Patterns, Associated Risk Factors, And Management of Congenital Anomolies

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ABSTARCT

Background:

Congenital anomalies (CA) are one of the leading causes of morbidity and mortality in paediatric patients. The study aims to evaluate the patterns, associated risk factors, and management of congenital abnormalities to prevent such conditions in the paediatric population.

Methodology:

An observational study was conducted in a tertiary care hospital, Gandhi hospital, Secunderabad, India. 80 cases were identified, included and analysed for the study from inpatients units of Paediatrics department. Data were collected from medical records and interviews with parents. The collected data was analysed using statistical methods.

Results and Discussion:

Out of sixty cases, the incidence of CA was higher in male babies. In our study, twenty-one children were between the age group of 3 months to 2 years. The occurrence of CA was more frequent among individuals with birth weight of 2 - 2.5 kg. It was observed that 26.7% of the children with CA were born out of consanguineous marriages and most of the congenital malformation cases had no significant risk factors in the study. The central nervous system was the commonest system associated with CA, followed by the genitourinary system. The commonly prescribed medications were analgesics, multivitamins, beta-lactam antibiotics, and cephalosporins. Oral and intravenous routes of administration were most commonly preferred in children with CA.

Conclusion:

This study emphasizes the need for early detection, prevention, and appropriate management of congenital anomalies in paediatric patients. The study highlights the importance of monitoring the birth weight, sex, and family history in predicting the risk of CA. The findings of this study can help healthcare providers improve their understanding of congenital anomalies and provide better care to affected children.

Keywords:-Congenital anomalies, consanguineous marriage, malformations, paediatrics, risk factors.

INTRODUCTION

Congenital anomalies are defects in early embryonic development or innate abnormalities appear in the course of development present at birth.[1] Congenital anomalies are referred as congenital abnormalities or malformations. These are abnormalities that are developmental defects.[2] Congenital anomalies appear due to flawed embryogenesis and also intrinsic anomalies during the developmental process.[3] Congenital malformations are the major contributors to infant mortality in both developed and developing nations.[4] They can be discrete abnormalities or components of the syndrome.[5] Congenital anomalies birth prevalence is impacted by social, ethnic, economic, and ecological factors, according to studies conducted worldwide.[6] They are remarkably highly frequent in consanguineous couples.[7] Males develop birth defects more than females, white people than non-white people, and the babies of mothers who are at the start or end of their fertile lives are more prevalent to develop malformations.[8] To standardise records and improve knowledge of the prevalence of common and unusual forms of congenital malformations, population-based registries for epidemiological surveillance of congenital anomalies are crucial.[9] In actuality, several links amongst risk factors and categories of birth abnormalities have been found by epidemiologic research analyses.[10] A study conducted

on a bigger population would likely assist us in determining the frequency and pattern of limb malformations and assist us in guiding the parents.[11] The embryonic membrane of the human embryo provides excellent protection while it is inside the uterus, but teratogens may damage development if they are exposed to the mother during the first few weeks of pregnancy.[12]

It is a rare disease and Uncertain but well-known hereditary problems, environmental toxins, infectious diseases, medications, and untreated medical conditions including diabetes and epilepsy in the prenatal period will all be causal factors in 60% of cases multifactorial inheritance was the major cause for anomalies.[13,14] Pregnancy-associated conditions are vital or heavily influence the occurrence of CA. The occurrence of congenital defects is higher for pregnancy with hypertension, insulin-dependent diabetes with pregnancy, antepartum pregnancy, and twin pregnancy.[15] In order to provide appropriate guidance for examining other acquaintance, perform prenatal diagnosis and monitoring, initial diagnosis of patient with numerous abnormalities are essential.[16] Chest x-rays remain the standard methodology for locating the endotracheal tube, despite ultrasonography which has been proven to be an alternative to traditional radiography to identify birth defects.[17]

Ultrasound antenatal diagnosis is becoming more common, especially in situations with aplasia. The parents will be made aware of the deficit before the baby is born.[18] Major CA in children may require numerous complicated surgical operations and technologically sophisticated monitoring equipment's, that may be frequently unavailable in underdeveloped nations.[19] Many surgical procedures, protracted new-born hospitalisation, and frequently unknowable future life expectancy are just a few of the issues that CA-affected children must deal with. Delayed diagnosis of a symptomatic pattern of abnormality or the final picture of concomitant defects may significantly increase parental anxiety, especially in the instance of multiple congenital anomalies.[20] Some parents believed that the clinicians' information was biased by their own ideas rather than being neutral. This was deemed unprofessional by parents.[21] In this study we assessed the patterns, risk factors and management of congenital anomalies.

