

## **Original Research Article**

### **Frequency of common congenital anomalies among neonates in tertiary care hospital: A cross-sectional study**

#### **Abstract**

**Aim:** To determine the frequency of common congenital anomalies among neonates in tertiary care hospital

**Study design:** A cross-sectional study

**Place and Duration:** Children Hospital Larkana Pakistan from April to October 2018

**Methodology:** One hundred twenty-four newborns with congenital anomalies were included in this study after taking parental consent. All the newborns were assessed by prenatal, natal, and postnatal history, general physical examination, and relevant investigations like ultrasound abdomen, echocardiography, and CT Scan Brain were done; Questionnaire was filled out immediately within 24 hours after hospital admission

**Results:** The mean maternal age of the patients was  $24.16 \pm 4.13$  years. Central nervous system (CNS) anomalies were observed in 21% of cases. Neural tube defects like hydrocephalus 6.5%, sacrococcygeal teratoma 2.4%, meningomyelocele 11.3%, and encephalocele 0.8%. Cardiovascular system (CVS) anomalies were found in 11.3% cases in which cyanotic 2.4%, (2 were Tetralogy of Fallot and 1 was Transposition of great arteries) Acyanotic 8.9% (10 Ventricular septal defects and 1 atrial septal defect). Gastrointestinal tract (GIT) Anomalies

(39.5%) like, duodenal atresia (0.8%), anorectal malformations (25%), Pyloric Stenosis (0.8%), exomphalocele (5.6%), diaphragmatic hernia (2.4%), Hirschsprung disease (4.8%).

Genitourinary system abnormalities (16.9%) like Hypospadias (14.5%), undescended testes (0.8%), cloacal exstrophy (0.8%) and hydrometrocolpos (0.8%). Musculoskeletal abnormalities (11.3%) like clubfoot (0.8%), developmental dysplasia of Hip (0.8%), polydactyly (1.6%) and cleft lip & palate (8.1%) of cases.

**Conclusion:** Congenital abnormalities are prevalent in our setup, and the CNS is the most commonly involved system, followed by the gastrointestinal system, genitourinary system, cardiovascular system, and musculoskeletal system. The prevalence and pattern of congenital defects must be known in order for healthcare providers to create preventive interventions at various levels.

**Key Words:** Congenital Anomalies, CNS anomalies, Neural tube defects, newborns

## Introduction:

Birth defects and congenital malformations are terms used to describe congenital anomalies.

"Congenital abnormalities can be defined as any structural or functional aberration (e.g. metabolic diseases) that arises during intrauterine life and can be diagnosed prenatally, at birth, or postnatally." [1] The prevalence of congenital anomalies varies from one geographic area to other. Many studies have been done in different parts of the world, the worldwide incidence is estimated as 3-7%. The estimated incidence in the United States & the United Kingdom is 2-5%, [2] Japan 1.96%, [3] while in developing countries incidence is highest among India 2.22%, (64.3%,) Bangladesh 1.75%. [4] Studies conducted in various locales of Pakistan showed that the prevalence of congenital malformations was 11.5/1000 births. [5] But no such studies have been

conducted in interior regions of Sindh Pakistan. In Pakistan, the incidence rate varies like in Abbottabad incidence is highest 31%, [6] Lahore 21.5%, [7] Rahim Yar khan 15%, Peshawar 2.9%, [8] Hyderabad 15.7%, Faisalabad 2.9%, Multan 2.95%, Karachi (LNH 15.8/1000, Civil hospital 4.1%). [9]

Prenatally various tools are available to detect the risk of different congenital anomalies like family history, family pedigree, vaccination status for rubella and chickenpox, and screening tools for genetic conditions and birth defects. DNA analysis for carrier risk identification can be performed. Amniocentesis, cordocentesis, Chorionic villus sampling, ultrasonography, maternal serum markers, TORCH profile are the other modalities. Clinical examination for deafness and heart anomalies are all part of neonatal screening. On basis of these modalities, various manipulations and interventions can be done for certain congenital malformations like hydrocephalus, Posterior Urethral Valve, and hydronephrosis. Neonatal surgical intervention is performed shortly after birth to restore not only the anatomy but also the functionality of the baby. Because of increased reporting of congenital anomalies, this study would be helpful in early identification, intervention, and reducing morbidity & mortality. The current study was designed to determine the frequency of common congenital anomalies among neonates in tertiary care hospitals.

