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# EPIDEMIOLOGY AND PATTERN OF CONGENITAL MALFORMATIONS AMONG 1900 CONSECUTIVE LIVE BIRTHS AT A TERTIARY CARE HOSPITAL IN PAKISTAN

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## Abstract

*Congenital abnormalities remain a leading cause of perinatal morbidity and mortality globally, particularly in low- and middle-income countries with weak surveillance systems. In Pakistan, hospital-based data on the prevalence and distribution of congenital abnormalities are limited, particularly in the federal capital region.*

*Over the course of four months, a descriptive epidemiological study was carried out at an Islamabad tertiary care hospital. A total of 1,967 consecutive deliveries to 1,889 moms were evaluated for congenital abnormalities using clinical assessments and medical record reviews. A standardized questionnaire was used to collect information about maternal, neonatal, and demographic characteristics. Congenital abnormalities were identified using the ICD-10 and OMIM criteria, and prevalence was computed per 1,000 births. Chi-square tests and odds ratios were used to examine the relationships between selected maternal and neonatal variables. Out of 1,967 newborns, 108 were identified with congenital abnormalities, resulting in an overall prevalence of 54.9 per 1,000 births. Males were more likely to have congenital abnormalities (5.5% vs. 4.9%). Malformations were more common in stillbirths and neonatal deaths (20%) than in live births (4.7%; $p < 0.0001$). The musculoskeletal system was the most often damaged system (24.3%), followed by central nervous system anomalies (22.4%), urogenital malformations (15.0%), and cardiovascular anomalies (12.2%). The most common individual malformations were clubfoot and hydrocephalus. Congenital abnormalities were more common among moms aged 25-35 years, as well as primigravida and multigravida women, however these correlations were not statistically significant. This hospital-based study found a high prevalence and complex pattern of congenital abnormalities among consecutive newborns in a*

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*tertiary care environment in Pakistan. The prevalence of musculoskeletal and central nervous system defects, along with a much greater probability of stillbirths, emphasizes the importance of systematic congenital anomaly surveillance, enhanced antenatal screening, and strengthened perinatal care facilities. These results provide critical baseline data for public health planning and future population-based investigations in the region.*

**Introduction**

Congenital malformations (CMs) or birth defects are a significant worldwide health issue, and the primary cause of neonatal morbidity, chronic disability, and baby death. As per the recent estimates of the world, in the countries with low and middle income there is a significant percentage of the death of the newborns because of some congenital anomalies, and its prevalence is higher due to the lack of access to preventive care, early diagnosis, and the surveillance system (Banu et al., 2023). Nevertheless, in most of the areas, the number of congenital malformations keeps increasing regardless of the progress in prenatal screening and newborn care, which is mostly attributed to demographic changes, environmental exposures, nutritional inadequacies, and disparities in healthcare accessibility (Bai et al., 2024; Shetty et al., 2023).

Minor anomalies refer to the non-vital organs that have minimal or no effect. They do not result in any suffering in the baby and in most instances there is no urgency in fixing them at least during the neonatal period. Major or serious anomalies, conversely, harm or have great cosmetic importance. They can even prove fatal. Therefore, they need an urgent correction. Otherwise, even a big problem can ruin the well being and development of the child if those anomalies are not corrected in their early stages (Balakumar, 2004; Singh et al., 2006).

The geographical distribution of the epidemiology and pattern of congenital malformations is significantly different, which is attributed to variations in genetic background, maternal health, socioeconomic status, and healthcare infrastructure (Wissmann, 2024; Pan et al., 2022). Musculoskeletal, central nervous system, cardiovascular, and urogenital anomalies are always on the list of the most recurring systems involved in the world, even though their proportional distribution differs between populations (Denaro et al., 2023). Significantly, the hospital-based studies continue to be an essential source of default epidemiological data in the countries where the registries of congenital anomalies on the population level have not been created yet (Luo et al., 2025).

The issue of congenital malformations is escalated in South Asian nations such as Pakistan due to low-coverage of antenatal screening, late manifestation in pregnancy, and inappropriate postnatal surveillance systems (Zaki et al., 2025). The studies available in Pakistan indicate a large spectrum of reported prevalence numbers representing methodology heterogeneity, variation in study settings, and underreporting of anomalies noticed post-birth (Afzal et al., 2025). In addition, the majority of the available research is limited to the chosen cities, and the data on the federal capital and its surroundings is lacking.

It has been demonstrated that maternal and neonatal factors (maternal age, parity, birth weight, neonatal sex, and pregnancy outcome) have an effect on the incidence and severity of congenital malformations (Ananth et al., 2023). Increased incidences of congenital anomalies in male neonates and greatly enhanced incidence rates in stillbirths and neonatal mortality have been reported in several studies, highlighting the importance of congenital malformations in outcomes related to adverse perinatal mortality (Mehare et al., 2025). These epidemiological correlates are important to understand in order to implement specific preventive measures and enhance early diagnosis.

Even though the study of consanguinity, fertility and reproductive compensation is complex, the net result is a lower rate of elimination of deleterious genes in the gene pool thereby decreasing the process of cleansing of the gene

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pool that has been linked to inbreeding (Hilliard, 2024).

When human Genetics, couples are considered to be consanguineous when they share common ancestry one or more. Since the vast majority of couples in one place have a common ancestor somewhere on their family tree, in reality the search to find a common ancestor often does not go more than three or four generations. Medical genetics use the definition of a consanguinity of marriage usually referring to the marriage of two individuals of the same sex who are second cousins or fewer. The second cousins have 1/32 common ancestor genes and, on average, their offspring will be homozygous at 1/64 loci (Temaj et al., 2022).

The given study was aimed at identifying the cases of congenital defects in neonates in a systematic manner with the help of the standardized diagnostic prediction models, such as the Online Mendelian Inheritance in Man (OMIM) and the International Classification of Diseases, Tenth Revision (ICD-10). It also sought to investigate the connection between the malformations at birth and the socio-biological factors of the investigated population with special reference to maternal, neonatal, and demographic factors. The other major aim was to assess the relationship between parental consanguinity and the health outcomes of the newborns in terms of morbidity and mortality rates. Besides this, the research was aimed at the determination of the coefficient of inbreeding (F) and finding its correlation with various patterns of marriage and prevalence of congenital malformations. Lastly, the research sought to examine if there were a temporal changes in inbreeding by comparing the consanguinity pattern of the parental generation to the study subjects to give the information on the changes in genetic structure across generations.

