

NEED FOR NOTIFYING AND PREVENTION OF CONGENITAL ANOMALIES

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ABSTRACT

Objective: To determine the frequency of different systems involved in congenital anomalies to prevention and notifying strategies.

Study Design: Descriptive observational study.

Place and Duration of Study: The study was carried out at Military Hospital, Rawalpindi, from Jan 2011 to Aug 2013.

Material and Methods: The study was carried out at Military Hospital, Rawalpindi. The data of all the neonates with congenital anomalies was analyzed in term of sex and various system involved. Various type of tests were used for the diagnosis of different defects like, barium studies, computed topography, magnetic resonance imaging, echocardiography, x-rays, ultrasound.

Results: Total number of admissions during the study period were 4201 out of which 371 (8.83%) were diagnosed cases of congenital anomalies. Of these babies 220 (59.30%) were male, 142 (38.27%) were females and the remaining 9 (2.43%) were with ambiguous genitalia 215 (57.95%) were delivered by normal vaginal delivery and rest 156 (42.05%) by lower segment caesarian section. The most common system involved was central nervous system (CNS) 89 (23.99%), followed by gastro intestinal tract 74 (19.95%), cardiovascular system 61 (16.44%), respiratory system 35 (9.43%) and genito urinary system 18 (4.85%). Babies with dysmorphic features were 48 (12.94%) and with cleft lip and palate 17 (4.58%). The remaining 29 (7.82%) were grouped in miscellaneous category.

Conclusion: Congenital anomalies are an important cause of admission to NICU. The prevalence of congenital anomalies was found more in males than in females. In our set up the most common system involved in congenital anomalies is central nervous system followed by gastrointestinal tract and cardiovascular systems

Keywords: Congenital anomalies, Cleft lip, Hydronephrosis, Meningomyelocele, Tetra logy of fallot.

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INTRODUCTION

Congenital anomalies can be defined as an abnormality of function, body metabolism or structure that is present at birth and is fatal or result in mental or physical disability¹. In both developed and developing countries congenital anomalies remain important cause of mortality². In the developing countries prevalence of congenital anomalies is under estimated due to lack of diagnostic capabilities and poor record of health statistics³. There is considerable variation in the prevalence of congenital anomalies from one region to another⁴. This variation may be due to racial, social and ecological differences in

different part of the world⁵. To plan for predunction of the incidence of congenital anomalies it is very important to know the prevalence of congenital anomalies not only for every country but also for every region⁶.

The incidence of congenital anomalies is about 5%¹. Of these 5%, 2 or 3% of congenital anomalies can be diagnosed prenatally through invasive or noninvasive tests or at birth and the remaining 2% can be diagnosed during the first year of life¹. The etiology of congenital anomalies cannot be established in 60 to 80% of the cases. Environmental causes are responsible for 10 to 20% of cases and genetic factors are responsible for the remaining 10 to 20% of the cases. Congenital anomalies are responsible for 12 to 32% of the peri natal deaths⁷. Use of multivitamins containing folic acid by women

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during peri conception period not only reduce the incidence of neural tube defects but also decrease the incidence of other birth defects like limb defects, heart defects, oral facial clefts and pyloric stenosis etc⁸.

To reduce the incidence of congenital diseases it is important to adopt comprehensive and integrated strategy combined with population screening, genetic counseling, community education and best possible treatment and prevention⁹.

Rationale of our study is that in most of our institutes there is no congenital anomalies registry. Our study will highlight the heavy load of congenital anomalies which in most cases are preventable. It will further signify the need for notifying and prevention of congenital anomalies. The aim of our study was to determine the frequencies of different systems involved in congenital anomalies in neonates admitted in our NICU.

