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 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 history, general physical examination, and relevant investigations; Questionnaire was filled out immediately within 24 hours after hospital admission

Results: The mean maternal age of the patients was 24.16±4.13 years. 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%. CVS anomalies were found in 11.4% 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). 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

Comment [WU1]: A specific information is needed; which type of investigations?

(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 damaged system. In neonates with congenital abnormalities, prematurity, low birth were linked risk factors. The prevalence and pattern of congenital defects must be known in order for healthcare providers to create preventive interventions at various levels.

Comment [WU2]: All these results have not been presented in the manuscript!
The author/s indicated that the frequency of anomalies was included in table2, but there were no actual detailed data regarding the anomalies were found in table2!

Comment [WU3]: This conclusion is not supported by the study results.

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 later in life," [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].

Various tools are available to detect different congenital anomalies prenatally by asking family history and screening tools for genetic conditions and birth defects. Family pedigree and DNA analysis for carrier risk identification, vaccination status for rubella and chickenpox, by Amniocentesis, cordocentesis, Chorionic villus sampling, ultrasonography, maternal serum markers, TORCH profile. Clinical examination and screening for blood, metabolism, and hormone production abnormalities, 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/posterm 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, 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 ± 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) To evaluate the effect modification, mode of delivery, consanguinity, booking status & family history of congenital anomalies were stratified followed by application of chi-square with a p-value.

Comment [WU4]: How did these anomalies have been diagnosed?

Comment [WU5]: Repeated data!

Results:

Comment [WU6]: Very poor presentation of the results. The most important data that reflect the study title were not available!

This study included one hundred twenty-four Newborns aged 0 hours -28 days. The mean maternal age of the patients was 24.16±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 2.

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).

Comment [WU7]: No any data regarding the congenital anomalies has been mentioned in table2!!

Comment [WU8]: Where are these comparisons?!

Table 1: Descriptive statistics of the study participants

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

Comment [WU9]: There is a confusion, does the study on fetuses or on neonates?

Table 2: Demographic characteristics of the study participants

Variables	**Number**	**Percentage**
Residence		
Urban	70	56.45
Rural	54	43.55

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

Discussion

Comment [WU10]: This discussion has been based on unavailable results!

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±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

Comment [WU11]: This conclusion is not supported by the study results!

Congenital anomalies are not rare in our setup and CNS was the most commonly affected system in our study. Prematurity, LBW, male gender, consanguinity, advanced maternal age, and family history of congenital anomalies were associated risk factors in neonates for congenital

anomalies. Knowledge of incidence and pattern of congenital anomalies are important to plan preventive strategies at different levels by healthcare providers.

Permission:

It was taken from the ethical review committee of the institute

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.

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