## Materials and methods

### Sampling site

In the current study, Pakistan institute of Medical Sciences (PIMS), Islamabad was studied on genetic epidemiology and prevalence of congenital malformations in Pakistan. PIMS is a tertiary care hospital that was set up in 1985 and is located in the heart of Federal Capital, Islamabad. The hospital is divided into various sections such as Islamabad Hospital, Children Hospital, Maternal and Child Health Care Centre (MCH) and Quaid-i-Azam Postgraduate Medical College (QPGMC) as well as College of Medical Technologies. It is well endowed with a very good geographic location, which also results in a huge number of patients being referred in twin cities, Rawalpindi and Islamabad as well as adjacent districts such as: Bara Kahu, Ali Pur, Ari Syedan, Bani gala, Pind Bhagwal, Nilore, Talhaar and Phulgiran etc. which also reflects a large number of subjects referred to PIMS.

The Maternal and Child Health Care Centre (MCH) was with the help of Japanese government. The Centre has highly developed medical facilities and it can cater to any type of emergency situation. Such facilities are diagnostic set, Pathology laboratory, Radiology, Blood bank, CTG (Cardiotocography) machines, ultrasound and Emergency resuscitation facilities.

To survey CM on the neonates MCH General Wards such as Gynecology Ward, Prenatal Ward, Postnatal Ward and Nursery were visited. The most common places of examining neonates were unit 1 and 2 of Postnatal Ward and Nursery. General Ward has 90 beds capacity; Labor Ward floor has capacity of 10 beds and the Private Ward has 35 beds capacity. The rate of delivery has risen in MCH labor room to over 5,000/year in 2003 as compared to 1,500 in 1994.

### Consent Approval

My research proposal has been discussed at Review Committee of Department of Animal Sciences, Quaid-i-Azam University (QAU) Islamabad, before the actual introduction of this project. The research proposal was also reviewed by Ethical Review Committee of PIMS. The concerned authorities of PIMS (i.e. the Director and In charge of the General Ward and Nursery) gave their go-ahead to the project to have the data collected. Mother/family heads of every neonate also gave consent in filling in this proforma.

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**Study Design**

It was a descriptive epidemiology of births and still-born babies at PIMS, a maternity hospital, throughout the four months from October 2010 to January 2011. The descriptive study design examines the variability in disease rates among a population by considering factors such as age, gender, caste, and environmental conditions. This sort of study design can determine trends and patterns of occurrence of a given condition within a specific geographical place, as well as the factors that are connected with it. However, it is unable to identify the causal agents. It includes case reports, surveillance systems, cross-sectional studies, and cluster investigations.

**Questionnaire Designing**

The data were collected through the standard questionnaires that were designed based on the objectives of the study and it had two sections (Annexure I, II). The variables were initially captured under the category of Demography, maternal age, parental consanguinity and detailed birth record of the pregnancy including previous pregnancy detailed, birth order, type of delivery, type of delivery, gestational age and history of CM in other children. The second section was on neonatal characters such as live or still-birth, sex, body length, birth weight, head circumference and APGAR (Activity, Pulse, Grimace, Appearance and Respiration) score and presence of CM and type of it, which was gathered in the medical records.

**Research Team**

In the case of data collection, a team of researchers was developed. The team members consisted of two researchers of Human Genetics lab, Department of Animal Sciences Quaid-i-Azam University Islamabad, and I was among them. In addition to this, the team also included staff members who were in-service at PIMS like resident doctors and nurses.

**Questionnaire Filling**

Pediatricians screened and tested all babies born in PIMS against malformation of congenital malformations. It entailed ultrasound during ante-natal, apparent at birth congenital anomaly and anomaly at birth, which were apparent under ultrasound or X-ray. The newborns medical records of the malformations that were in-congenital to the newborn were relaxed to study. The structured interview was conducted to complete the questionnaire. The data were collected by interviewing the delivering mother and examining the neonates and filling the questionnaires. Part 1 (Annexure I, II) of the questionnaire was done through interviewing the mother followed by the current pregnancy part which was done in line with the availability of record file. The second section of the questionnaire was responded to after researching about the neonate. The physical measurements were measured at the spot or at the Discharge Slip of neonate and medical record.

The data was encompassed on all the pregnant women and the babies who were live and admitted in the General Ward after the delivery time. The data of all the neonates were also recorded in Nursery of General Ward. The information gathered in the Private Ward did not occur since parents, medical and paramedical staff did not collaborate. The deliveries, which were commencing on holidays (weekends, Eid, Muharram), were not also included in this study.

**Dysmorphology Record and Clinical Ascertainment**

The clinical characteristics of the neonates were recorded in discharge slip of neonates or maternal file in the case of abnormal condition in the neonates. Where there was any suspicion, diagnosis confirmation was performed with the help of the medical officer who was on duty. The neonate file was also discussed in detail with the doctors in the Nursery and the entire detail of anomaly was noted.

**Anomalies classification**

The first determination of the anomaly was carried out by the child ward gynecologist and the neonatologist. This data was noted in the maternal file. Abnormalities were denoted based on the system used by ICD-10 codes

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(International Classification of Diseases) and OMIM (Online Mendelian Inheritance in Man).

**Statistical Analysis, storage and Data entry**

Proforma data were typed and stored in the MS Excel (ver. 2007). Prior to any processing and analysis data, any errors were eliminated before counter checking its data. MS Excel 2007, GraphPad and SPSS were used to analyze data. Simple frequency tables and the distributions matrices, Chi-square test, one way Analysis of variance (ANOVA) and odd ratio were used to summarize and analyze the data in order to determine the significance of the variables. A descriptive analysis was done to compute frequency distribution and percentages. The Chi-square test was applied to evaluate differences of different groups of data. The significance level taken as p=0.05. Odd ratio test (OR) was used to determine the relative risk of occurrence of malformation in neonates on the basis of various maternal parameters and parental parameters. The coefficient of inbreeding (F) was obtained. In order to compare the disparities among various parameters and to obtain the trend of change, the data were plotted with the assistance of GraphPad.

