Study of Congenital Malformation in a Tertiary Care Teaching Hospital

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ABSTRACT

Background: Congenital anomaly is one of the most important causes & being the 5th most common cause of neonatal mortality & morbidity. It may present as a structural or functional abnormality. These defects occur due to defective embryogenesis. Associated factors may be maternal age, maternal TORCH infection, drugs, genetic factors. Antenatal USG reduces the incidence.

Materials and Methods: A cross-sectional study was done in the Pediatric department over 1 year. Diagnosis of all congenital anomalies was done by the concerned pediatrician& pediatric surgeon. Data was collected in the specified format.

Results: A total of 10205 cases of age group 1 month to 5 years presented to the paediatric OPD, out of which 193 children were diagnosed as congenital anomalies in 1 year. Males were found to be affected the most. The most common system involved was found to be the genitourinary system (36.78%). The second most common system involved was the gastrointestinal system (33.67%). The least common system involved was the musculoskeletal system.

Conclusion: Congenital anomalies are a major cause of neonatal & infantile mortality & morbidity. Routine screening with a level II targeted scan for all the pregnant mothers should be mandatory. Adequate nutrition, parental education & Rubella vaccination of the mother can decrease the prevalence of congenital anomalies to some extend.

Keywords: Congenital anomalies, Genitourinary system, Gastrointestinal system, Hypospadiasis (Siriraj Med J 2021; 73: 609-613)

INTRODUCTION

The period of organogenesis or early fetal age (5-8 weeks of gestation) is the most vital period for the normal development of the fetus. Better maternal care & improved life standards of living, impact the outcome of congenital birth anomalies.[1] Congenital anomalies are caused due to structural or functional abnormalities that occurred in intrauterine life. A congenital anomaly is the most important cause of neonatal morbidity & mortality in developing and developed countries. It accounts for 8-15 % of perinatal deaths & 13-16% of neonatal deaths.[2,3] About 94% of congenital anomalies are seen in low to middle-income countries.[4] Maternal nutrition, infectious diseases & social stress are the most important factors for congenital anomalies in developing country like India. Several factors affect the incidence, e.g. maternal age, consanguinity, nutrition, TORCH infection, genetic factors and certain medicinal & recreational drugs, including alcohol, tobacco & radiation.[4] Vaccinations, adequate folic acid intake, iron, iodine fortification & proper antenatal care are the few preventable measures.

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Congenital defects are the emerging causes of morbidity & mortality.[5] Most congenital anomalies have serious medical, surgical & cosmetic consequences contributing long term defect impacting family life.

MATERIALS AND METHODS

Our study is a cross-sectional study done over 1 year from January 2019 to December 2019 in the pediatric department at IMS & SUM Hospital, Bhubaneswar. Children presented to the paediatric outpatient department of age group 1 month to 5 years were taken as a subject. The study aimed to find out the incidence and different proportion of congenital anomaly presenting to our hospital using a structured form containing the age of presentation, sex, type of congenital anomalies & its association with various maternal risk factors, e.g. maternal anemia, parity, education of mother, antenatal check-up & antenatal iron & folic acid intake.

The diagnosis was made by the concerned pediatricians & pediatric surgeon. Informed consent was taken from the parents. Detailed general physical & systemic examination was done. Ultrasonogram, neurosonogram, X-Ray & 2D ECHO were done to rule out the internal anomalies. CT, MRI brain & karyotyping were done in selected cases.

Out of all congenital cases collected, all are divided into genitourinary, gastrointestinal, vascular, musculoskeletal, cardiovascular system (CVS), central nervous system (CNS). The variables were analysed by frequencies & chi-square test using SPSS Version 20.

RESULTS

A total of 10205 children presented to the pediatric OPD, out of which 193 children were diagnosed with congenital anomalies and referred to the peadiatric surgery department of IMS & SUM Hospital during the study period with different congenital anomalies. It has been seen that the most common age group of presentation is 1-3 years (38%) (Table 1) with a male predominance (93.3%). It is found that the genitourinary system is the most common type of system involved (36.78%) & musculoskeletal system anomalies being the least involved one (1%) (Table 2).

