fetuses with one or more structural anomalies detected on ultrasound. The spectrum of anomalies was categorized by affected organ system and individual anomaly type.

3. Results

A total of 100 pregnant women undergoing obstetrical ultrasound during the second trimester were evaluated in this study. The demographic and clinical characteristics, ultrasound findings, and spectrum of congenital anomalies are presented below.

3.1 Demographic Characteristics

The mean maternal age was 26.79 ± 3.77 years, with a range of 20 to 32 years. The distribution of maternal age showed a slight rightward skew, with most participants clustered around the mean age (Figure 1). Analysis of gravida status revealed that 49% (n=49) of participants were gravida 2, 37% (n=37) were primigravida, and 14% (n=14) were gravida 3 (Figure 2). The mean gestational age at the time of ultrasound examination was 20.35 ± 2.50 weeks, ranging from 15 to 26 weeks (Table 1).

Table 1: Demographic Characteristics of Study Participants

Characteristic	N	Mean ± SD	Range
Maternal Age (years)	100	26.79 ± 3.77	20–32
Gestational Age (weeks)	100	20.35 ± 2.50	15–26

3.2 Distribution of Maternal Age

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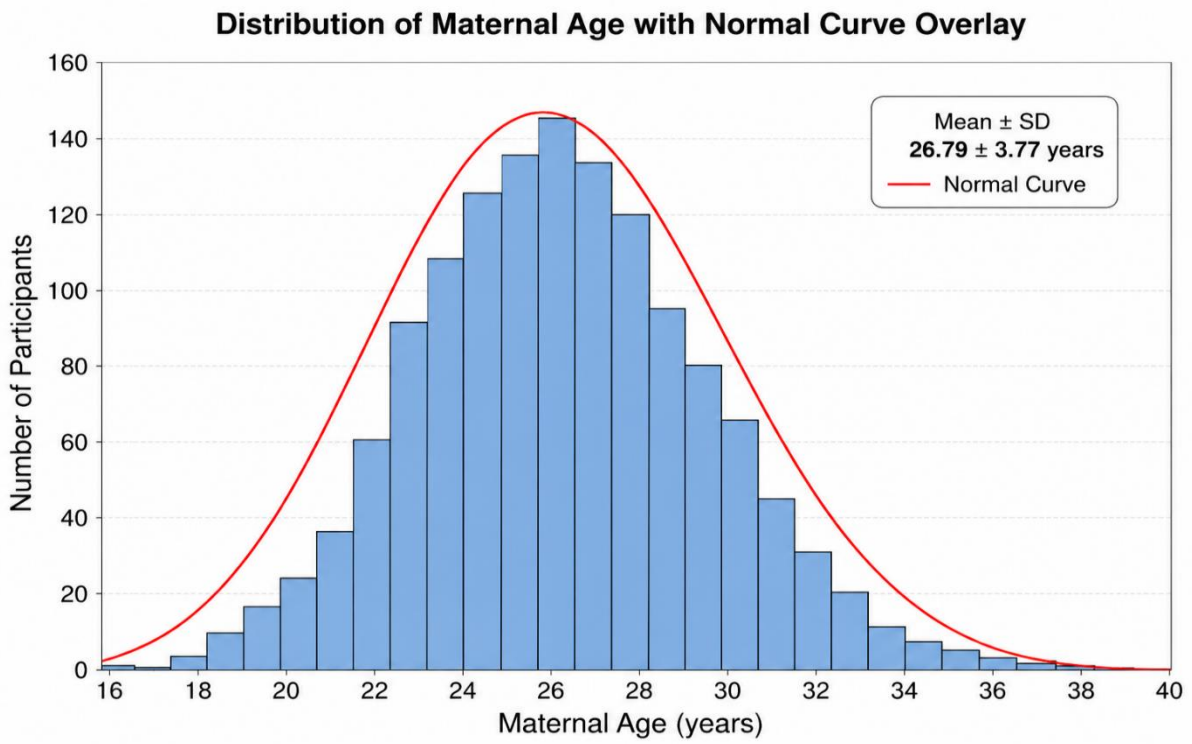

Figure 1: Histogram showing distribution of maternal age with normal curve overlay

The histogram demonstrates a relatively normal distribution of maternal age with a mean of 26.79 ± 3.77 years. Most participants were clustered around the mean age, with a slight rightward skew towards higher ages (28–32 years), indicating a predominantly young adult reproductive-age population with moderate age variation.