After classifying their prevalence, proportion and confidence interval were obtained. The calculation included prevalence per 1,000 of anomaly which is the number of neonate with anomaly/neonates total/1,000. Proportion was done by dividing the number of neonates with particular anomaly by the total number of neonates. To establish the relationships between various neonatal and parental parameters as well as determining the level of significance Chi-square test was conducted. To determine significance of the statistical 95% CI (confidence interval) was also considered and this is the true prevalence ratio with confidence.

**Statistical expressions used in the analysis**

$$\text{Percentage} = \frac{\text{No. of neonates with a specific anomaly}}{\text{No. of total anomalies}} \times 100$$

$$\text{Prevalence} = \frac{\text{No. of neonates with a specific anomaly}}{\text{No. of total neonates}} \times 1000$$

$$\text{Proportion} = \frac{\text{No. of neonates with a specific anomaly}}{\text{No. of total anomalies}}$$

$$95\ \%\ CI = p \pm 1.96 \sqrt{\frac{p(1-p)}{N}}$$

(Where p = proportion, N = total number of neonates

CI = Confidence Interval)

**Table 1: The following online databases were surveyed during the course of this project:**

| Databases | Description | URL |
|---|---|---|
| NCBI, PubMed | National Centre for Biotechnology Information | www.ncbi.nlm.nih.gov/ www.ncbi.nlm.nih.gov/pmc/ |
| OMIM | Online Mendelian Inheritance in Man | www.ncbi.nlm.nih.gov/omim/ |

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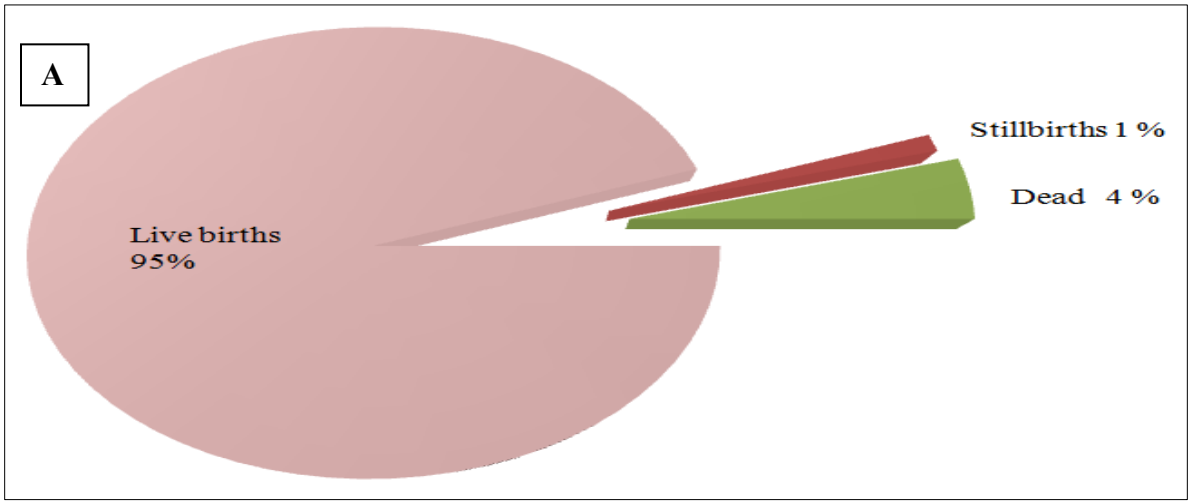
| | | |
|---|---|---|
| London Dysmorphology Database | London Dysmorphology Database | www.lmdatabases.com |
| CARIS | Congenital Anomaly Register & Information | www.wales.nhs.uk |
| ICD | International Classification of Diseases | www.who.int/classifications/icd/en |
| EUROCAT | European surveillance of Congenital Anomalies | www.eurocat-network.eu/ |
| Pak Medi Net | Pakistan Medical Journals repository | www.pakmedinet.com/ |

## Results

### Demographic distribution of neonates

During the four months of observational study at PIMS, 1,967 newborns were delivered to 1,889 mothers. The detailed distribution of neonates was explored with respect to various demographic parameters. Neonates were classified based upon their birth status, maternal origin, maternal language and rural urban status of parents. It also included the anthropometric parameters of neonates.

Among the 1,967 newborns, 1,857 were live-births, 24 stillbirths, and 86 dead (Figure 1A). The total 1,967 newborns included 1,010 males, 938 females and 19 with non- specific gender (include abortion cases also). A total of 108 newborns were diagnosed with congenital malformations. With respect to pregnancy outcome of the total 1,889 deliveries 93% were singleton and 7.47% multiple pregnancies (Figure 1B).