METHODS

An observational study was conducted in department of paediatrics, Gandhi hospital, Secunderabad, Telangana. This study was approved by Institutional Ethics Committee. The children with congenital anomalies were included in this observational study. It was conducted for a period of 6 months from October 2022 to March 2023. The sample size taken was 80 out of which 60 were included in our study as they met our inclusion criteria and 20 were excluded as they did not meet our criteria. SPSS version 29 was the software used in this study for calculation of mean, standard deviation, cross tabulations taking age, gender and birthweight into consideration. In the study the inclusion criteria of patients were children below the age of 12 years, Inpatients of paediatric department, PICU, NICU, follow up of cases until discharge and cases with complete information. Patients above 12 years age, unconscious patients, outpatient's department patients and cases with incomplete information or without a proper discharge summary were excluded from the study. Dropouts in this study were absconded patients, if the patient dies in the process or if patient left the hospital in between treatment course. All the children below 12 years of age admitted in the paediatric department were included. The diagnosis of the congenital anomalies was based on the antenatal investigations (ultrasound or magnetic resonance imaging), postnatal clinical examination and relevant investigations as per findings. Risk factors like complications like thyroid, hypertension, diabetes etc during pregnancy, history of similar complaints in the family, history of abortions, siblings with genetic disorders, grandparents with complications were taken into consideration. The demographic details of the children like birth weight, gender, consanguinity, age, immunization history, management etc were collected. The data was entered into excel sheet and analysed.

RESULTS

We have collected 80 cases in total, and we considered only 60 cases according to inclusion criteria(n=60).

Table : 1 Patterns of Congenital Anomalies

Disease	No. of Cases	Percentage
Acyanotic congenital heart disease	1	1.7
Addison's disease with autoimmune encephalitis	1	1.7

Bronchogenic cyst	1	1.7
Cerebral palsy	2	3.3
Closed lumbosacral myelomeningocele	1	1.7
Congenital airway malfunction	1	1.7
Congenital heart disease	1	1.7
Congenital hypothyroidism	2	3.3
Cyanotic congenital heart disease	2	3.3
Cystic fibrosis	1	1.7
Dilated cardiomyopathy	2	2.4
Downs syndrome	1	1.7
Encephalocele	1	1.7
Epidermolysis bullosa simplex	1	1.7
Focal cortical dysplasia	1	1.7
Global development delay	1	1.7
Hereditary motor sensory neuropathy	1	1.7
Hydrocephalus	1	1.7
Hydrocephalus	3	5.0
Job's syndrome	1	1.7
Juvenile myasthenia gravis	1	1.7
Left undescended testis	1	1.7
Liver hemangioma	1	1.7
Lymphangioma of upper lip	1	1.7
Microcephaly	3	5.0
Mild penile hypispadias with chordee	1	1.7
Myelomeningocele	1	1.7
Myoclonic jerk syndrome with autoimmune encephalitis	1	1.7
Neonatal cholestasis	1	1.7
Neuroblastoma	1	1.7
Niemann Pick Disease	1	1.7
Omphalomesenteric duct cyst	1	1.7
Osteogenesis imperfecta	1	1.7
Perimembranous ventricular septal defect	1	1.7
Phimosis	4	6.7
Postnatal hydroureteronephrosis	1	1.7
Quadriplegia cerebral palsy	1	1.7
Right inguinal hernia with undescended testis	1	1.7
Schimke immuno osseous dysplasia	1	1.7
Sickle cell anaemia	1	1.7
Spastic cerebral palsy	1	1.7
Spastic diplegic cerebral palsy	1	1.7
Spastic quadriplegia cerebral palsy	1	1.7
Spinal dysraphism	1	1.7
Thalassemia	1	1.7
Tuberous sclerosis	1	1.7
Undescended testis	2	2.4
X-Linked Hypoglobulinemia	1	1.7
Total	**60**	**100.0**

In the total number of cases collected, 17 are female cases (28.3%) and 43 are male cases (71.3%). This shows that the number of patients diagnosed with congenital anomalies is more in males when compared to females.