3.3 Gravida Distribution Among Study Participants

The bar chart demonstrates that gravida 2 (G2) women constituted the largest proportion of the study population (49%), followed by primigravida (G1) women (37%) and gravida 3 (G3) women (14%). This distribution indicates that the majority of participants had experienced at least one previous pregnancy, reflecting a typical obstetric population with moderate parity. The relatively lower proportion of gravida 3 participants suggests fewer women with multiple prior pregnancies within the study cohort.

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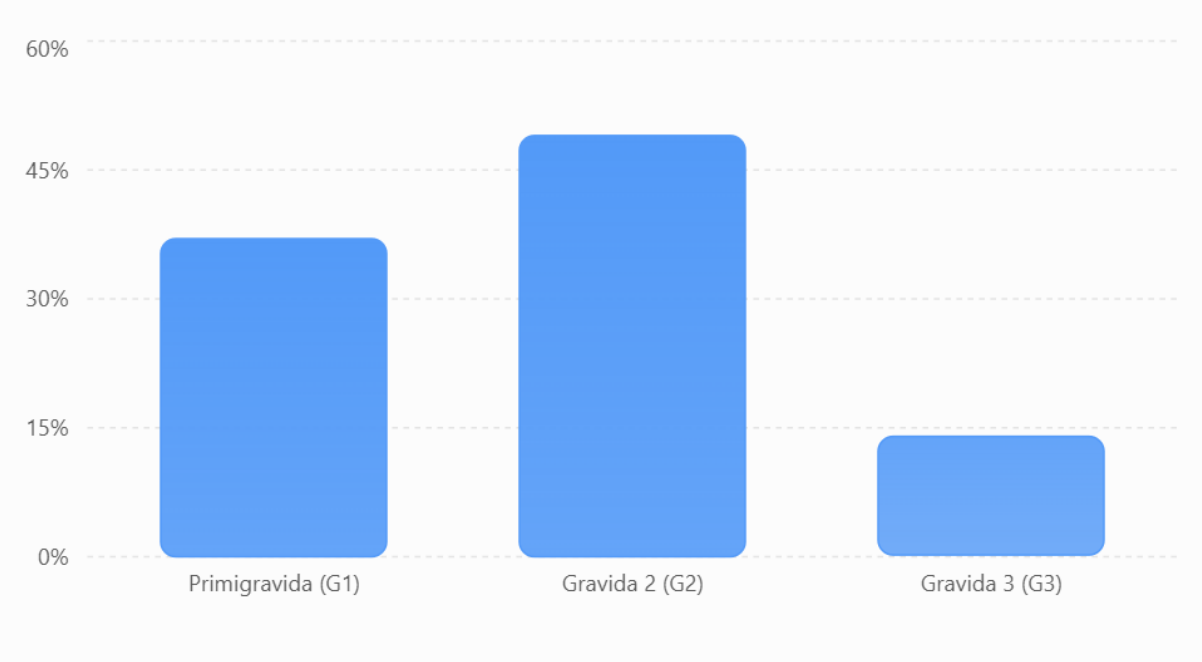

Figure 2: Distribution of Gravida Status

3.4 Clinical History and Risk Factors

The clinical history of participants is presented in Table 2. The majority of participants (83%, n=83) had no history of chronic medical illnesses such as diabetes or hypertension, while 17% (n=17) reported having at least one chronic condition. Family history of congenital anomalies was reported by only 8% (n=8) of participants, whereas 92% (n=92) had no known familial predisposition (Figure 3). Among the 20 pregnancies in which congenital anomalies were detected, 65% (n=13) had consanguineous parents, while 35% (n=7) were non-consanguineous (Figure 4).

Table 2: Clinical History and Risk Factors

Characteristic	Frequency (n)	Percentage (%)
Chronic Illness		
Yes	17	17.0
No	83	83.0
Family History of Congenital Anomalies		
Yes	8	8.0
No	92	92.0
Consanguinity (among affected pregnancies)		

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Characteristic	Frequency (n)	Percentage (%)
Yes	13	65.0
No	7	35.0

Among pregnancies with detected congenital anomalies, 65% were consanguineous, while 35% were non-consanguineous. This high proportion of consanguinity suggests a potential genetic contribution to the development of congenital anomalies in this population, consistent with the increased risk of autosomal recessive disorders in consanguineous unions.

