# Frequency of Fetal Congenital Anomalies and Associated Risk Factors Observed in Pregnant Women in a Public Sector Hospital of Karachi

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## Abstract

**Objective:** To identify congenital anomalies and their risk factors in pregnant women presenting at a public sector hospital of Karachi.

**Methods:** A cross sectional study was conducted in the department of Obstetrics, Orangi town, a public sector hospital in Karachi, from January 2014 to December 2015 after obtaining institutional ethical approval. Convenience sampling was used in this study. All those women who presented with diagnosed congenital anomalies consecutively either in outpatient department or came directly to ward for the termination of pregnancy were included after their informed verbal and written consent. A predesigned proforma was used to take a complete history, associated risk factors and relevant information of the congenital anomalies. The data was analyzed quantitatively by using SPSS version 20.

**Results:** A total of 11,946 women were admitted in Obstetrics department during the study period. Data was calculated on daily bsis from outpatients' and inpatients' records of the hospital. The data was analysed by using descriptive statistic; frequency distribution technique. Out of the total 87 (0.72%) presented with different fetal congenital anomalies. Among them, 34 (39.0%) were male, 47 (54.0%) were female and 6 (7.3%) of the fetuses with gender not clear. Majority of the women who delivered babies with congenital anomaies, were not taking folic acid during pregnancy 49 (56.3%). Maternal age parameter revealed that out of 87 women, 37 (42.2%) were between the age of 26-30 years. Majority of patients belonged to poor socioeconomic status i.e. 71 (81.6%). Congenital anomalies presented more in fetuses of multiparous women than in those with first pregnancy.Most commonly involved body systemwere central nervous system 45 (51.7%) followed by musculoskeletal 16 (18.3%), gastrointestinal system 15 (17.2%), congenital syndromes 7 (8.0%), genitourinary 2 (2.4%), cardiac 1 (1.2%) and respiratory system 1 (1.2%).

**Conclusion:** The central nervous system was affected the most. Healthcare authorities must consider primary prevention in the form of vaccination such as Rubella vaccine, nutrition and drugs such as folic acid to reduce preventable congenital anomalies. It will further help in developing awareness amongst women.


**Keywords:** Congenital anomalies, fetuses, Pakistan, frequency, risk factors.

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## Introduction

Worldwide, congenital anomaly is a global health issue and plays a major role in perinatal mortality and morbidity, as well as childhood death, chronic illness and disability[1,2]. Several studies have reported that mostfamilies health care system and even societies bear long life ill health effects due to congenital anomalies[2-5]. According to Parmar et al, 66% of congenital anomalies are of unknown etiology[2]. Congenital anomalies or birth defects can be defined as structural or functional anomalies including metabolic disorders, which are present at the time of birth[2]. Many studies have confirmed that 3rd to 8th weeks of gestation are the most important period for the development of organs and any alteration during this period causes malformations[2-6].

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The incidence of congenital anomaly varies in different countries. However, it ranges between 3 to 7%. It is estimated that 276,000 infant mortality occurs annually due to congenital anomalies globally[4]. It is speculated that 20 -30% infant mortality and 30-50% deaths after neonatal period occurs due to congenital defects[3]. In the UK an estimated number of congenital anomaly of 15, 966 was recorded[7]. World Health Organization (WHO) has estimated that in Pakistan 26,627 or 2.08% deaths occurs due to congenital defects in 2011 and this has increased by 2.34% in the year 2014[8,9].

The most prevalent congenital anomalies reported in a study were heart defects followed by orofacial, cleft lip, Down syndrome and neural tube defects[10]. Another study by Malhotra and Thapar[11], indicates that the most common anomalies were related to gastrointestinal tract followed by the nervous system. However, several other studies reported neural tube defects as the most frequent anomaly[2,3,12-16,18-20].

The etiological risk factors for congenital malformation include genetic and environmental factors, age of the mother, socioeconomic status, maternal exposure to teratogen, radiation,maternal illness, smoking and alcohol consumption as well as consanguinity[3-5]. It is also evident that congenital defects occur more in low birth weight and consanguineous marriages[1,5,19]. In addition, studies have reported that congenital anomalies are more common in male fetuses and in elderly mothers[18,21,22]. Moreover, lack of folate supplementation, smoking and alcohol consumption were also most common risk factors of congenital anomalies[5,14,16]. The public health rationale behind this study was to confirm the main risk factors of fetal congenital anomalies which will help in implementation of effective intervention in prevention of congenital anomalies.

The objective of this study was to identify the most commonly seen congenital defects in pregnant women of a public sector hospital of Karachi, their cause and the main risk factors that will further help in developing awareness amongst women.