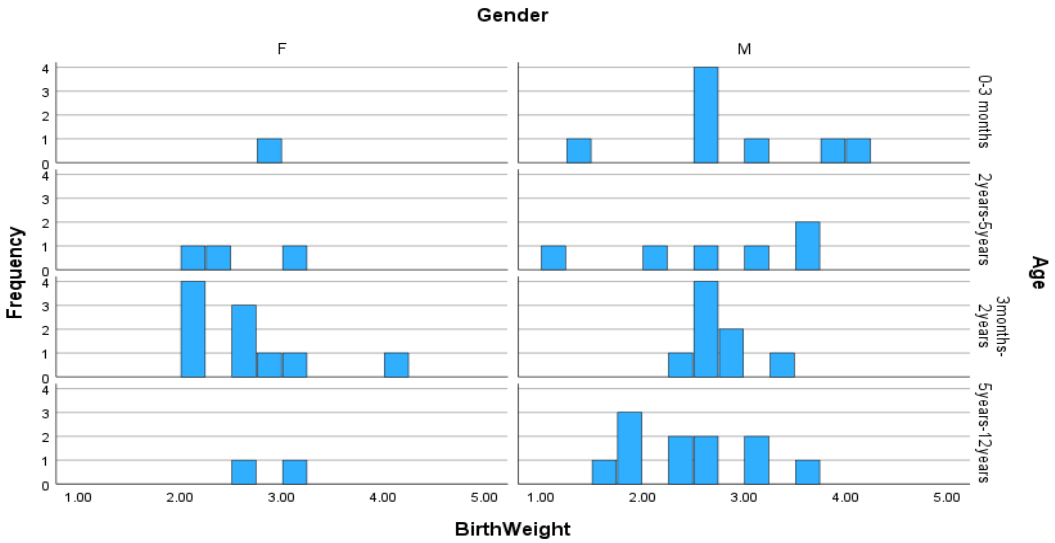

Fig:1 Graphical representation of the distribution of cases according to birth weight, age, and gender.

Patients with ages 3 months to 2 years were highly diagnosed with congenital anomalies, and patients with 2 years to 5 years were least diagnosed when compared to patients of other ages. 44 cases (73.3%) were immunized, 2 cases (3.3%) were partially immunized, 4 cases (6.7%) were not immunized, and we were unable to find the status of immunization in 10 cases (16.7%). In the total 60 cases, 16 cases (26.7%) were presented in which the child was born out of a consanguineous marriage, and 35 cases (58.3%) were presented in which the child was not born out of a consanguineous marriage. We were unable to find information about consanguinity in 9 cases (15%). The mean birth weight is 2.6 with a standard deviation of 0.62. Out of 60 cases, the minimum birth weight is 1.20 kg and the maximum birth weight is 4.2 kg. In our study, we were unable to find the risk factors associated with congenital anomalies in 27 (45.8%) cases. Out of all the risk factors mentioned in the graph, the highest number of cases (15.3%) are with a history of similar complaints in the family, followed by 8 cases (13.6%) of consanguineous marriages. In our research study, we found the highest number of cases (31.7%) with congenital anomalies that belong to the central nervous system, which is followed by the genitourinary system (18.3%). Females from ages 3months to 2 years were most commonly presented with central nervous system abnormalities. Males from ages 5 to 12 years were most commonly presented with central nervous system abnormalities. In our study, we have also considered the management patterns in patients with congenital anomalies. We consider only the common drugs used in the hospital while excluding the disease-specific treatment medication.

Table :2 Summary of drugs used in the management

Drugs	Frequency	Percentage
Tetracycline antibiotics	1	0.6
Glycopeptide antibiotics	4	2.6
Aminoglycoside antibiotics	5	3.3
Antihistamines	8	5.3
Nitroimidazole antimicrobials	9	6.0
Mucolytics	11	7.3
Oral rehydrating solution	11	7.3
Cephalosporin	16	10.7

Proton pump inhibitors	17	11.4
Beta-lactamase inhibitors	18	12.0
Multivitamins	21	14.0
Analgesic	28	18.7
Total	**149**	**100.0**

This graph shows that analgesics were highly prescribed in the greatest number of cases (28 cases) followed by multivitamins (21 cases). In terms of antimicrobial agents, beta-lactam antibiotics (18 cases) were the most used, followed by cephalosporins (16 cases). The highest number of patients were prescribed with drugs that should be taken via the oral route, which is followed by intravenous injection administration.