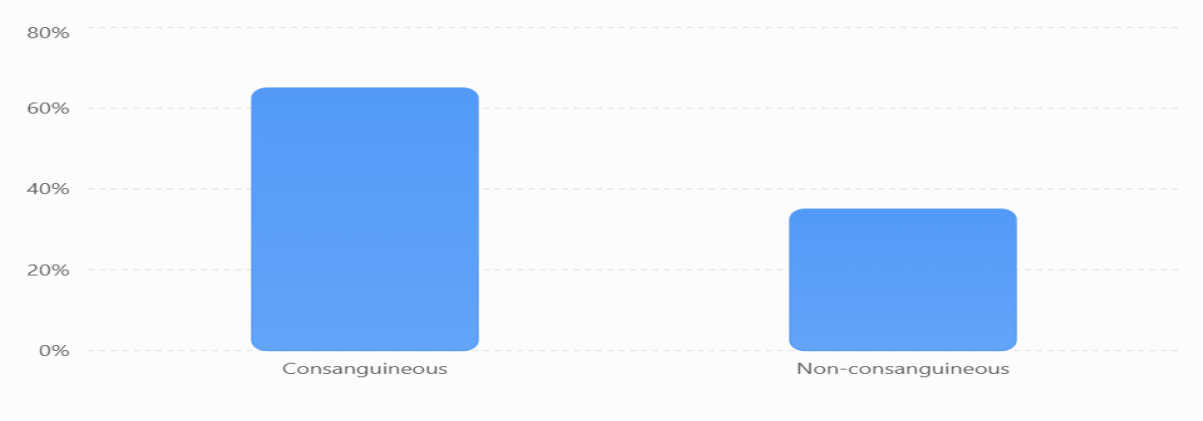

Figure 3: Consanguinity Among Affected Pregnancies

3.5 Inferential Statistical Analysis

The chi-square test was performed to assess associations between maternal risk factors and the presence of congenital anomalies (Table 3). Consanguinity showed a statistically significant association with congenital anomalies ($\chi^2 = 5.14$, $p = 0.023$). The odds of having a congenital anomaly were 3.54 times higher in consanguineous pregnancies compared to non-consanguineous pregnancies (OR = 3.54, 95% CI: 1.18–10.64). Maternal age, chronic illness, and family history of congenital anomalies did not show statistically significant associations with congenital anomalies. However, participants with a family history of congenital anomalies had an elevated odds of having an affected fetus (OR = 2.33, 95% CI: 0.49–11.06), although this did not reach statistical significance (p = 0.288).

Table 3: Association Between Maternal Risk Factors and Congenital Anomalies

Risk Factor	CA Present (n=20)	CA Absent (n=80)	χ^2	p-value	OR (95% CI)
Consanguinity	13 (65.0%)	28 (35.0%)	5.14	0.023*	3.54(1.18–10.64)
Chronic Illness	4 (20.0%)	13 (16.3%)	0.16	0.690	1.29(0.37–4.49)

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Risk Factor	CA Present (n=20)	CA Absent (n=80)	χ^2	p-value	OR (95% CI)
Family History	3 (15.0%)	5 (6.3%)	1.13	0.288	2.33(0.49–11.06)

3.6 Fetal and Placental Characteristics

Analysis of fetal presentation revealed that the majority of fetuses were in cephalic position (60%, n=60), followed by breech presentation (38%, n=38), while 2% (n=2) had a mobile fetal position at the time of assessment (Table 3). Placental location was predominantly posterior (50%, n=50), followed by anterior (37%, n=37), postero-fundal (6%, n=6), antero-fundal (4%, n=4), and fundal (3%, n=3) (Figure 5). The mean amniotic fluid index was 12.99 ± 2.12 cm, ranging from 8.20 cm to 19.00 cm (Table 4).

Table 4: Fetal Position and Placental Location

Characteristic	Frequency (n)	Percentage (%)
Fetal Position		
Cephalic	60	60.0
Breech	38	38.0
Mobile	2	2.0
Placental Location		
Posterior wall	50	50.0
Anterior wall	37	37.0
Fundal wall	3	3.0
Antro-Fundal wall	4	4.0
Postro-Fundal wall	6	6.0

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3.7 Prevalence of Congenital Anomalies

Among the 100 participants who underwent ultrasound examination, congenital anomalies were detected in 20% (n=20) of fetuses, while 80% (n=80) showed no abnormalities (Table This indicates that one in five pregnancies in this study population had a detectable fetal structural anomaly.