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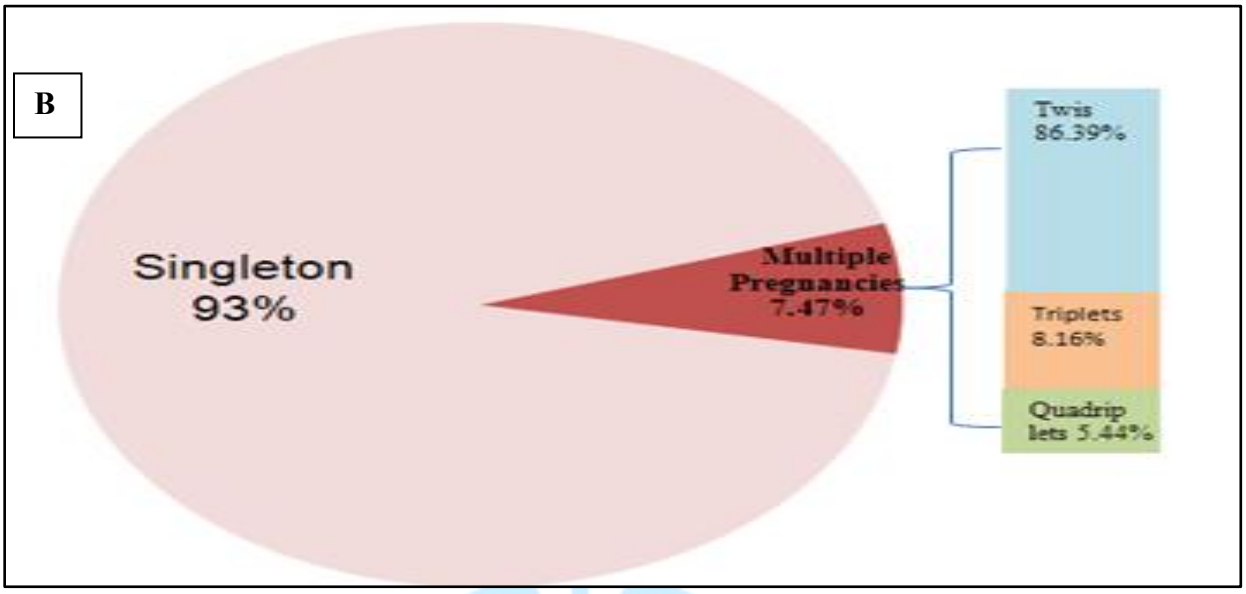
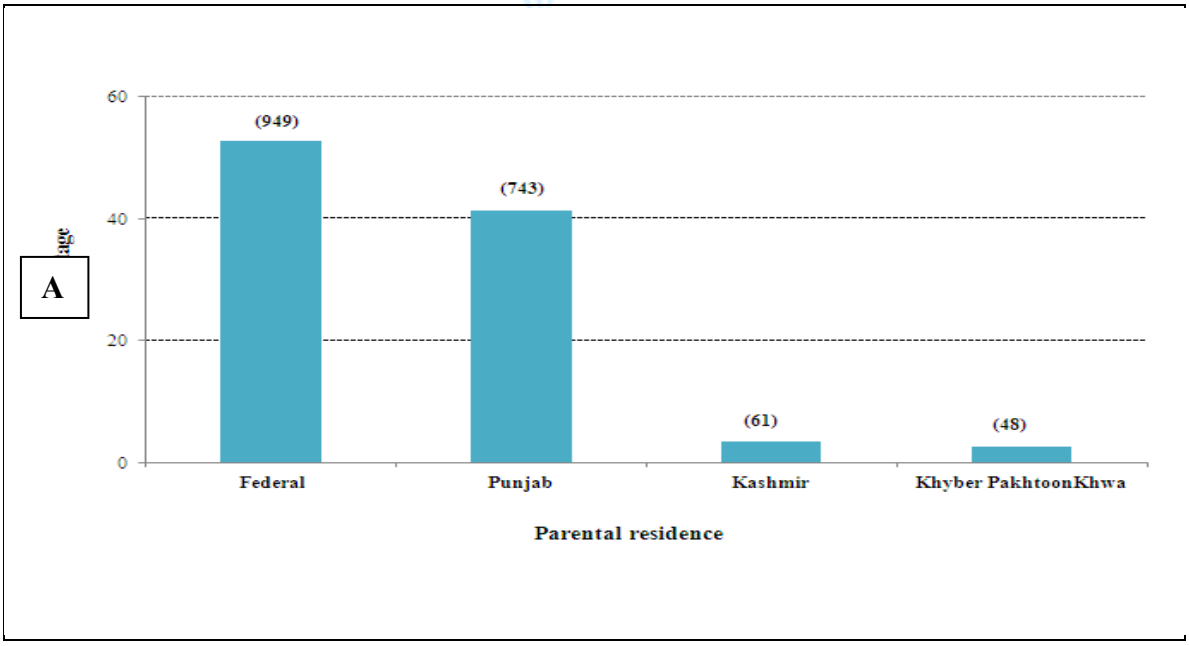



**Figure 1 (1A-1B): Distribution of Neonates delivering at PIMS (A), Distribution of Neonates according to pregnancy outcome (singleton, multiple)**

The neonates were categorized according to their parental residence (Figure 2A). Most of the mothers delivering at PIMS were form Federal Capital and Punjab. Of the total 1,967 neonates, 52.69% were from Federal Capital, 41.25% were from Punjab, 3.39% from Kashmir, and 2.67% from Khyber Pakhtoonkhwa (KPK).

The geographic distribution of neonates according to parental origin (rural/urban) was also observed. Most of the subjects delivering at PIMS were from rural areas (66.06%) and urban representatives were 33.94%. Figure 2B depicts the rural/urban status of neonate's parents in each province.



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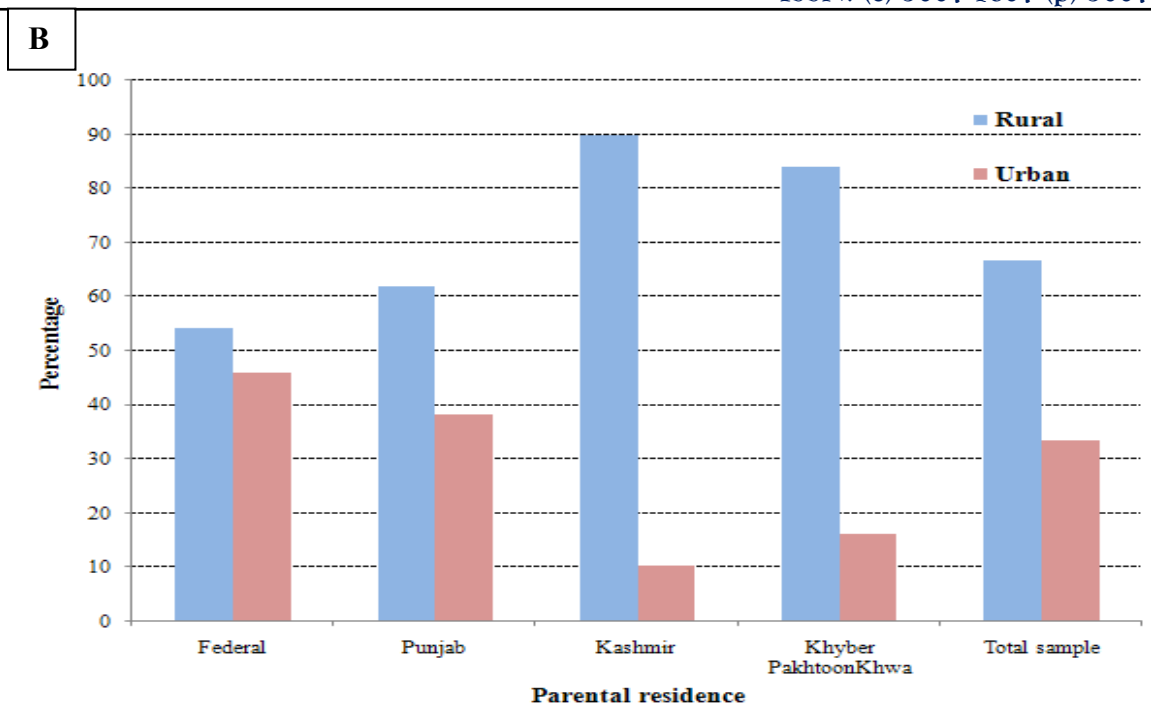


**Figure 2 (2A-2B): Distribution of neonates according to parental residence in Pakistan (A), Distribution of neonates according to rural/urban status of parental residence (B)**

Distribution of neonates according to maternal language was also studied (Figure 3). Punjabi speaking mothers were the most predominant (64.7%). Second and third significant linguistic representatives were Pahari and Pushtoo comprising 12.4% and 12% subjects, respectively. The other minor representative language groups were Urdu (3.9%) and Hindko (2.7%).

