

**Research Article****Congenital Anomalies****<sup>1</sup>Kainaat, <sup>2</sup>Qamar Abbas  
and <sup>3</sup> Muhammad Ali**<sup>1</sup>King Edward Medical University Lahore, Pakistan  
<sup>2</sup>Quaid-E- Azam Medical College Bahawalpur, Pakistan  
<sup>3</sup>Jinnah Post Graduate Medical Centre Karachi, Pakistan**ABSTRACT**

**Objective:** Frequency determination in the various systems responsible for the congenital anomalies for the identifying strategies and prevention is the objective of our research.

**Study Design:** Descriptive observational study.

**Place and Duration of Study:** Research was carried out in Mayo Hospital Lahore in the time span of January, 2017 to December, 2017.

**Material and Methods:** The venue of research was Mayo Hospital Lahore and analysis of the congenital complexities was carried out in every neonate involving numerous systems and sex. Our research used diagnosis to identify numerous defects such as barium studies, magnetic resonance imaging, computed topography, x-rays, echocardiography and ultrasound.

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

**Conclusion:** Important reason of hospital admission are congenital anomalies specially in the admission of NICU. Congenital anomalies prevalence was observed dominant in males in comparison to females and common most system was observed as Central Nervous System in 89 cases and Gastro Intestinal Tract in the 74 followed by 61 cases of CVD.

**Keywords:** Congenital Anomalies, Hydronephrosis, Cleft lip, Tetralogy of Fallot and Meningocele.

**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 nations 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 five percent congenital anomalies incidence; whereas, prenatal diagnosis can be made in the case of 2 – 3 percent of the cases 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 percent of the children[2]. There is environmental involvement in the 10 – 20 percent of the cases for the incidence of congenital involvement. Pre-natal deaths because of the congenital anomalies are reported as 12 – 32 percent. 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**

We conducted this descriptive observational research study in Mayo Hospital Lahore, which is a tertiary healthcare facility in the locality and research was completed in the time period that extended from January, 2017 to the December, 2017 in the ICU. 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 cases of deliveries in such cases. 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 total of (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 cases (2.43%) with a dominance of males over females. Vaginal delivery cases were 215 (57.95%) and 156 cases were of lower segment caesarian section (42.05%). CNS was most common system 89 cases (23.99%), with hydrocephalus and meningomyelocele sharing 60 cases (69.77%) and 21 cases of CNS deformations (23.60%). Gastro intestinal tract 74 cases (19.95%) were taken as 2<sup>nd</sup> repeated anomaly. CVD system 61 cases (16.44%) was 3<sup>rd</sup> repeated anomaly followed by 35 cases (9.43%) of respiratory system. Genitourinary system involvement was observed in 18 cases (4.85%).

### **DISCUSSION**

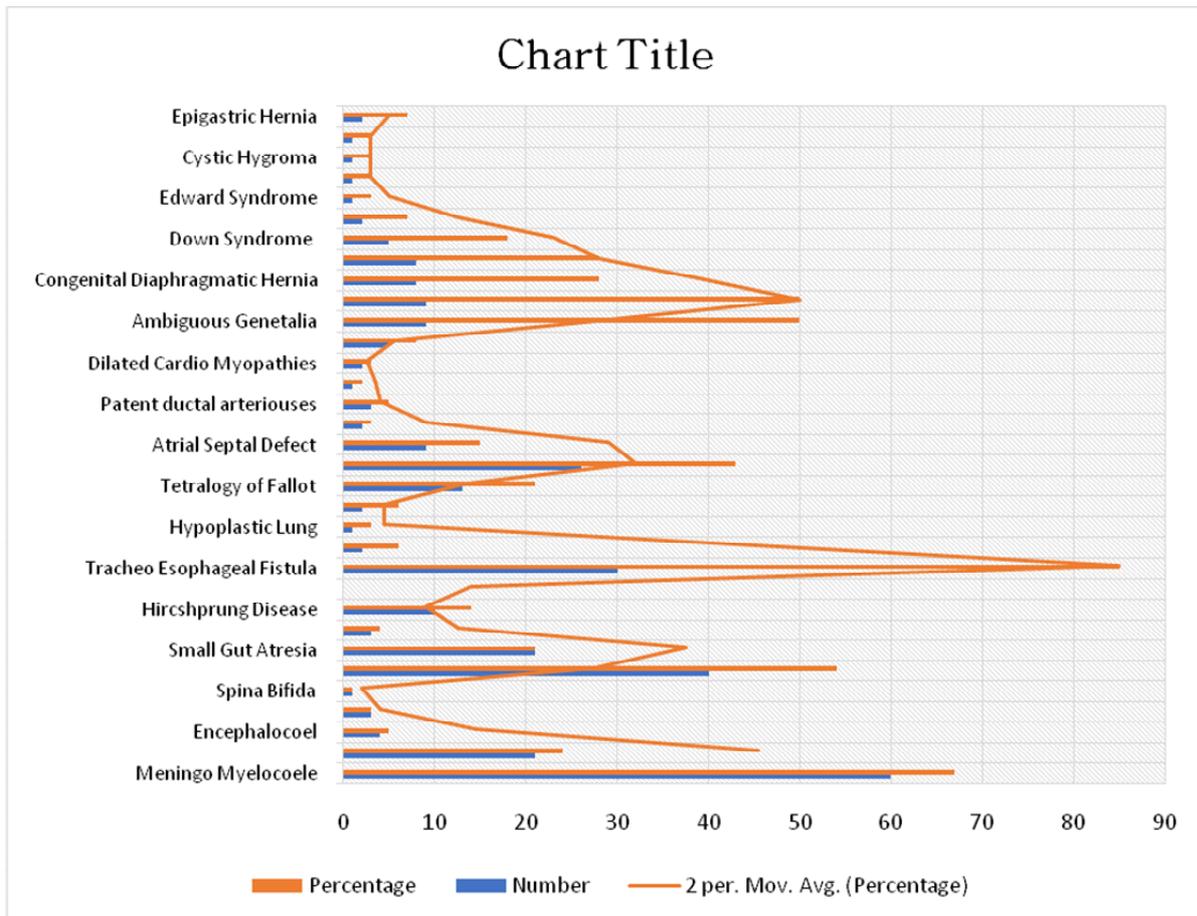
All the children who managed to survive any of the congenital anomalies during infancy are at enhanced morbidity risk for the health issues including few of the other problems such as mental, physical and social limitations[4]. Developing countries also face the effects of an ante-partum death in the congenital anomalies cases[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 cases (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 3<sup>rd</sup> or 4<sup>th</sup> 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 Myelocele	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
	Hirschprung 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
	Coarctation 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 Genitalia	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 Hemia	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 cases (19.95%) were taken as 2<sup>nd</sup> repeated anomaly[10]. CVD system 61 cases (16.44%) was 3<sup>rd</sup> repeated anomaly followed by 35 cases (9.43%) of respiratory system. Genitourinary system involvement was observed in 18 cases (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