### **Methodology:**

This study was carried out at Children's Hospital Larkana from April to October 2018 by the Non-Probability consecutive sampling technique. The ethical review committee permitted the study. The sample size was 124 was calculated by Rao soft Sample size calculator, according to the prevalence of congenital anomalies (8.84%) in Pakistan [5] with a confidence interval of

95% and margin of error less than 5%. All Newborns aged 0 hours -28 days of either gender either preterm or term/postterm with congenital anomalies were included in the study. Mothers of newborns having a systemic illness (Diabetes mellitus, Hypertension, Tuberculosis, and Epilepsy) that are confirmed via detailed history & relevant investigations were excluded. All the newborns were assessed by history, general physical examination, and relevant investigations like Complete Blood Count and ultrasound abdomen were performed. Echocardiography and Brain CT Scan was performed when needed. The questionnaire was filled out on hospital admission by the researcher herself.

All data were entered and analyzed in SPSS version 23. Maternal age, birth weight, parity, gestational age, was expressed in mean & standard deviation (Mean  $\pm$  SD). Percentage & frequency was calculated for Categorical variables like the presence of demographic age, gender, mode of delivery, consanguinity, booking status, education status of parents, family monthly income, rural/urban, family history of congenital anomalies & congenital anomalies (Neural tube defect, sacrococcygeal teratoma, cyanotic, acyanotic heart disease, Anorectal malformation, esophageal atresia, duodenal atresia, posterior urethral valve, hypospadias, hydrocele, undescended testes, limb deformity, developmental dysplasia of hip, polydactyly)

## **Results:**

This study included one hundred twenty-four Newborns aged 0 hours -28 days. The mean maternal age of the patients was  $24.16 \pm 4.13$  years. Similarly mean birth weight, gestational age, parity is also shown in Table 1. There were 54 (43.55%) rural and 70 (56.45%) urban patients. The education status of the parents and income status of the family is also shown in Table 1. Regarding mode of delivery, 54(43.55%) were vaginal delivery and 70(56.45%) cesarean

section. In this study, consanguinity was observed in 40 (32.26%) cases. The gender status of the neonate was female 33(26.61%) and male 91(73.39%). The frequency of common congenital anomalies among neonates is presented in **Table 3**.

The rate of common congenital anomalies among maternal age groups was not significant. The rate of GIT anomalies was significantly high with multiparty women as compared to primiparity women. While other anomalies differences were insignificant between multiparous and primiparous women. There was also no significant difference in the rate of congenital anomalies between the gestational age group and the birth weight of the babies. The rate of genitourinary system congenital anomalies was also significantly high in the male neonates as compared to female neonates ( $p=0.013$ ) while the rate of musculoskeletal was significantly high in females as compared to male neonates ( $p=0.006$ ).

**Table 1: Descriptive statistics of the study participants**

Variables	Mean $\pm$ SD
Maternal age (Years)	24.16 $\pm$ 4.13
Birth weight (Kg)	2.62 $\pm$ 0.29
Gestational age (Weeks)	37.18 $\pm$ 1.73
Parity	2.13 $\pm$ 0.77

**Table 2: Demographic characteristics of the study participants**

Variables	Number	Percentage
<b>Residence</b>		
Urban	70	56.45

Rural	54	43.55
<b>Booking Status</b>		
Booked	72	58.03
Unbooked	52	41.94
<b>Educational Status</b>		
Illiterate	59	48
Graduate	9	7
<b>Monthly Income</b>		
<25000	82	66.13
>25000	42	33.87
<b>Mode of delivery</b>		
Cesarean section	70	56.45
Vaginal delivery	54	43.55
<b>Consanguinity</b>		
Yes	84	67.74
No	40	32.26
<b>Family history of congenital anomalies</b>		
Yes	27	21.77
No	97	78.23
<b>Gender</b>		
Male	91	73.39
Female	33	26.61

