

# Prevalence of Congenital Birth Defects among Pediatric Patients of Interior Punjab

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## ABSTRACT

**Objective:** To find out the prevalence of congenital birth defects among the pediatric patients of Sialkot.

**Methods:** A descriptive cross-sectional study was conducted at the pediatrics medicine, pediatric surgery and obstetrics ward of Khawaja Muhammad Safdar Medical College and hospital Sialkot Pregnant women irrespective of the parity were enrolled in the study. Demographic variables, detailed history and examination before and after delivery were noted on a preformed proforma. The data was entered and analyzed through Statistical Package for Social Science (SPSS) version 20. p-value less than 0.05 was considered as significant.

**Results:** The mean age of the pregnant mothers included in the study was  $25.45 \pm 5.12$ . The most common age group affected was having age more than 30 years (58.1%) and majority were multiparous (52.4%). Cousin marriage was the most common factor leading to birth defects among their children and the result was significant (p-value  $\leq 0.05$ ). The prevalence of congenital birth defect was 14.7%. The cleft palate was the most common anomaly followed by hydrocephalus, cleft lip, down syndrome, polydactyly, undescended testes, club foot, microcephaly, meningomyelocele, ambiguous genitalia and bradydactyly.

**Conclusion:** The prevalence of congenital birth defect was 14.7% and cleft palate was the most common anomaly followed by hydrocephalus and down syndrome. The most common associated factor was cousin marriages.

## INTRODUCTION

The term congenital birth defect is labelled when there is developmental defect during intrauterine life, it may be the structural abnormality or the functional abnormality. Its diagnosis is simple and easy during prenatal life but if remains undiagnosed then diagnosis can be made soon after birth. Few of the diseases, like hearing defects cannot be diagnosed prenatally or even after birth, these types of defects can be diagnosed during infancy<sup>(1)</sup>.

The prevalence of congenital defects depends upon the level of development of a country, like in developed country its prevalence is bit low that is 39.1/1000 live births while in developing country like Pakistan, the prevalence is high 82/1000 live births<sup>(2-4)</sup>. Literature revealed that the most common congenital birth defect is congenital heart disease followed by neural tube defects including down syndrome<sup>(5)</sup>. Considering specifically Pakistan the prevalence is between 6-11% as per available literature<sup>(6,7)</sup>. Children with congenital abnormality, usually die during early childhood because of malfunctioning of body mechanism<sup>(7)</sup>. Majority of the congenital birth defects are sporadic as the underlying cause cannot be identify<sup>(8)</sup>.

There are multiple factors, including genetic and environmental factors, that can lead to congenital anomalies. Focusing Pakistan, the genetic causes like chromosomal disorders are rare as comparing environmental factors, that accompanies 10% of total burden of disease and the most common is frequent exposure of mother to infections<sup>(9)</sup>. Majority of middle east and Asian's prefer consanguineous marriages which is also a leading cause of congenital anomalies now a day<sup>(10)</sup>. Cousin marriage is a very common trend in Pakistan, specially the interior areas. So the aim of current study was to find out the prevalence of congenital birth defects among the pediatric patients of Sialkot.

## METHODOLOGY

A descriptive cross-sectional study was conducted at the pediatrics medicine, pediatric surgery and obstetrics ward of Khawaja Muhammad Safdar Medical College and hospital Sialkot during the period of April to September 2021. The study got ethical approval from the concerned institute. Sample size was calculated through Open Epi calculator and was 295. Pregnant women irrespective of the parity were enrolled in the study, prior to that informed consent was taken. Demographic variables, detailed history and

examination before and after delivery were noted on a preformed proforma.

The data was entered and analyzed through Statistical Package for Social Science (SPSS) version 20. All the numerical variables were presented as mean with standard deviation while categorical variables as frequency and percentages. To find out the association of numerical and categorical variables with the occurrence of congenital birth defects, the t-test and chi-square was used respectively. p-value less than 0.05 was considered as significant.

## RESULTS

The mean age of the pregnant mothers included in the study was  $25.45 \pm 5.12$  with average height and weight. The most common age group affected was having age more than 30 years (58.1%) and majority were multiparous (52.4%) with strongly significant p-value (p-value 0.001). Cousin marriage was the most common factor leading to birth defects among their children and the result was significant (p-value 0.000). About 48.9% pregnant women delivered through normal vaginal delivery but no significant association with congenital birth defect as reported in Table 1. The prevalence of congenital birth defect was 14.7% as shown in figure 1. The frequency of commonly occurring congenital anomalies was also noted, the cleft palate was the most common anomaly followed by hydrocephalus, cleft lip, down syndrome, polydactyly, undescended testes, club foot, microcephaly, meningomyelocele, ambiguous genitalia and bradydactyly as shown in Figure 2.