## Patients and Methods

A cross sectional hospital based study was conducted in the department of Obstetrics, Orangi town, a public sector hospital in Karachi, from January 2014 to December 2015. The institutional ethical approval was initially taken. A total of 11,946 women were admitted in Obstetrics department during the study periodof which 100 had congenital anomalies. However of the 100 questionnaires issued, only 87 were valid and 13 excluded because these patients were delivered outside study setup.

Consecutive convenience sampling was used for the purpose of this study. All those women who presented with diagnosed congenital anomalies either in outpatient department or came directly toward for the termination of pregnancy were included after their informed verbal consent. Those women who visited outpatient department had given consent for study and proforma was filled but did not deliver in study setup were excluded from study. A predesigned proforma was used to take a complete history of the women to find out the associated risk factors of congenital anomalies. The variable included are shown in Table 2 and 3. After history and examination, delivery was conducted according to the standard protocol followed in labour room. All the relevant information of the anomalies was documented in the Performa. Karyotyping was not performed due to non-affordability.

The data was analyzed quantitatively by using SPSS version 20. Descriptive statistics were tabulated in the form of frequency with percentages.

## Results

A total of 11,946 women admitted in Obstetrics department during the study period. Data was calculated on daily basis from outpatients and in patient records of hospital. The data was analyses by using descriptive statistics; frequency distribution technique. Out of the total 87 (0.72%) presented with different congenital anomalies fetuses. Among them, 34 fetuses were male (39.0%), 47 (54.0%) were female fetuses and 6 (7.3%) having no clear

gender identification. Among all,majority of the fetuses werewith weight >600 gmi.e. 34 (39.0%), Table 1. Regarding gestational age,majority of fetuses were born between 3rd trimester 26-30 weeks 24 (29.2%) 11 (13.4%) between 31-35 weeks and 10 (12.17%) were more than 35 weeks followed by 2nd trimester 21 to 25 weeks 22 (26.8%) and 17 (19.5%) less than 20 weeks. Maternal age parameter revealed that out of 87 women 37 (42.2%) were between 26-30 years Table 1. Congenital anomalies presented more in fetuses of multiparous women, those having five or more children 27(31.0%) than in those with first pregnancy 14 (17.0) (Table 1). Consanguinity was not so high in present study 35 (40.2%) couples, family history of congenital anomaly was positive in 21 (24.1%). Significant number of women 67 (78.1%) had no previous affected child. Only 19 (23.1%) of women gave a history of having a child with congenital anomaly. History of folic acid intake was positive only in 38 (46.3%) while majority 44 (53.6%) were not taking folic acid during pregnancy. Majority of patients were poor 71 (81.6%) assessed by their occupation and income. Only 12 (13.7%) women were exposed with infection like chicken pox, measles and syphilis in first trimester infection. On the other hand, great number 75 (86.2%) of women had no history of such infection in first trimester. Mothers with Rh positive blood group had more congenital anomalies 74 (85.0%) than those of Rh negative Table 2. Most commonly involved body system were central nervous system 45 (51.7%) followed by musculoskeletal 16 (18.3%), gastrointestinal system 15 (17.2%), congenital syndromes 2 (2.4%), genitourinary 2 (2.4%), cardiac 1 (1.2%) and respiratory system 1 (1.2%), Table 3.

## Discussion

Congenital anomaly plays a significant role in perinatal mortality and morbidity, as well as childhood death, chronic illness and disability[1,2]. It is reported that most families' health care system and even societies bears lifelong ill health effects due to congenital anomalies[2-5].

A total of 87 women presented with congenital anomalies were seen during the study period. Therefore frequency of congenital anomalies in our study is 0.72%. Which corresponds to the study of Liaquat Memorial Hospital Kohat[24] of 0.97%. On the other hand study from Banglore and Egypt reported a high frequency 14.25% and 9.7% respectively[20,25]. The variations in the figures from different settings may be due to specific risk factors like ethnicity, geographical distribution, and consanguinity, socioeconomic status, cultural and nutritional factors.

In our study, congenital anomalies were found more in female fetuses than male fetuses (54.0% females and 39.0% males). This could be because present study did not represent the whole population. Similar to the Gul, Jabeen and Khan's study[24]. in contrast, number of studies have reported male fetuses with congenital anomalies were more than female fetuses[3,17,21].

The mean weight suggested that most fetusesweight is normal in relation to gestational age[23] (Table 1). Present study did not find association of birth weight with congenital defects. Our findings are consistent with the findings of the study done in Civil Hospital Karachi[5]. This is surprising as several studies reported association between birth weight and congenital anomalies[1,3,12].