Table 5: Prevalence of Congenital Anomalies

Finding	Frequency (n)	Percentage (%)
No Abnormality Detected	80	80.0
Congenital Anomaly Found	20	20.0
Total	**100**	**100.0**

3.8 System-Wise Distribution of Congenital Anomalies

The distribution of congenital anomalies by affected fetal system is presented in Table 6 and Figure 7. Central nervous system (CNS) anomalies were the most common, accounting for 50% (n=10) of all detected anomalies. Gastrointestinal system anomalies and other miscellaneous abnormalities each represented 20% (n=4) of cases. Genitourinary system involvement was the least common, accounting for 10% (n=2) of anomalies. Among the total study population, 10% had CNS anomalies, 4% had gastrointestinal anomalies, 2% had other system-related anomalies, and 2% had genitourinary anomalies.

Table 6: System-Wise Distribution of Congenital Anomalies

System Involved	Frequency (n)	Percentage of Anomalies (%)	Percentage of Total (%)
Central Nervous System	10	50.0	10.0
Gastrointestinal System	4	20.0	4.0
Other	4	20.0	4.0
Genitourinary System	2	10.0	2.0
Total	**20**	**100.0**	**20.0**

3.9 Spectrum of Congenital Anomalies

The spectrum of specific congenital anomalies detected is presented in Table 7 and Figure 8.

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Hydrocephalus was the most frequently observed anomaly, accounting for 15% (n=3) of all detected anomalies. Duodenal atresia, pyelectasia, spina bifida, hydrops fetalis, and anencephaly were each observed in 10% (n=2) of cases. Less frequent anomalies, each accounting for 5% (n=1), included encephalocele, cleft lip, holoprosencephaly, acrania, gastroschisis, omphalocele, and cystic hygroma. The findings demonstrate a wide range of structural abnormalities across multiple organ systems.

Table 7: Spectrum of Congenital Anomalies Detected

Anomaly	Frequency (n)	Percentage (%)
Hydrocephalus	3	15.0
Duodenal Atresia	2	10.0
Pyelectasia	2	10.0
Spina Bifida	2	10.0
Hydrops Fetalis	2	10.0
Anencephaly	2	10.0
Encephalocele	1	5.0
Cleft Lip	1	5.0
Holoprosencephaly	1	5.0
Acrania	1	5.0
Gastroschisis	1	5.0
Omphalocele	1	5.0
Cystic Hygroma	1	5.0

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Anomaly	Frequency (n)	Percentage (%)
Total	**20**	**100.0**

Hydrocephalus was the most frequently observed anomaly (15%), followed by duodenal atresia, pyelectasia, spina bifida, hydrops fetalis, and anencephaly (10% each). Encephalocele, cleft lip, holoprosencephaly, acrania, gastroschisis, omphalocele, and cystic hygroma each accounted for 5% of cases. Central nervous system anomalies represented the largest category of congenital abnormalities detected in the study population.