Table 1: Demographic characteristics of Pregnant mothers included in the study

Variables	Mean $\pm$ SD	%	p-value
Mean age (years)	25.45 $\pm$ 5.12	-	0.001
Mean years of miscarriage	4.49 $\pm$ 3.1	-	0.04
Mean weight (kg)	67.11 $\pm$ 7.45	-	0.59
Mean height (meters)	1.01 $\pm$ 0.38	-	0.721
Age groups			
$\geq 30$ years	-	58.1	0.001
$\leq 30$ years	-	41.9	
Parity			
Multiparous	-	52.4	0.382
Nulliparous	-	33.2	
Grandmultiparous	-	14.4	
Cousin marriage	-	69.7	0.000
Normal vaginal delivery	-	48.9	0.869

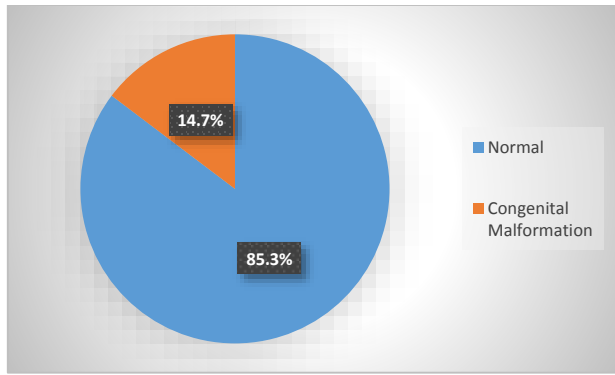


Figure 1: Prevalence of congenital birth defect

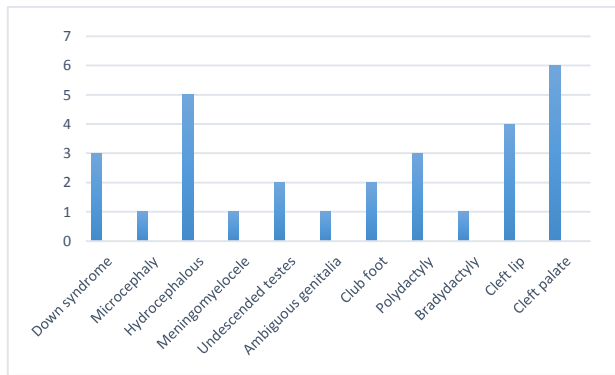


Figure 2: Frequency of different congenital anomalies

**DISCUSSION**

Congenital birth defects not only affect the child but the whole family by affecting the quality of life. Globally about 8 lac children affected with congenital birth defect per annum, out of them 3 lac die under 5 years of age while remaining survive with disability<sup>(11)</sup>. Due to the current burden of disease, multiple researches are going on to minimize the incidence rate of congenital birth defects by inventing new medical technologies for early diagnosis so that preventive measures can be taken before time<sup>(12)</sup>.

Early detection of congenital anomalies is not easy in developing countries, because of lack of facilities so prevalence rate is rising day by day as compared to developed countries<sup>(13)</sup>. The current study is important to highlight the prevalence of congenital birth defect and results showed its about 14.7%, comparing with another study done in Pakistan reported prevalence of 11.3% in 2019<sup>(13)</sup>. This is an alarming situation that the prevalence of congenital birth defects rises to 1% per year from 2019 to 2022 which is an economical burden. Current study found that the cleft palate was the most common anomaly followed by hydrocephalus, cleft lip, down syndrome, polydactyly, undescended testes, club foot, microcephaly, meningomyelocele, ambiguous genitalia and bradydactyly. There are some variations in the frequency of different congenital birth defects in international literature. Asendi et.al found the gastrointestinal disorders as the most common birth defect followed by neurological disorders<sup>(14)</sup>. Current findings are also supported by another study from Spain<sup>(13)</sup>.

Looking over Pakistan, a developing country, the Fouzia et.al. conducted a study in Karachi and found that the neural tube defects are the most commonly occurring congenital anomaly with prevalence of 65%<sup>(15)</sup>. Another study done in interior done in interior Sindh reported 11.3% cases of congenital abnormalities, out of which majority cases were of CNS and GIT malformations<sup>(13)</sup>.

There is high prevalence of congenital abnormalities in Pakistan because of cousin marriages<sup>(16)</sup> and the prevalence is increasing day by day as current study compared with previous

study and found 1% rise in prevalence rate per year. Current study reported that 69.7% cases of congenital anomalies had cousin marriages in their parents. The ratio of cousin marriages is even higher in Peshawar i.e. near about 80%<sup>(17)</sup>. Our neighboring country, India has a very low ratio of cousin marriage (1-4%), just because of their religious beliefs<sup>(18,19)</sup>.

The prevalence of congenital defects is varying between developed and developing countries because of the two main reasons, including cousin marriages and failure of prenatal diagnosis because of poor medical facilities to remote areas of country.

**CONCLUSION**

The prevalence of congenital birth defect was 14.7% and cleft palate was the most common anomaly followed by hydrocephalus and down syndrome. The most common associated factor was cousin marriages.

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