**Table 3: Congenital malformations in study participants**

<b>CNS Anomalies</b>	<b>21%</b>
Hydrocephalus	6.5%
Sacroccygeal Teratoma	2.4%
Meningomyelocele	11.3%
Encephalocele	0.8%
<b>CVS Anomalies</b>	<b>11.3%</b>
Cyanotic Heart Disease	2.4%
Acyanotic Heart Disease	8.9%
<b>GIT Abnormalities</b>	<b>39.5%</b>
Duodenal Atresia	0.8%
Anorectal Malformation	2.5%
Pyloric Stenosis	0.8%
Exampholos	5.6%
Diaphragmatic Hernia	2.4%
Hirschsprung Disease	4.8%
<b>Genitourinary Anomalies</b>	<b>16.9%</b>
Hypospadias	14.5%
Undescended Testis	0.8%
Cloacal Exstrophy	0.8%
Hydrometrocolpos	0.8%
<b>Musculoskeletal Anomalies</b>	<b>11.3%</b>
Club foot	0.8%
DDH	0.8%
Polydactyly	1.6%
Cleft lip/palate	8.1%

## Discussion

This study observed that congenital anomalies of the central nervous system (CNS) were the most common anomalies. CNS anomalies were observed in 21%. Neural tube defects like

hydrocephalus 6.5%, sacrococcygeal teratoma 2.4%, meningomyelocele 11.3%, and Encephalocele 0.8%. According to a study from Pakistan congenital anomalies were assessed in newborns of hypothyroid mothers. About 147/662 of the newborn had some form of congenital anomalies, among them cardiovascular defects were the most common. In our study central nervous system was most commonly involved, this difference may be due to hypothyroid mothers in their study. [10]

In our study, CVS congenital anomalies were found in 11.4% of cases, GIT Anomalies (39.5%) Genitourinary system abnormalities (16.9%) Musculoskeletal abnormalities (11.3%). A study from Bangladesh observed the incidence of a congenital anomaly as 1.54%. Clubfoot abnormalities were the most common birth defects, while cardiovascular abnormalities were the most common organ system involved (28%). Cardiovascular involvement was twice as compared to our study. This may be due to different socio-economic and cultural differences between us. [11] In another international study, there were interesting results, although the most commonly involved system was the nervous system after that gastrointestinal and musculoskeletal systems were most commonly involved. [12]

There were different results in a study from Nigeria, showing the musculoskeletal system as a predominant system involved as having congenital anomalies. This difference may be due to maternal diabetes and hypertension in their study while all of our study participants were healthy. [13]

In our study, males were most commonly involved. In a similar African study, 52% of newborns with congenital anomalies were male. The incidence of congenital malformations was just



0.62%. Although most of the mothers in their study were diabetic the type of anomalies were similar to us ie nervous system anomalies. [14]

The age of the mother has a significant impact on the delivery of a fetus with congenital abnormalities. As a result, ladies over the age of 30 should be inspected more closely, as the probability of giving birth to a fetus with congenital abnormalities is higher. Multiparty and multigravidas, in addition to maternal age, are linked to an increased occurrence of CAs. The growing age of mothers has been linked to an increase in chromosomal meiotic mistakes and is thought to be the only non-genetic risk factor for trisomy in humans. The average maternal age of the patients in our study was  $24.16 \pm 4.13$ . In our study, the rate of GIT anomalies was significantly high with multiparty women as compared to primiparity women. In a similar international study from Ethiopia, it was revealed that mothers' mean age was 25.7 years. The prevalence rate of congenital malformation was 1%. Similar to our results Anencephaly, and hydrocephalus were the most common congenital anomalies. [15]

### **Limitations:**

One of the study's shortcomings is the well-established link between folic acid deficiency and neural tube abnormalities. Because of their high cost, serum and blood folate levels could not be assessed. As a result, due to a lack of relevant tests, a conclusive diagnosis of chromosomal abnormalities could not be made. The findings of this study may not be applicable to the full population because it was a cross-sectional descriptive study. Nonetheless, these findings highlight a significant public health issue and serve as a foundation for further research.

### **Conclusion**

Congenital abnormalities are prevalent in our setup, and the CNS is the most commonly involved system, followed by the gastrointestinal system, genitourinary system, cardiovascular system, and musculoskeletal system. Knowledge of incidence and pattern of congenital anomalies are important to plan preventive strategies at different levels by healthcare providers.

### **Ethical Approval:**

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

### **Consent**

As per international standard, parental written consent has been collected and preserved by the author(s).

### **COMPETING INTERESTS DISCLAIMER:**

Authors have declared that no competing interests exist. The products used for this research are commonly and predominantly use products in our area of research and country. There is absolutely no conflict of interest between the authors and producers of the products because we do not intend to use these products as an avenue for any litigation but for the advancement of knowledge. Also, the research was not funded by the producing company rather it was funded by personal efforts of the authors.