Age of the mother associated with congenital anomaly is considered important factors. Maternal parameter in our study presented congenital anomalies more common in age between 26 and 30 years (43.9%), which is similar with study done in India[18]. In contrast studies also reported that elderly mothers or age above 30 had high incidence of producing congenital malformed babies[3,17]. Whereas in our study only 3.6% women with age more than 35 years were found with congenital malformed fetuses. This could be because number of women in age group less than 35 years were few, hence this is a bias in our study design. However, this study suggests that women presenting with congenital anomalies in our setup were with other risk factors Most of the mothers in our study belonged to poor

**Table 1.** Demographic Characteristics (n= 87)

| Parameter | Frequency | Percentage % |
|---|---|---|
| Parity | | |
| G1 | 15 | 17.2 |
| G2 | 22 | 25.2 |
| G3-G4 | 23 | 26.4 |
| >G5 | 27 | 35.6 |
| Total | 87 | |
| Maternal Age | | |
| <20 years | 4 | 4.5 |
| 20-25 years | 14 | 16.0 |
| 26-30 years | 37 | 42.5 |
| 31-35 years | 26 | 29.8 |
| >35 years | 6 | 6.8 |
| Total | 87 | |
| Gestational age | | |
| <20 weeks | 17 | 19.5 |
| 21 to 25 weeks | 22 | 25.2 |
| 26 to 30 weeks | 24 | 27.5 |
| 31 to 35 weeks | 13 | 14.9 |
| >35 weeks | 11 | 12.6 |
| Total | 87 | |
| Birth weight | | |
| <400g | 16 | 18.3 |
| 400-500g | 14 | 16.0 |
| 500g-600g | 23 | 26.4 |
| >600g | 34 | 39.0 |
| Total | 87 | |
| Gender of baby | | |
| Male | 34 | 39.0 |
| Female | 47 | 54.0 |
| Gender not clear | 6 | 6.8 |
| Total | 87 | |

G = Gravida

**Table 2.** Risk factors (n=87)

| Parameter | Frequency | Percentage % |
|---|---|---|
| Maternal blood group | | |
| O+ | 27 | 32.9 |
| O- | 2 | 2.4 |
| A+ | 26 | 29.8 |
| A- | 3 | 3.6 |
| B+ | 18 | 21.9 |
| B- | 5 | 6.0 |
| AB+ | 3 | 3.6 |
| AB- | 3 | 2.4 |
| Total | 87 | |
| Consanguinity | | |
| Yes | 35 | 40.2 |
| No | 52 | 63.4 |
| Total | 87 | |
| Previous infected child | | |
| Yes | 19 | 23.1 |
| No | 68 | 78.1 |
| Total | 87 | |
| Family history | | |
| Yes | 21 | 24.1 |
| No | 66 | 75.8 |
| Total | 87 | |
| H/O Folic acid intake | | |
| Yes | 38 | 46.3 |
| No | 49 | 56.3 |
| Total | 87 | |
| Socioeconomic status | | |
| Poor | 71 | 81.6 |
| Average | 16 | 19.5 |
| Total | 87 | |
| First trimester infection | | |
| Yes | 12 | 13.7 |

**Table 3.** Distribution of congenital malformation (n=87)

| System | Malformation | Frequency | Percentage % |
|---|---|---|---|
| CNS | Hydrocephalus | 22 | 26.8 |
| | Anencephaly | 18 | 20.6 |
| | Meningocele | 3 | 3.6 |
| | Agenesis of corpus collosum | 1 | 1.2 |
| | Encephalocele | 1 | 1.2 |
| Total | | 45 | 51.7 |
| Musculoskeletal | | | |
| | Talipes | 3 | 3.6 |
| | Spina bifida | 7 | 8.0 |
| | Achondroplasia | 1 | 1.2 |
| | Skeletal dyplasia | 5 | 6.0 |
| Total | | 16 | 18.3 |
| Genitourinary | | | |
| | Posterior urethral valve | 1 | 1.2 |
| | Hydronephrosis | 1 | 1.2 |
| Total | | 2 | 2.4 |
| Cardiovascular | | | |
| | | 1 | 1.2 |
| Total | | 1 | 1.2 |
| Digestive | | | |
| | Omphelocele | 2 | 2.4 |
| | Imperforate anus | 1 | 1.2 |
| | Fetal ascites | 5 | 6.0 |
| | Esophageal atresia | 1 | 1.2 |
| | Duodenal atresia | 1 | 1.2 |
| | Cleft lip | 5 | 6.0 |
| | Cleft palate | | |
| Total | | 15 | 17.2 |
| Respiratory | Plural infusion | 1 | 1.2 |
| Total | | 1 | 1.2 |
| Congenital Syndromes | | | |
| | Arnold chari malformation | 1 | 1.2 |
| | Dandy walker syndrome | 1 | 1.2 |
| | Hydropfetalis | 5 | 6.0 |
| Total | | 7 | 8.0 |
| Grand Total | | 87 | |
| | | | |
| Gestational age | | | |
| <20 weeks | | 17 | 19.5 |
| 21 to 25 weeks | | 22 | 25.2 |
| 26 to 30 weeks | | 24 | 27.5 |
| 31 to 35 weeks | | 13 | 14.9 |
| >35 weeks | | 11 | 12.6 |
| Total | | 87 | |