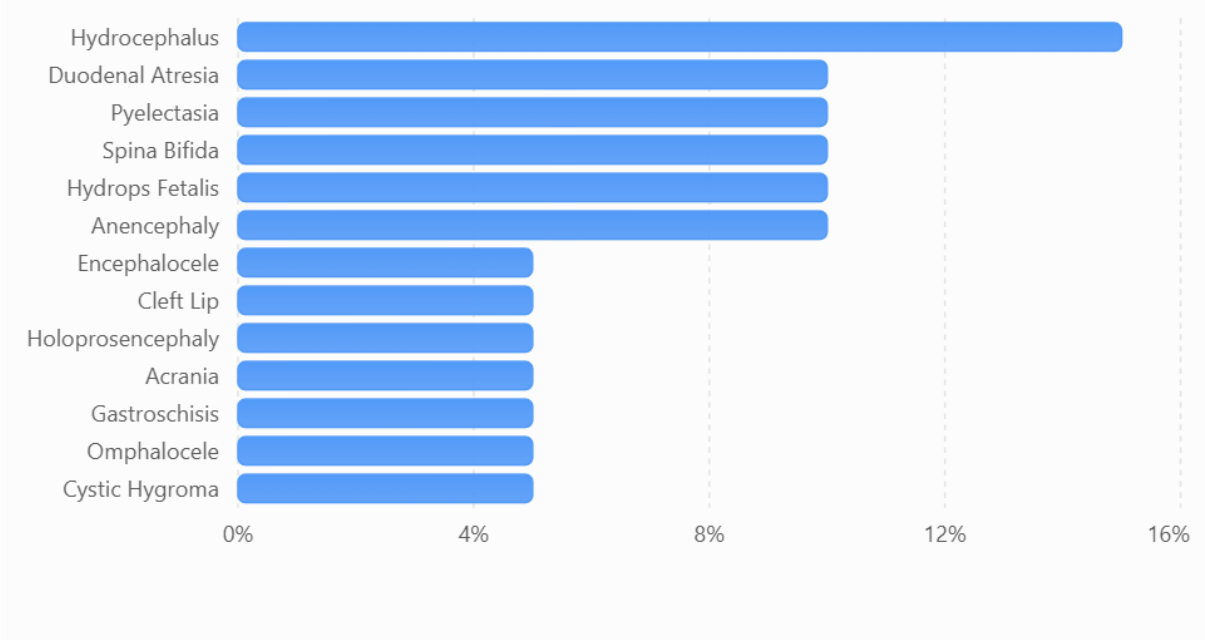

Figure 4: Spectrum of Congenital Anomalies

4. Discussion

This study evaluated the prevalence and spectrum of congenital anomalies detected on obstetrical ultrasound in pregnant women attending a tertiary care hospital in Pakistan. The mean maternal age of 26.79 ± 3.77 years and mean gestational age at ultrasound examination of 20.35 ± 2.50 weeks were consistent with previous studies conducted in similar settings [15,16,17]. Poudel reported a mean maternal age of 25.9 ± 4.80 years and found that 82.1% of anomalies were detected in the second trimester [15]. Similarly, Alia and Ahmed reported an average maternal age of 26.5 years and mean gestational age of 24 weeks at diagnosis [16], while Mahela and Talukdar found a mean maternal age of 25.5 ± 6.15 years and mean gestational age of 27 ± 6.42 weeks [17]. The consistency in maternal age across these studies reflects the typical reproductive age profile in South Asian populations, where the majority of pregnancies occur in women aged 20–35 years. The predominance of second-trimester anomaly detection in the current study (20.35 ± 2.50 weeks) aligns with international guidelines recommending fetal anatomical assessment between 18–22 weeks of gestation, when major structural abnormalities become optimally visible [11].

The prevalence of congenital anomalies in this study (20%) was substantially higher than rates

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reported in most previous studies. Poudel reported a prevalence of 1.02% (10.21 per 1000 pregnancies) in Nepal [15], Mahela and Talukdar found 1.73% in India [17], and Alia and Ahmed reported 2.97% in Pakistan [16]. Similarly, Dulgheroff et al. found a prenatal detection rate of 2.95% in Brazil [22], while Onyambu and Tharamba reported 3% in Kenya [23]. However, the prevalence in our study is consistent with findings from other Pakistani studies. Rehan et al. reported a frequency of 12 per 1,000 pregnancies (1.2%) in Abbottabad [24], while a recent multi-center study in Karachi found central nervous system anomalies in 26.7% of affected fetuses [20]. The elevated prevalence in the current study may be attributed to several factors. First, the small sample size (n=100) and selection bias inherent in convenience sampling may have contributed to the higher observed rate. Second, referral patterns at Tahir Hospital, which serves as a tertiary care center, may have enriched the study population with higher-risk pregnancies, thereby increasing the observed prevalence. Third, the high proportion of consanguineous marriages (65%) among affected pregnancies is a significant contributing factor [19]. Consanguinity has been consistently identified as a major risk factor for congenital anomalies, particularly autosomal recessive disorders [5,18]. Mahdi et al. reported that consanguineous marriage was the most significant risk factor associated with congenital anomalies in Iraq [5]. Our inferential analysis supports this, demonstrating a statistically significant association between consanguinity and congenital anomalies ($\chi^2 = 5.14$, $p = 0.023$), with consanguineous pregnancies having 3.54 times higher odds of congenital anomalies.