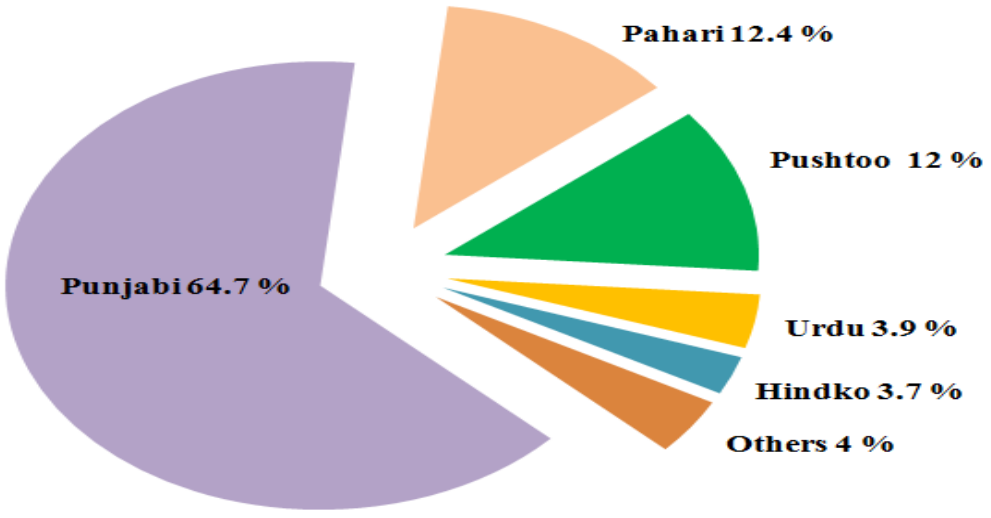


**Figure 3: Distribution of Neonates according to Maternal Language in different areas of Pakistan**

**Anthropometric parameters of neonates**

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Table 2 Anthropometric measurements of alive, normal and singleton neonates was provided in Table 2 with neonates with anomalies, dead, abortion, twins and triplets and quadruplets excluded. All the anthropometric measures were compared between males, females, and total neonates. Neonatal birth weight was divided into four ranges. The majority of the neonates (63%)-weight between 2.6-3.5 Kg with an average birth weight of 3.01±0.26 Kg.

The mean birth weight did not show a sex difference between the male and female babies. Body length of neonates was classified into five length ranges with maximum number of neonates having body length mean of 48.60/1.30 cm. Again sex difference was not observed in mean body length of neonates. Unless specified otherwise, head circumference or occipital-frontal circumference (OFC) were measured in centimeters and classified into three. The final anthropometric measurement of neonate was of APGAR score (appearance, pulse, grimace, activity and respiration) at the birth of the infant (1 min and 5 min). The APGAR score of the newborns of neonates is between 0 to 10. The mean value (8.60±0.50) of APGAR score at 5 minute in most of the neonates (86%) was 8.1±0.48 in the males and 8.50±0.49 in the females with no apparent sex difference in the mean score of APGAR.

**Table 2: Physical and biological parameters of alive, singleton and normal neonates**

| Neonatal Parameters | Male | | Female | | Total | |
|---|---|---|---|---|---|---|
| | No. | Mean ± SD | No. | Mean ± SD | No. | Mean ± SD |
| **Weight Intervals (Kg)** | | | | | | |
| 0.5 - 1.5 | 19 | 1.31±0.23 | 31 | 1.31±0.17 | 50 | 1.31±0.19 |
| 1.6 - 2.5 | 187 | 2.23±0.25 | 198 | 2.24±0.26 | 386 | 2.24±0.25 |
| 2.6 - 3.5 | 530 | 3.02±0.26 | 484 | 3.00±0.26 | 1014 | 3.01±0.26 |
| 3.6 - 4.5 | 92 | 3.79±0.20 | 67 | 3.78±0.18 | 159 | 3.78±0.19 |
| Total | 830 | 2.89±0.56 | 782 | 2.80±0.58 | 1,609 | 2.85±.57 |
| | | | | | | |
| **Body Length Ranges (cm)** | No. | Mean ± SD | No. | Mean ± SD | No. | Mean ± SD |
| 35 – 40 | 4 | 37.82±2.90 | 8 | 38.80±1.81 | 12 | 38.41±2.10 |
| 41 – 45 | 46 | 43.80±1.40 | 43 | 44.01±1.36 | 89 | 43.93±1.36 |
| 46 – 50 | 398 | 48.60±1.30 | 430 | 48.60±1.30 | 828 | 48.60±1.30 |
| 51 – 55 | 288 | 52.30±1.25 | 219 | 52.12±1.25 | 507 | 52.20±1.25 |
| 56 – 60 | 11 | 56.50±0.70 | 8 | 56.30±0.56 | 19 | 56.41±0.60 |
| Total | 747 | 49.79±2.88 | 708 | 49.39±2.80 | 1,455 | 49.60±2.85 |
| | | | | | | |
| **Head Circumference Ranges (cm)** | No. | Mean ± SD | No. | Mean ± SD | No. | Mean ± SD |
| 25 – 30 | 16 | 29.22±1.31 | 19 | 28.71±1.60 | 35 | 28.91±1.55 |
| 30 – 35 | 658 | 33.61±1.00 | 622 | 33.50±1.00 | 1280 | 33.60±1.00 |
| 35 – 40 | 57 | 36.40±0.86 | 49 | 36.30±0.90 | 106 | 36.40±1.10 |
| Total | 731 | 33.71±1.41 | 690 | 33.59±1.50 | 1,421 | 33.67±1.47 |
| | | | | | | |
| **APGAR Score at 1 MIN.** | No. | Mean ± SD | No. | Mean ± SD | No. | Mean ± SD |
| 0.0 - 1.0 | 3 | 0.67±0.58 | | | 4 | 0.80±0.50 |
| 2.0 - 3.0 | 19 | 2.68±0.48 | 16 | 2.31±0.47 | 35 | 2.50±0.50 |
| 4.0 - 5.0 | 38 | 4.45±0.50 | 45 | 4.40±0.49 | 83 | 4.40±0.50 |
| 6.0 - 7.0 | 405 | 6.74±0.44 | 389 | 6.69±0.46 | 794 | 6.71±0.50 |