### **References**

1. Aliyu LD. Fetal Congenital Anomalies in Africa: Diagnostic and Management Challenges. Congenital Anomalies in Newborn Infants-Clinical and Etiopathological Perspectives 2021 Sep 15. IntechOpen.
2. Forci KH, Alami MH, Bouaiti E, Slaoui ME, Alaoui AM, Izgua AT. Prevalence of congenital malformations at the “Les Orangers” maternity and reproductive health Hospital of Rabat: a descriptive study of 470 anomalies. BMC pediatrics. 2020; 20(1):1-0.

3. Ikehara S, Kimura T, Kakigano A, Sato T, Iso H, Japan Environment Children's Study Group, Saito H, Kishi R, Yaegashi N, Hashimoto K, Mori C. Association between maternal alcohol consumption during pregnancy and risk of preterm delivery: the Japan Environment and Children's Study. *BJOG: An International Journal of Obstetrics & Gynaecology*. 2019; 126(12):1448-54.
4. Siddika M, Sen S, Islam MN, Bhuiyan MK. Pattern and Risk Factors of Congenital Anomaly in Newborn in a Tertiary Level Private Medical College Hospital, Bangladesh. *Mymensingh Medical Journal: MMJ*. 2018; 27(4):805-12.
5. Hussain S, Asghar I, Sabir MU, Chattha MN, Tarar SH, Mushtaq R. Prevalence and pattern of congenital malformations among neonates in the neonatal unit of a teaching hospital. *J Pak Med Assoc*. 2014; 64(6):629-34.
6. Alanazi AF, Naser AY, Pagan P, Alanazi AF, Alanazi AA, Alsairafi ZK, Alsaleh FM. Trends of Hospital Admissions Due to Congenital Anomalies in England and Wales between 1999 and 2019: An Ecological Study. *International Journal of Environmental Research and Public Health*. 2021; 18(22):11808.
7. Sharif F, Mahmood F, Azhar MJ, Asif A, Zahid M, Muhammad N, Rehman IU, Neil SM. Incidence and management of cleft lip and palate in Pakistan. *J Pak Med Associat*. 2019; 69(5):632-39.
8. Kiran Z, Sheikh A, Humayun KN, Islam N. Neonatal outcomes and congenital anomalies in pregnancies affected by hypothyroidism. *Annals of medicine*. 2021; 53(1):1560-8.
9. Aziz A, Saleem S, Nolen TL, Pradhan NA, McClure EM, Jessani S, Garces AL, Hibberd PL, Moore JL, Goudar SS, Dhaded SM. Why are the Pakistani maternal, fetal, and

newborn outcomes so poor compared to other low and middle-income countries?

Reproductive Health. 2020; 17(3):1-2.

10. Kiran Z, Sheikh A, Humayun KN, Islam N. Neonatal outcomes and congenital anomalies in pregnancies affected by hypothyroidism. *Annals of medicine*. 2021; 53(1):1560-8.
11. Amin T, Pervez M, Nur AN. Frequency of Birth Defects in a Tertiary Care Hospital in Bangladesh. *Bangladesh Journal of Child Health*. 2020 Oct 12; 44(1):8-12.
12. Silesh M, Lemma T, Fenta B, Biyazin T. Prevalence and Trends of Congenital Anomalies among Neonates at Jimma Medical Center, Jimma, Ethiopia: A Three-Year Retrospective Study. *Pediatric Health, Medicine and Therapeutics*. 2021; 12:61.
13. Chukwubuike KE, Ozor I, Enyi N. Prevalence and pattern of birth defects in the two tertiary hospitals in Enugu, southeast Nigeria: A hospital-based observational study. *African Journal of Paediatric Surgery: AJPS*. 2020 Jul; 17(3-4):85.
14. Mekonnen D, Worku W. Congenital anomalies among newborn babies in Felege-Hiwot Comprehensive Specialized Referral Hospital, Bahir Dar, Ethiopia. *Scientific Reports*. 2021 May 26; 11(1):1-8.
15. Gedamu S, Sendo EG, Daba W. Congenital Anomalies and Associated Factors among Newborns in Bishoftu General Hospital, Oromia, Ethiopia: A Retrospective Study. *Journal of environmental and public health*. 2021 Mar 31; 2021.