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AN OBSERVATIONAL STUDY ON THE FREQUENCY OF CONGENITAL ANOMALIES

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Abstract:

Objective: *The aim of this study was to observe the determination in the various systems responsible for the congenital anomalies.*

Material and Methods: *The design of this study was an observational study. In this study analysis of the congenital anomalies was carried out in neonate involving numerous systems and sex. This study used diagnosis to identify numerous defects such as barium studies, magnetic resonance imaging, computed topography, x-rays, echocardiography and ultrasound.*

Results: *Hospital admissions in the duration of this study were 4201 and 371 patients (8.83%) of congenital anomalies were diagnosed with male and female ratio respectively 220 male patients (59.30%), 142 female patients (38.27%) remaining 9 patients (2.43%) had an ambiguous genitalia 215 patients were (57.95%) of normal delivery and 156 patients were of lower segment caesarian (42.05%). Common involvement was observed about the Central Nervous System (CNS) 89 patients (23.99%), 74 Gastro Intestinal Tract patients (19.95%), 61 Cardiovascular System patients (16.44%), 35 Respiratory System patients (9.43%) and 18 Genito Urinary System patients (4.85%). Dysmorphic featured babies were 48 patients (12.94%) and 17 palate and cleft lip patients (4.58%) and the rest were treated as 29 miscellaneous patients (7.82%).*

Conclusion: *Congenital anomalies prevalence was observed dominant in males in comparison to females and common most system was observed as Central Nervous System in 89 patients and Gastro Intestinal Tract in the 74 followed by 61 patients of CVD.*

Keywords: *Ambiguous Genitalia, Genito Urinary, Gastro Intestinal Tract, Congenital Anomalies.*

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INTRODUCTION:

To define congenital anomalies, we can say that functional abnormality, structure or body metabolism present at birth or also be referred to physical and mental abnormality. Developing and developed countries face an important issue of mortality in the children because of congenital anomalies. Under developed countries lack in the diagnostics of these congenital anomalies and other contributing reason is the poor documentation of the disease and anomalies in the children and poor managed statistical figures of health [1]. The congenital anomalies vary in the different areas and regions; its prevalence is differing in many regions. The major reasons behind the difference may be the involvement of social, racial and ecological difference in the different parts of the world. We need to consider varying parameters for the various regions in order to predict the incidence of congenital anomalies. There is 5% congenital anomalies incidence; whereas, prenatal diagnosis can be made in the case of 2 – 3 % of the patients with the help of non-invasive or invasive test methods to in the first year of the children age. Congenital anomalies etiology cannot be recognized in 60 – 80 % of the children [2]. There is environmental involvement in the 10 – 20 % of the patients for the incidence of congenital involvement. Pre-natal deaths because of the congenital anomalies are reported as 12 – 32 %. Folic acid as multivitamin use by the women before conceiving may reduce or eliminate the chances of these abnormalities, neural tube defects, related birth defects including heart defects, limb defects, facial and oral clefts, pyloric stenosis etc. A comprehensive strategy is required for the reduction of congenital disease incidence as an integrated plan that screen the population, councils and educates the communities for the better prevention and treatment of such anomalies. Our research study is very important in this regard as most of the hospitals and institutes do not even register or document these anomalies [3]. Research also points out the preventable anomalies in the case of congenital abnormalities and also aims at the signification of the disease prevention. Frequency determination in the various systems responsible for the congenital anomalies for the identifying strategies and prevention is the objective of our research.

MATERIAL AND METHODS:

The duration of this study was from January, 2019 to the December, 2019. Diagnostic equipment was up to date which made it a perfect referring spot of the congenital anomalies' patients from every part of Pakistan including complex patients of deliveries in such patients. NICU register was used for the documentation of such congenital anomalies including every disorder in the child including every child. Radiological investigations such as MRI, X-Rays, CT-Scan, USG and Barium needed for further diagnosis were completed. Data entry and analysis was made through SPSS-18. Percentage and frequencies were calculated for delivery mode, sex and involved system.

RESULTS:

We included 371 congenital anomalies neonates which was a (8.83%) of the NICU admission out of 4201. Male and female ratio was as that 220 males (59.30%) and 142 females (38.27%) including 9 ambiguous patients (2.43%) with a dominance of males over females. Vaginal delivery patients were 215 (57.95%) and 156 patients were of lower segment caesarian section (42.05%). CNS was most common system 89 patients (23.99%), with hydrocephalus and meningomyelocele sharing 60 patients (69.77%) and 21 patients of CNS deformations (23.60%). Gastro intestinal tract 74 patients (19.95%) were taken as 2nd repeated anomaly. CVD system 61 patients (16.44%) was 3rd repeated anomaly followed by 35 patients (9.43%) of respiratory system. Genitourinary system involvement was observed in 18 patients (4.85%).