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| | | | | | | |
|---|---|---|---|---|---|---|
| 8.0 - 9.0 | 292 | 8.02±0.15 | 269 | 8.01±0.12 | 561 | 8.01±0.20 |
| Total | 758 | 6.99±1.24 | 719 | 6.93±1.23 | 1,477 | 6.96±1.23 |
| | | | | | | |
| **APGAR Score at 5 MIN.** | No. | Mean ± SD | No. | Mean ± SD | No. | Mean ± SD |
| 0.0 - 1.0 | 2 | | | | | |
| 2.0 - 3.0 | 2 | 0.67±0.58 | 3 | 3 ±0 | 5 | 2.84±0.41 |
| 4.0 - 5.0 | 10 | 4.81±0.40 | 4 | 5 ±0 | 14 | 4.92±0.48 |
| 6.0 - 7.0 | 46 | 6.58±0.49 | 60 | 6.70±0.44 | 106 | 6.70±0.50 |
| 8.0 - 9.0 | 660 | 8.1±0.48 | 621 | 8.50±0.49 | 1,281 | 8.60±0.50 |
| 10.0 - | 36 | 8.02±0.15 | 32 | 10.00 ±0 | 68 | 10.00 ±0.0o |
| Total | 756 | 8.46±1.01 | 720 | 8.44±0.88 | 1,474 | 8.45±0.95 |

**Dysmorphology profile of neonates**

Out of 1,889 deliveries, 108 (5.7%) neonates were diagnosed with congenital malformations. These anomalies were categorized according anatomical systems involved. Distribution of these anomalies is shown in Figure 4 According to ICD-10 the anomalies were classified and ranked in descending order of frequency which is as follow: musculoskeletal defects 24.3 % (prevalence 13.211, CI 0.222-0.259), central nervous system defects 22.43% (prevalence 12.195, CI 0.201-0.238), urogenital system defects 14.95% (prevalence 8.130, CI 0.134-0.165), cardiovascular anomalies 12.15% (prevalence 6.606, CI 0.105-0.134), syndromic anomalies 10.28% (prevalence 5.589, CI 0.088-0.115), orofacial defects 7.48% (prevalence 4.065, CI 0.058-0.081), digestive system problem 6.54% (prevalence 3.557, CI 0.049-0.070), respiratory defects 0.93% (prevalence 0.508, CI 0.005-0.014), and others 1.87% (prevalence 1.016, CI 0.014-0.026). Multiple congenital anomalies were counted only once by major organ system involved.

Overall prevalence of congenital malformations was 54.9/1,000. The frequency, prevalence, proportion and 95% confidence interval was calculated for individual anomaly in each group (Table 3). Congenital anomalies of musculoskeletal system were most common (26/108) and in this group common anomaly was club foot accounted 10 of the total musculoskeletal anomalies (10/26). Among 10 cases of club foot three cases were associated with genitalia problem and CNS defects. Congenital nervous system anomalies were the second most common group of anomalies (24/108), with hydrocephaly accounting for 15 of the 24. Eight cases of hydrocephaly were isolated, whereas seven were linked with other nervous system abnormalities. Anomalies of the urogenital system were the third most prevalent anomaly, accounting for 16 of the total detected anomalies. The most common aberration in this group was underdeveloped genitalia, which affected 12/16 instances. Congenital abnormalities of the circulatory, orofacial, digestive, and respiratory systems were also found in modest numbers (Table 3).

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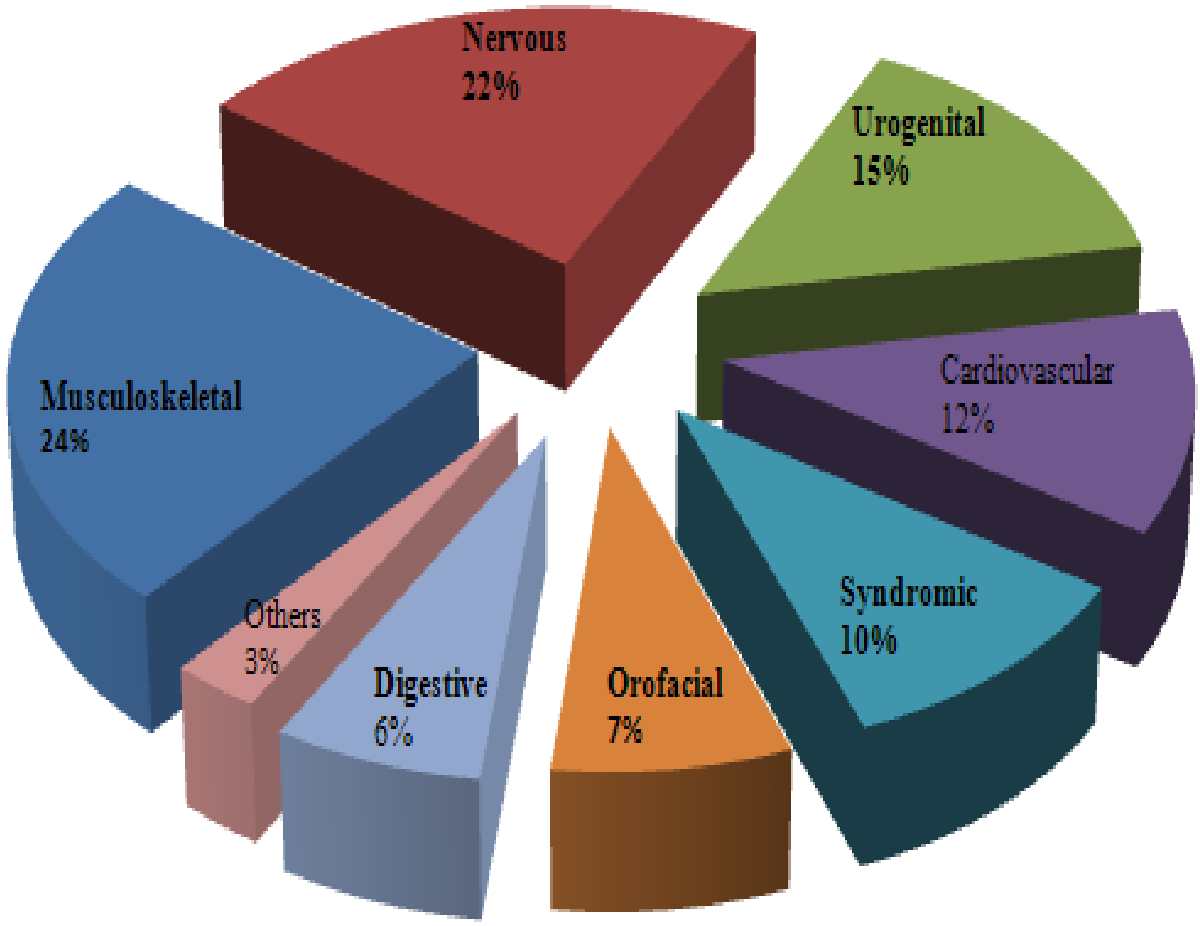