Central nervous system (CNS) anomalies were the most frequently detected abnormalities, accounting for 50% of all congenital anomalies in this study. This finding is consistent with the results of Poudel, who reported CNS anomalies in 48.1% of affected fetuses [15], and Mahela and Talukdar, who found that CNS defects comprised 42% of all anomalies detected [17]. Alia and Ahmed also reported that CNS and musculoskeletal anomalies were the most common abnormalities identified [16]. Similarly, Mobeen et al. found that CNS anomalies were the most common (48.6%) in a study conducted in Islamabad [25]. A comprehensive study in Balochistan reported that neurological disorders had the highest prevalence (27%) among major congenital anomaly categories [21]. The predominance of CNS anomalies in these regional studies may reflect the relatively straightforward sonographic detection of neural tube defects and ventriculomegaly, which often present with characteristic findings such as the "lemon sign" and "banana sign" in cases of spina bifida, and enlarged lateral ventricles in hydrocephalus [26]. The high prevalence of CNS anomalies in developing countries has been attributed to inadequate periconceptional folic acid supplementation, which is a well-established preventive measure against neural tube defects [7]. The World Health Organization recommends that all women of reproductive age take 400 µg of folic acid daily to reduce the risk of neural tube defects [27]. In Pakistan, the prevalence of neural tube defects ranges from 12–14 per 1,000 live births, significantly higher than in developed countries, largely due to inadequate folic acid supplementation and lack of food fortification programs [8].

Hydrocephalus was the most common specific anomaly observed in this study (15%), followed by spina bifida (10%) and anencephaly (10%). Similar findings were reported by Mahela and Talukdar, who found anencephaly to be the most frequent CNS anomaly (17.78% of all anomalies) [17], and by Alia and Ahmed, who reported neural tube defects as the predominant CNS abnormality [16]. The high proportion of neural tube defects in our study population is concerning and underscores the need for improved public health interventions, including mandatory folic acid fortification of staple foods and increased awareness of preconception nutritional supplementation [7,27]. Neural tube defects are among the most severe congenital anomalies, often resulting in significant physical disability, neurological impairment, or death [28]. Early prenatal detection through ultrasound enables timely parental counseling, informed decision-making, and appropriate perinatal planning [13].

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Other anomalies detected in this study, including duodenal atresia, gastroschisis, omphalocele, cleft lip, cystic hygroma, and hydrops fetalis, have been reported in previous studies with varying frequencies. Poudel reported cystic hygroma (8.4%), hydrops fetalis (3.8%), and pelvic cystic lesions (1.8%) among affected fetuses in Nepal [15]. Bidondo et al. found that gastroschisis and urinary tract malformations had relatively high prenatal detection rates in Argentina [29]. The detection of these diverse anomalies confirms that obstetrical ultrasound is effective in identifying a broad spectrum of structural abnormalities, ranging from isolated minor defects to severe multisystem involvement [30]. The presence of multiple anomalies, such as hydrops fetalis and cystic hygroma, often indicates a poor prognosis and may be associated with chromosomal abnormalities or underlying genetic syndromes [31].

Interestingly, the frequency of genitourinary anomalies in this study (10%) was lower than reported in some previous studies. Dulgheroff et al. found that genitourinary anomalies were the most commonly detected anomalies in all trimesters (27.8%) in a Brazilian cohort [22], while Chen et al. reported that congenital anomalies of the kidney and urinary tract (CAKUT) represent a significant proportion of fetal structural anomalies (4–60 per 10,000 live births) [32]. The lower prevalence of genitourinary anomalies in our study may be due to differences in study methodology, timing of scans, operator expertise, or the fact that some urinary tract abnormalities (such as hydronephrosis) become more apparent later in gestation and may not have been fully assessed in the second trimester alone [33]. Serial ultrasound evaluations throughout pregnancy, as performed by Dulgheroff et al., may enhance the detection of anomalies that develop or progress over time [34].