DISCUSSION:

All the children who managed to survive any of the congenital anomalies during infancy are at enhanced morbidity risk for the health issues including few of the other problems such as mental, physical and social limitations [4]. Developing countries also face the effects of an ante-partum death in the congenital anomalies patients [5]. In this research, we included 371 congenital anomalies neonates which was a total of (8.83%) of the NICU admission out of 4201 [6]. Male and female ratio was as that 220 males (59.30%) and 142 females (38.27%) including 9 ambiguous patients (2.43%) with a dominance of males over females. Same has been observed in many of the other research studies by various authors [7]. Central nervous system was most affected system having neural tube defects, which makes it in the category of most repeated anomalies. Defects of birth mainly develop because of the neural tube closure lack after the conception in the 3rd or 4th of gestation [8]. This incidence of CNS has also been reported by various other authors.

Table: Frequency of Different Systems Involved in Congenital Anomalies

Category	Systems involved	Number	Percentage
Central Nervous System	Meningo Myelocoele	60	67
	Hydrocephalus	21	24
	Encephalocoel	4	5
	Microcephaly	3	3
	Spina Bifida	1	1
Gastro Intestinal Tract	Imperforate Anus	40	54
	Small Gut Atresia	21	21
		3	4
	Hircshprung Disease	10	14
	Anorectal Stenosis		
Respiratory System	Tracheo Esophageal Fistula	30	85
	Tracheal Atresia	2	6
	Hypoplastic Lung	1	3
	Choanal Atresia	2	6
Cardio Vascular System	Tetralogy of Fallot	13	21
	Ventricular Septal Defect	26	43
	Atrial Septal Defect	9	15
	Coarcatation of Aorta	2	3
	Patent ductal arteriosus	3	5
	Transposition of great arteries	1	2
	Dilated Cardio Myopathies	2	3
	Complex Anomalies	5	8
Genito Urinary System	Ambiguous Genetalia	9	50
	Congenital Hydronephrosis	9	50
Miscellaneous	Congenital Diaphragmatic Hernia	8	28
	Omphalocele	8	28
	Down Syndrome	5	18
	Pierre Robin Syndrome	2	7
	Edward Syndrome	1	3
	Sacrococcygeal Teratoma	1	3
	Cystic Hygroma	1	3
	Right Malformed Ear	1	3
	Epigastric Hernia	2	7

Apparent anomalies can be observed easily at the time of birth, which also makes it a reason for the increased incidence of congenital anomalies [9]. Multi-vitamin supplements use by the mothers in the developed countries has decreased the neural tube anomalies as it contains folic acid. There was also an involvement of the other systems except CNS such as Gastro intestinal tract 74 patients (19.95%) were taken as 2nd repeated

anomaly [10]. CVD system 61 patients (16.44%) was 3rd repeated anomaly followed by 35 patients (9.43%) of respiratory system. Genitourinary system involvement was observed in 18 patients (4.85%). According to the research of Tuncbilek, involved systems are urinary system, musculoskeletal system and CVD systems respectively 14.4%, 11.70% and 8.28% repeated mostly after CNS involvement [11]. Whereas, as per the outcomes of Himmetoglu, there is a reverse or decreasing order involvement of these systems. A Pakistani research held at Abbottabad in Ayub Teaching Hospital observes CNS as (31%) and NTD's as (77%) [12]. Cardiac defects are observed as (16%) and urogenital anomalies as (6%), which are similar to the outcomes of our research. Our research also notices 4.58 % patients of palate and cleft lip. Palate and Cleft lip cause the incidence of deaths in the 600 – 800 live birth, occurrence with some other anomalies are observed as (60%) [13]. It is also observed that almost half of the anomalies can be prevented. Birth defects can be decreased through documentation of these patients and regular ante-natal checkup for the assurance of the food supplementation and multi-vitamins before the birth of the child in the pregnancy and child bearing age.

CONCLUSION:

Congenital anomalies prevalence was observed dominant in males in comparison to females and common most system was observed as Central Nervous System in 89 patients and Gastro Intestinal Tract in the 74 followed by 61 patients of CVD.

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