**Figure 4: Percent distribution of total anomalies in Islamabad City Pakistan**

**Table 3: Distribution and prevalence of Congenital Malformations (n=108)**

| Congenital malformation | No | %age | Prevalence | Proportion | CI | ICD code | OMIM |
|---|---|---|---|---|---|---|---|
| **Musculoskeletal system anomalies** | **26** | **24.3** | **13.211** | **0.241** | **0.222-0.259** | **Q65-Q79** | |
| Club foot | 10 | 9.35 | 5.081 | 0.093 | 0.077-0.102 | Q66 | 119800 |
| Skeletal and limb defect | 7 | 6.54 | 3.557 | 0.065 | 0.049-0.070 | | |
| Polydactyly | 6 | 5.61 | 3.049 | 0.056 | 0.049-0.070 | Q69 | 603596 |
| Brachydactyly | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q69 | 112500 |
| Abdomen distended | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | | |
| Erbs palsy | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q65-Q79 | |

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|---|---|---|---|---|---|---|---|
| **Nervous system defects** | **24** | **22.43** | **12.195** | **0.222** | **0.201-0.238** | **Q00-Q07** | |
| Hydrocephalous | 8 | 7.48 | 4.065 | 0.074 | 0.058-0.081 | Q00-Q07 | 236600 |
| Hydrocephalous associated with other CNS defects | 7 | 6.54 | 3.557 | 0.065 | 0.049-0.070 | Q00-Q07 | |
| Anencephaly | 4 | 3.74 | 2.033 | 0.037 | 0.031-0.048 | Q00-Q07 | 206500 |
| Meningeocele | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | Q00-Q07 | |
| Micro cephaly | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | Q00-Q07 | |
| Spina bifida | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q00-Q07 | |
| **Urogenital system anomalies** | **16** | **14.95** | **8.13** | **0.148** | **0.134-0.165** | **Q50-Q64** | |
| Underdeveloped genitalia | 12 | 11.21 | 6.098 | 0.111 | 0.096-0.123 | Q50-Q64 | |
| Kidney problem | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | Q80-A89 | 173900 |
| Epispadiasis | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q50-Q56 | 607306 |
| Ambigous genitalia | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q50-Q64 | |
| **Cardiovascular defects** | **13** | **12.15** | **6.606** | **0.12** | **0.105-0.134** | **Q30-Q34** | |
| Congenital Heart defect | 10 | 9.35 | 5.081 | 0.093 | 0.077-0.102 | Q30-Q34 | |
| VSD | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | Q30-Q34 | |
| Cardiomegaly | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q30-Q34 | |
| Syndromic | 11 | 10.28 | 5.589 | 0.102 | 0.088-0.115 | | |
| **Orofacial anomalies** | **8** | **7.48** | **4.065** | **0.074** | **0.058-0.081** | **Q10-Q18** | |
| Skin problem/Ichthyosis, peeling of skin | 3 | 2.8 | 1.524 | 0.028 | 0.022-0.037 | Q80 | 607602 |
| Isolated cleft lip | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | Q35-Q37 | 600625 |
| Cleft palat | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q35-Q37 | 119540 |
| Cleft lip with cleft palat | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q35-Q37 | |
| Turned ears | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | Q10-Q18 | |
| **Digestive system anomalies** | **7** | **6.54** | **3.557** | **0.065** | **0.049-** | **Q38-** | |

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|---|---|---|---|---|---|---|---|
| | | | | | 0.070 | Q45 | |
| Transesophageal Fistula | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | | |
| Deodanal Atresia/Esophagal atresia | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | | |
| Transesophageal Fistula/duedenal atresia | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | | |
| Esophagocele | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | | 189960 |
| **Respiratory system anomalies** | **1** | **0.93** | **0.508** | **0.009** | **0.005-0.014** | **Q30-Q34** | |
| Lung failure | 1 | 0.93 | 0.508 | 0.009 | 0.005-0.014 | | |
| Others | 2 | 1.87 | 1.016 | 0.019 | 0.014-0.026 | | |

Sex distribution of congenital malformations is shown in Table 4. The total 1,967 newborns includes 1010 Males, 938 Females and 19 with gender not identified (includes abortion cases also). The male neonates was found to have more congenital malformations than female neonates, out of 1,010 males 59 (5.5%) were reported with congenital malformations and 46 (4.9%) females were diagnosed with congenital malformations of the total 938 females.

The frequency of CM in live and still births is shown in Table 5. In this study, there were 105 stillbirths or neonatal deaths among 1,967 babies (5.3%). The frequency of CM in live births was 4.6%, whereas it was 20% in stillbirths and neonatal deaths. A statistically significant difference was reported between the frequency of CM in stillbirth and live birth ($p<0.0001$).