DISCUSSION:

A prospective observational study was conducted to assess and evaluate the patterns and associated risk factors with congenital anomalies in the paediatric patients. The focus of this study was to evaluate the patterns of congenital anomalies and help prevent such conditions in the paediatric patients.

According to this study, genitourinary malformations like undescended testis (phimosis) and central nervous system malformations (hydrocephalus, microcephaly etc) were common among the paediatric patients. In this study, the paediatric patients of various groups were observed that is 0-3 months (20%),13 years (1.7%), 2-5 years (16.5%), 3 months – 2 years (35%),5 – 12 years (26.7%) out of this the percentage of paediatric patients of 3 months – 2 years (35%) was found to be more. In a total of 60 cases collected 21 children with congenital anomalies were between the age group of 3 months to 2 years which was more compared to the other age groups in the study and among these children hydrocephalus and phimosis were mostly observed than other conditions. In our study the total percentage of male patients (71.1%) was comparatively higher than the female patients (28.3%) which is similar to the study reported by J M Lary *et al* 2001.[22] In a study conducted by Vikram Datta *et al* and Verma *et al*, [23] no difference was observed in the distribution of malformations between both the genders which is contradicting to our study. The Male: Female ratio was 43:17 and it was statistically significant. 35 paediatric patients were born out of a non-consanguineous marriage, 16 children were born out of consanguineous marriage and 9 of them were not applicable. The percentage of paediatric patients born out of non-consanguineous marriage was found to be more when compared to children born out of consanguineous marriage and NA. In a study done by Liascovich R, Rittler M,[24] a significant association was observed between parental consanguinity and congenital malformations. Mili *et al* 1991,[25] observed birth defects are linked to extra morbidity in low-birth weight new-borns similar to the results observed in present study where the incidence of congenital anomalies was observed more in children with birth weight between 2 – 2.5 kgs (31.7%) according to the observed data.

Wodi A P *et al* 2022,[26] explained that vaccines and immunization of children can help prevent serious illness and reduce the severity of congenital anomalies in children. In our study the data collected shows most of the children were immunized. The major risk factor observed in this study was patients with H/O similar complaints in the family, patients born out of a consanguineous marriage, children whose mothers had complications during pregnancy. Ali Raza Brohi *et al* 2008,[27] explained that for majority of malformations, prenatal period and multifactorial inheritance were to blame. Viviana Hodosan *et al* 2022 explained that many classes of antibiotics used during pregnancy by women belong to category B most prescribed being cephalosporins which is similar to our results. The drugs were mostly given through oral route (58%) followed by intravenous (32.2%) and least given by intranasal route (9.8%). Farmer D *et al* 2015,[28] in his studies concluded that improving the quality of paediatric surgical care and accessibility in low income and lower middle-class countries has potential to reduce mortality and lifelong disability.[29] As observed in the data collected, surgical procedures performed in the children with congenital anomalies were circumcision, laparoscopy, homograft debridement, umbilectomy, VP shunting, herniotomy, splenectomy, excision of lesion. The congenital anomalies of central nervous system (16%) were found to be highest among all the others followed by genitourinary(9.2%),

immune system(7.6%), cardiovascular(5.9%), musculoskeletal(2.5%), respiratory(2.5%), endocrine(1.7%), gastro(1.7%), multisystem(1.7%), lymphatic system (0.8%,) and peripheral nervous system (0.8%).

CONCLUSION

The incidence of congenital anomalies was significantly higher in male babies. The incidence of congenital anomalies was higher in patients with ages of 3 months to 2 years. The incidence of congenital anomalies was observed to be higher in individuals with birth weight of 2-2.5 kg. Most of the congenital malformation cases had no significant risk factors in our study. 26.7% of the patients with congenital anomalies were born out of consanguineous marriage. The most common system involved was the central nervous system followed by genitourinary system. The most commonly used medications were analgesics, multivitamins, beta lactam antibiotics, and cephalosporins. Oral and intravenous routes of administration were commonly preferred in cases with congenital anomalies.

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