| System | Malformation | Frequency | Percentage |
|---|---|---|---|
| Birth weight | | | |
| <400g | | 16 | 18.3 |
| 400-500g | | 14 | 16.0 |
| 500g-600g | | 23 | 26.4 |
| >600g | | 34 | 39.0 |
| Total | | 87 | |
| Gender of baby | | | |
| Male | | 34 | 39.0 |
| Female | | 47 | 54.0 |
| Gender not clear | | 6 | 6.8 |
| Total | | 87 | |

G = Gravida

and age may not be the contributing factor of congenital anomaly in these women.

We found more congenital anomalies in the fetuses of maltiparus women which was consistent with the earlier results[3]. This high frequency (Table 1) indicates that the congenital anomalies may increases as the birth order increases. Therefore, it seems that multiparous women are the ideal target group for preventive measure and public health education campaign.

Although our study found that significant number of women (Table 2) belong to Rh positive blood groups. We could not find any definite explanation for this.

It is reported at large that consanguineous marriages has close association with congenital anomalies[1,3-5,7] because of the homozygous expression of recessive genes inherited from blood relations (first cousin marriages)[3,4,7]. However, our results established contrary findings 63.4% of women had non consanguineous marriage, which was in accordance with earlier study done in Aga Khan University Hospital, Karachi[19]. Hence this information provides ground for further research in this area.

According to our study findings 23.1% women had history of previous affected child. This is further supported by the non-compliance of nutrients especially folic acid intake, multiparity and poor socioeconomic status in the study group.

Folic acid deficiency is known risk factor of having baby with neural tube defects1, 3-4, 5, 7, 19,24. In our study 46.3 % of women were non-compliant in folic acid intake during early pregnancy. In addition, in our study central nervous system was the most affected system 26.8 % hydrocephalus, 20.0 % anencephaly and 8.0% cases of spina bifida suggested strong association with folic acid deficiency. This suggested further need of implementation for improving diet of women particularly folic acid intake.

Most of the mothers in our study belonged to poor socioeconomic status. This could be one of the reasons for high frequency of congenital anomalies in our setting. These findings are consistent with the finding of study done at Karachi Civil hospital by Raza MZ et al[5].

Family history of congenital malformation was 24.1 percent in our study and majority 75.8% women had no family history. Which was further

consistent of with study finding of non-consanguineous marriages 63.4%. Findings are similar with study done in Kharian, Pakistan that only 22.1% were with consanguineous marriages[3].

In our study the most common affected system was the CNS followed by musculoskeletal and then GIT. This is also supported by other researches by Hussains et al[3]. In addition, several other studies have reported that CNS was the most affected system such as the studies from UK[15], Egypt[20], India[2,17,18] and Pakistan[12-14,16,19]. In contrast, the study by Butt et al reported that most common congenital anomalies were heart defects followed by orofacial, cleft lip, down syndrome and CNS defects[10]. Another study found gastrointestinal related defects were more common followed by CNS defects[11]. Similarly, a study from India reported musculoskeletal related anomalies were most common followed by CNS[22]. The limitations of our study were that it was secondary care hospital study therefore it is not representative of the community at large. In addition, interventions like serum alfa-fetoprotein, acetylcholinesterase, amniocentesis, karyotyping are not available at secondary care hospital center for early diagnosis of congenital malformations.

It is recommended that healthcare awareness campaigns should be available at all public sector hospitals, prenatal and antenatal classes should be available at all public health sectors, awareness regarding folic acid intake during women prenatal and antenatal visits should be the area of consideration and government should facilitate prenatal screening test at public health center.

## Conclusion

Congenital anomalies are frequent in our set up and CNS was the most commonly affected system. Female gender, multiparity, poor socioeconomic status, folic acid intake were associated risk factors. Knowledge of frequency and related risk factors of congenital anomalies are important to plan preventive measures at different levels by healthcare professionals.

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## Conflict of Interest

Authors have no conflict of interests and no grant/ funding from any organization for this study.

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