**Table 4: Prevalence of Congenital malformations by sex of neonates**

| Sex | No. of newborn delivered | No. with CM | Percentage of CM |
|---|---|---|---|
| Male | 1,010 | 56 | 5.54 |
| Female | 938 | 46 | 4.90 |
| Sex not known | 19 | 6 | 27.27 |
| Total | 1,967 | 108 | 5.49 |

**Table 5: Frequency of congenital malformations in live birth and still birth**

| No. of newborn delivered | Total | No. of newborn with CM | Percentage of CM |
|---|---|---|---|
| Live birth | 1,857 | 87 | 4.67 |
| Still birth/neonatal mortalities | 110 | 21 | 20.00 |
| All birth | 1,967 | 108 | 5.49 |

**Discussion**

Congenital malformations (CMs) remain a major contributor to neonatal morbidity, long-term disability, and infant mortality worldwide, particularly in low- and middle-income countries where access to preventive care, early diagnosis, and structured surveillance systems is limited (Christianson et al., 2018; Dolk et al., 2021). In Pakistan, congenital anomalies continue to represent a substantial yet under-recognized public health burden, largely due to fragmented reporting systems and reliance on hospital-based

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data (Masood et al., 2011; Hasan et al., 2010). The present hospital-based study provides important baseline evidence on the prevalence, system-wise distribution, and neonatal correlates of congenital malformations from a tertiary care setting in Islamabad, a region for which published data remain scarce.

The overall prevalence of congenital malformations observed in this study was higher than that reported in several earlier Pakistani studies (Shamim et al., 2010; Masood et al., 2011), though comparable to rates reported from other tertiary care centers in South Asia (Taksande et al., 2020; Bhide and Kar, 2018). Such variability in prevalence estimates may reflect differences in study design, inclusion criteria, diagnostic capacity, and timing of anomaly detection. Hospital-based studies, particularly those conducted in referral centers, tend to report higher prevalence due to the concentration of complicated pregnancies and high-risk deliveries (Dolk et al., 2020). Nonetheless, these studies play a critical role in settings where population-based congenital anomaly registries are not yet established.

In the present study, musculoskeletal anomalies constituted the most frequently affected system, followed closely by central nervous system (CNS) anomalies. This pattern aligns with reports from other hospital-based studies conducted in Pakistan and neighboring countries, where musculoskeletal and neural defects consistently rank among the most commonly reported categories (Jabeen, 2011; Parker et al., 2019). The predominance of musculoskeletal anomalies may be partly attributable to their relative ease of clinical detection at birth and higher compatibility with life, compared with internal organ anomalies that may remain undiagnosed during the early neonatal period (Blencowe et al., 2018). Among CNS anomalies, hydrocephaly emerged as the most frequent diagnosis, a finding consistent with previous regional studies (Hasan et al., 2010; Nafees et al., 2003). Nutritional factors, particularly inadequate periconceptional folate intake, have been widely implicated in the etiology of neural tube defects and may contribute to this observed burden (Kancherla et al., 2019).

The system-wise distribution of anomalies observed in this study differed from that reported in some earlier Pakistani studies, where gastrointestinal or CNS anomalies predominated (Shamim et al., 2010; Jahangir et al., 2009). Such heterogeneity underscores the influence of regional, environmental, nutritional, and healthcare-related factors on the epidemiology of congenital malformations. Differences in referral practices, availability of diagnostic imaging, and clinician expertise may further contribute to variation in reported patterns across studies (Moorthie et al., 2018).

A higher frequency of congenital malformations was observed among male neonates compared with females, a trend that has been consistently reported in multiple epidemiological studies (Egbe et al., 2015; Sitkin et al., 2021). Although the biological basis of this sex disparity remains incompletely understood, it has been suggested that male fetuses may be more vulnerable to adverse intrauterine conditions and developmental disruptions (Parker et al., 2019). Additionally, congenital malformations were significantly more common among stillbirths than live births, highlighting the substantial contribution of structural anomalies to adverse pregnancy outcomes. Similar findings have been reported in studies from both high-income and low-income settings, emphasizing the critical role of congenital anomalies in perinatal mortality (Best et al., 2017; Blencowe et al., 2018).

Maternal age distribution in the present study showed that most affected neonates were born to mothers in the mid-reproductive age group, a finding that reflects the general childbearing pattern in Pakistan rather than indicating a specific age-related risk. Previous studies have demonstrated that both extremes of maternal age may be associated with an increased risk of congenital anomalies; however, this relationship often varies depending on anomaly type and population characteristics (Kancherla et al., 2019; Feldkamp et al., 2017). Similarly, congenital malformations were observed among both primigravida and multigravida mothers,

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consistent with earlier reports from Pakistan and other South Asian countries (Masood et al., 2011; Taksande et al., 2020).

The findings of this study must be interpreted in light of certain limitations. As a hospital-based study, the results may not be fully representative of the general population, particularly rural communities with limited access to tertiary healthcare facilities. Additionally, some maternal and neonatal data were incomplete due to logistical constraints and variations in record-keeping practices. Despite these limitations, the study provides valuable insight into the burden and pattern of congenital malformations in a major referral hospital serving a diverse population.

Overall, the high prevalence of congenital malformations observed in this study underscores the urgent need for the development of structured congenital anomaly surveillance systems in Pakistan. Strengthening antenatal screening services, improving periconceptional nutritional interventions, and enhancing neonatal diagnostic capacity are essential steps toward reducing the burden of congenital anomalies. The findings of this study contribute important baseline data that can inform public health planning and support the establishment of regional and national congenital anomaly registries.

## Conclusion

This study highlights congenital malformations as an important contributor to adverse neonatal outcomes in a tertiary care setting in Islamabad. The relatively high prevalence observed, along with the predominance of musculoskeletal and central nervous system anomalies, underscores the continuing public health significance of congenital malformations in Pakistan. The higher frequency of anomalies among male neonates and stillbirths further emphasizes their role in perinatal morbidity and mortality. The variation in the pattern of anomalies compared with previous national and regional studies reflects the influence of local demographic, environmental, and healthcare-related factors and reinforces the need for context-specific data. These findings underscore the importance of strengthening antenatal screening, improving neonatal diagnostic capacity, and establishing structured congenital anomaly surveillance systems. Such measures are essential for early identification, informed public health planning, and the reduction of preventable neonatal morbidity and mortality.

## Declarations

### Consent for publication

Not applicable

### Availability of data and materials

The data will be available from author on reasonable request

### Competing Interest

All authors declare that there are no competing interests.

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