TeachingHospital 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 percent cases 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 cases and regular antenatal 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**

Important reason of hospital admission are congenital anomalies specially in the admission of NICU. Congenital anomalies prevalence was

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

## REFERENCES

1. Miller, E., et al., Elevated maternal hemoglobin A1c in early pregnancy and major congenital anomalies in infants of diabetic mothers. *New England Journal of Medicine*, 1981. 304(22): p. 1331-1334.
2. Miller, D.T., et al., Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. *The American Journal of Human Genetics*, 2010. 86(5): p. 749-764.
3. Pagon, R.A., et al., Coloboma, congenital heart disease, and choanal atresia with multiple anomalies: CHARGE association. *The Journal of pediatrics*, 1981. 99(2): p. 223-227.
4. Hwang, D.-Y., et al., Mutations in 12 known dominant disease-causing genes clarify many congenital anomalies of the kidney and urinary tract. *Kidney international*, 2014. 85(6): p. 1429-1433.
5. Vivante, A., et al., Exome sequencing discerns syndromes in patients from consanguineous families with congenital anomalies of the kidneys and urinary tract. *Journal of the American Society of Nephrology*, 2017. 28(1): p. 69-75.
6. Cohen, E., J.G. Ray, and H.T. Sørensen, Re: addressing mortality in mothers of infants with congenital anomalies. *Journal of Public Health and Emergency*, 2018. 2(2).
7. Ludwig, W.W., S.D. Goldstein, and J.P. Gearhart, Bladder exstrophy and postoperative intussusception due to Meckel's diverticulum: A confluence of congenital anomalies. *Journal of Pediatric Surgery Case Reports*, 2017. 16: p. 22-24.
8. Vivante, A., et al., Single-gene causes of congenital anomalies of the kidney and urinary tract (CAKUT) in humans. *Pediatric nephrology*, 2014. 29(4): p. 695-704.
9. Wemakor, A., et al., Selective serotonin reuptake inhibitor antidepressant use in first trimester pregnancy and risk of specific congenital anomalies: a European register-based study. *European journal of epidemiology*, 2015. 30(11): p. 1187-1198.
10. Di Spiezio Sardo, A., et al., The comprehensiveness of the ESHRE/ESGE classification of female genital tract congenital anomalies: a systematic review of cases not classified by the AFS system. *Human reproduction*, 2015. 30(5): p. 1046-1058.
11. Heidet, L., et al., Targeted exome sequencing identifies PBX1 as involved in monogenic congenital anomalies of the kidney and urinary tract. *Journal of the American Society of Nephrology*, 2017. 28(10): p. 2901-2914.
12. Gili, J.A., et al., High birth prevalence rates for congenital anomalies in South American regions. *Epidemiology*, 2015. 26(5): p. e53-e55.
13. Ornoy, A., et al., Effect of maternal diabetes on the embryo, fetus, and children: congenital anomalies, genetic and epigenetic changes and developmental outcomes. *Birth Defects Research Part C: Embryo Today: Reviews*, 2015. 105(1): p. 53-72.