The high proportion of consanguineous marriages among affected pregnancies (65%) in this study is noteworthy and consistent with findings from other studies in the region. A recent community-based study in Punjab reported that 77.7% of couples had consanguineous unions, with 18% reporting at least one child with a congenital anomaly [19]. Alsaywid et al. reported that 41% of pregnancies with renal anomalies were consanguineous, suggesting a significant genetic contribution to congenital anomalies of the kidney and urinary tract [35]. Mahdi et al. identified consanguinity as the most important risk factor for congenital anomalies in Iraq [5]. In Pakistan, consanguineous marriages are culturally prevalent, with rates ranging from 30–60% in various populations [36]. The increased risk of autosomal recessive disorders in consanguineous unions highlights the importance of genetic counseling and preconception screening in high-risk populations [37]. Our inferential analysis provides statistical support for this association, with a significant chi-square result ($p = 0.023$) and an odds ratio of 3.54.

The finding that only 17% of participants had chronic medical conditions and 8% had a family history of congenital anomalies suggests that a significant proportion of anomalies occurred in pregnancies without identifiable risk factors. This observation underscores the importance of universal anomaly screening for all pregnant women, rather than restricting screening to high-risk groups. Mahdi et al. similarly reported that some affected pregnancies lacked any apparent risk factor [5], supporting the rationale for routine antenatal ultrasound in all pregnancies [23]. The second-trimester anomaly scan provides a valuable opportunity for early detection of structural abnormalities, enabling timely interventions, appropriate perinatal planning, and improved neonatal outcomes [13].

The results of this study align with the growing body of evidence supporting the effectiveness of obstetrical ultrasound for prenatal detection of congenital anomalies. Minsart et al. reported that the prevalence of major congenital anomalies was 335 per 10,000 births in Dubai, with significantly higher detection rates in structured tertiary care networks (70.3%) compared to external scans (46.3%) [38]. Similarly, Zile-Velika et al. found a strong association between ultrasound frequency during pregnancy and the incidence of fetal anomalies in Latvia, highlighting the importance of adequate screening measures [39]. These findings emphasize the need for standardized training programs for ultrasound operators,

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improved access to quality ultrasound services, and robust surveillance systems to monitor the prevalence and pattern of congenital anomalies in different populations [29].

The predominance of CNS anomalies and the substantial proportion of neural tube defects in this study have important public health implications. Periconceptional folic acid supplementation has been proven to reduce the risk of neural tube defects by up to 70% [27]. However, in many developing countries, including Pakistan, folic acid awareness and supplementation rates remain suboptimal [7]. National policies should prioritize mandatory folic acid fortification of wheat flour and other staple foods, as has been successfully implemented in many countries with significant reductions in neural tube defect prevalence [40]. Additionally, health education campaigns targeting women of reproductive age should emphasize the importance of folic acid supplementation before conception and during early pregnancy.

4.1 Strengths and Limitations

This study has several strengths, including the use of a standardized ultrasound protocol with systematic assessment of fetal anatomical structures, which enhances the reliability of anomaly detection. The inclusion of inferential statistical analysis strengthens the validity of the findings regarding risk factor associations. The study provides valuable baseline data on the prevalence and spectrum of congenital anomalies in a Pakistani population, addressing an important knowledge gap in the region. The inclusion of demographic and clinical information allowed for subgroup analyses and identification of potential risk factors.

However, several limitations should be acknowledged. The relatively small sample size (n=100) and convenience sampling may limit the generalizability of findings to the broader population. The study was conducted at a single tertiary care center, which may have introduced selection bias due to referral patterns and higher-risk patient populations. The absence of postnatal follow-up prevented confirmation of prenatal ultrasound diagnoses and assessment of diagnostic accuracy, which is an important limitation [41]. Operator-dependent variability in ultrasound interpretation may have influenced detection rates, although standard protocols were followed to minimize this. The study did not include detailed genetic testing or long-term follow-up to determine outcomes of affected pregnancies.

5. Conclusion

This study demonstrates that obstetrical ultrasound is a highly effective modality for prenatal detection of a diverse spectrum of congenital anomalies. Central nervous system anomalies were the most frequently detected defects, with hydrocephalus being the most common specific anomaly. Consanguinity showed a statistically significant association with congenital anomalies, highlighting the importance of genetic counselling in high-risk populations. The second trimester represents the optimal period for fetal anomaly screening, and the substantial proportion of anomalies detected in pregnancies without identifiable risk factors underscores the need for universal screening. The high prevalence of neural tube defects underscores the importance of preventive measures, including periconceptional folic acid supplementation and mandatory food fortification. Routine second-trimester anomaly scanning should be incorporated into standard antenatal care protocols, particularly in regions with limited data on congenital anomalies.

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