Inguinal hernia is the most common gastrointestinal anomaly in this study, consisting of 81.5%. Tongue-tie is the second most common anomaly, which is 9.2%. Neuroblastoma & umbilical granuloma are the least common types of gastrointestinal anomalies in this study. Hypospadiasis is the most common presentation (36.61%). Hydrocele is the 2nd most common congenital genitourinary anomaly in this study. Vaginal synachiae & Posterior urethral valve are the least common types. The most common cardiovascular presentation is patent ductus arteriosus (PDA), which accounts for 42.8%. The second most common CVS anomaly is a ventricular septal defect (28.6%) (Table 3).

Out Of 193 children, 6 children presented with CNS anomalies and all of them presented as a case of hydrocephalus. Only 2 children presented with musculoskeletal anomalies, one with dermoid cyst & another with supernumerary little finger 10 children presented with respiratory anomalies in the form of laryngomalacia. Only 4 children presented with vascular anomalies, e.g. Hemangioma. 5 children presented with a thyroglossal cyst, whereas 2 children with thyroglossal fistula.

Low maternal education significantly increases the risk of congenital anomalies (P-value- <0.00001). The association of maternal anemia & congenital anomalies is statistically significant in our study (P-value < 0.00001). Improper dosing intake of iron & folic acid statistically increases the risk of congenital anomalies P-value-0.003). There was no association found between parity & antental check-up with risk of congenital anomalies in our study. (P-value- 0.2 & 0.14 respectively) (Table 4)

DISCUSSION

Congenital anomalies are an important cause of childhood morbidity. The pattern of presentation & prevalence of congenital anomalies may vary according to geographical distribution.[1] Nutritional deficiency & maternal infection are the most common causes of congenital anomalies in developing countries like India. Approximately 1 in 33 infants & 3.2 million congenital defects are reported worldwide annually.[2] Every year around 2.7 lakhs neonates die during the neonatal period due to congenital anomalies worldwide. Praneshwari et al. 2019 did a study where they found congenital anomalies in male babies are more (61.5%).[6] Another study was done by Devi et al. 2018 which showed congenital anomalies are more in males (57.1%).[7] Vinodh et al. 2017 in their study done in 2017, found male predominance in congenital anomalies (54.4%).[8] Our study, it is found that congenital anomalies are more prevalent in males, which comprises 93.3%.

Our study's most common presenting age group in our study is between 1-3 years that is 38%. Other studies done by pabbati et al & Vinodh et al where they included newborns as their study population.[8,10] The most common system involved in a study done by Shatanik sarkar et al. in 2013 was the musculoskeletal system (33.2%).[9] Jayalakshmi pabbati et al in 2016 did a study where they found the most common system

TABLE 1. Age distribution of congenital anomalies.

Age Group	Percentage (%)
1 Month-1 Year	28
1-3 Year	38
3-5 Year	34

TABLE 2. System wise distribution of congenital anomalies.

System	Number (Percentage)
Cardiovascular System	28 (14.5%)
Gastrointestinal System	65 (33.67%)
Genitourinary System	71 (36.78%)
Masculoskeletal System	2 (1%)
Respiratory System	10 (5.1%)
Vascular System	4 (2%)
Central nervous system	6 (3.1%)
Miscellaneous	7 (3.8%)

TABLE 3. System wise distribution of congenital anomalies.

System	Number (Percentage)
Gastrointestinal System	
Inguinal Hernia	53 (81.5%)
Tongue tie	6 (9.2%)
Umbilical Hernia	3 (4.6%)
Neuroblastoma	2 (3%)
Umbilical granuloma	1 (1.5%)
Genitourinary System	
Hypospadiasis	26 (36.61%)
Undescended Testes	11 (15.49%)
Hydrocele	14 (19.7%)
Posterior urethral valve	3 (4.22%)
Phimosis	13 (18.3%)
Vaginal synache	1 (1.3%)
Cardiovascular system	
Dextrocardia	3 (10.7%)
PDA	11 (39.2%)
VSD	8 (28.6%)
ASD	5 (17.9%)
TAPVC	1 (3.6%)

TABLE 4. Association of various parameters with Congenital anomalies.

	Congenital Anomalies		
	Present	Absent	P-value
Maternal education			<0.00001*
Less than High school	109	4,123	
High school & above	84	5,889	
Maternal anemia			<0.00001*
Present	74	119	
Absent	2,107	7,905	
Parity			0.2
High Parity (>3)	91	102	
Low Parity (<3)	5,113	4,899	
Antenatal Check up			0.14
>4 Check up	60	3,621	
<4 Check up	133	6,391	
Iron- Folic Acid Intake			0.003*
Not/Partially Taken	149	6,738	
Taken	44	3,274	

*p <0.05 is considered significant

involved in congenital anomalies was the musculosketetal system (37.6%).[10] Another study was done by S Swain et al where they found CNS (39.5%) was the most common system involved.[11] Kokate P et al, in 2016 conducted a study & found that craniospinal involvement is the most common presentation.[12] Devi KR et al in 2018 did a study where they found the musculoskeletal system as the most common system involved in congenital anomalies (50.5%).[7] The most common system in our study is found to be a genitourinary system which comprises 36.78% of children presented with various congenital anomalies. In our study, hypospadiasis is the most common congenital genitourinary anomaly consist of 36.61%. A similar prevalence was found by Dr S. Lakshmi Vinodh et al & Rameswarapu et al in 2017 & 2013, respectively.[8] Inguinal hernia is the most common congenital gastrointestinal anomaly found in our study (81.5%). On the contrary omphalocele & diaphragmatic hernia are the most common presentations found in studies done by Devi KR et al in 2018 & Dr S. Lakshmi Vinodh et al in 2017 respectively.[7,8]

In this study the most common congenital CNS anomaly is hydrocephalus similar to the study done by Dr S. Lakshmi Vinodh et al. They found hydrocephalus was the most common CNS presentations.[8] Dr S. Lakshmi Vinodh et al in their study found VSD as the most common cardiovascular anomaly, whereas in our study PDA is the most common presentation which comprises of 42.8% of the cardiovascular cases.[8] In our study association of maternal anemia & antenatal intake of iron & calcium tablet with congenital anomalies is statistically significant (P-value <0.00001), whereas the association of high parity with congenital anomalies is statistically not significant. A similar result was found in a study done by Thaddanee R et al in 2016.[13] The Association of antenatal check-up with congenital anomalies is statistically not significant in our study.

On the contrary, the association was significant in the study done by Thaddanee R et al in 2016.[13] In our study it is found that the association of maternal education with congenital anomalies is statistically significant (P value- 0.00001). Similar result was found in a study done by Dingemann C et al in 2019.[14]

CONCLUSION

Congenital anomalies are one of the major causes of stillbirth & infant mortality. All pregnant mothers should be counseled regarding the level II targeted scan

at 18-22 weeks to rule out the congenital anomalies. Various management modalities should be discussed with the concerned neonatologist, pediatric surgeon & neurosurgeon if any anomaly is detected. If an anomaly is compatible with life & parents are willing to continue the pregnancy, then proper precautions with utmost care should be taken. If the anomaly is incompatible with life, parents should be advised for termination. Parental education, maternal vaccination for Rubella & maternal adequate nutrition with iron & folic acid supplements can reduce congenital anomalies. This study was done to see the prevalence of various congenital anomalies presented to a tertiary care hospital so that we should maintain a record & with emphasis on routine screening of every pregnant mother to decrease morbidity & mortality.

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