MATERIAL AND METHODS

This descriptive observational study was carried out in Military Hospital, Rawalpindi. It is a tertiary care hospital with well-equipped nursery intensive care unit. The study period extended from Jan 2011 to Aug 2013. This hospital has state of the art diagnostic tool, so patients with congenital anomalies or with suspicion of congenital anomalies are referred to this hospital from almost all part of Pakistan. Also complicated pregnancies or with antenatally diagnosed congenital anomalies are referred to this hospital for delivery. The record of all patients with congenital anomalies were documented in our NICU register. All the babies with congenital anomalies were included in the study. All radiological investigations like MRI, CT-Scan, X-Rays, Ultrasonography and Barium studies required for diagnosis were done in Armed Force Institute of Imaging and Radiology (AFIRI), blood tests required were sent to Armed Force Institute of Pathology (AFIP) and for echocardiography and angiography patients were sent to Armed Force Institute of Cardiology

(AFIC). Data was analyzed using SPSS Version 18. Frequencies and percentages were calculated for categorical variables like sex distribution, mode of delivery and system involved.

RESULTS

Total no of neonates with congenital anomalies was 371 making 8.83% of the total admissions to nicu which was 4201. Male babies with congenital anomalies were 220 (59.30%), 142 (38.27%) were females and the remaining 9 (2.43%) were with ambiguous genitalia.

Babies delivered by vaginal delivery were 215 (57.95%) and those delivered by lower segment caesarian section were 156 (42.05%). The most common system involved was CNS 89 (23.99%), with meningomyelocele and hydrocephalus making 60 (69.77%) and 21 (23.60%) of the CNS anomalies, respectively.

The second most common anomalies were that of gastro intestinal tract 74 (19.95%). Cardiovascular system 61 (16.44%) was the third most common system involved followed by respiratory system 35 (9.43%) babies and Genito urinary system 18 (4.85%) anomalies 8 (50%) as given in table.

DISCUSSION

Children with congenital anomalies who survive infancy are at increased risk of morbidity due to various health problem and most of them are affected socially, mentally and physically¹⁰. Congenital anomalies are still a major cause of antepartem death in developing countries¹¹. Total number of admissions to our neonatal intensive care unit (NICU) during the study period was 4201, out of which 371 were with congenital anomalies. The incidence of congenital anomalies was more common in males 59.30% than in females 38.27% with 2.42% having ambiguous genitalia. This is compatible with other studies which also showed increased incidence of congenital anomalies in male than in females^{12,13}. The most affected system in our study was central nervous system with neural tube defects making the most common CNS anomalies. These birth defects develop due to lack of closure of

neural tube during third and fourth week after conception¹⁴. Other studies have also reported CNS anomalies to be the most common^{15,16}. As

decreased possibly due to increased multi vitamins supplementation containing folic acid

Table: Frequency of different systems involved in congenital anomalies.

Systems involved	Frequency (%)
Central Nervous System (n=89) (29%)	
Meningo Myelocele	60 (67%)
Hydrocephalus	21 (24%)
Encephalocele	4 (5%)
Microcephaly	3 (3%)
Spina Bifida	1 (1%)
Gastro Intestinal Tract (n=74) (24%)	
Imperforate Anus	40 (54%)
Small Gut Atresia	21 (28%)
Hirschprung Disease	3 (4%)
Anorectal Stenosis	10 (14%)
Respiratory System (n=35) (11%)	
Tracheo Esophageal Fistula	30 (85%)
Tracheal Atresia	2 (6%)
Hypoplastic Lung	1 (3%)
Choanal Atresia	2 (6%)
Cardio Vascular System (n=61) (20%)	
Tetralogy of Fallot	13 (21%)
Ventricular Septal Defect	26 (43%)
Atrial Septal Defect	9 (15%)
Coarctation of Aorta	2 (3%)
Patent ductal arterioses	3 (5%)
Transposition of great arteries	1 (2%)
Dilated Cardio Myopathies	2 (3%)
Complex Anomalies	5 (8%)
Genito Urinary System (n=18) (6%)	
Ambiguous Genitalia	9 (50%)
Congenital Hydronephrosis	9 (50%)
Miscellaneous (n=29) (10%)	
Congenital Diaphragmatic Hernia	8 (28%)
Omphalocele	8 (28%)
Down Syndrome	5 (18%)
Pierre Robin Syndrome	2 (7%)
Edward Syndrome	1 (3%)
Sacrocygeal Teratoma	1 (3%)
Cystic Hygroma	1 (3%)
Right Malformed Ear	1 (3%)
Epigastric Hernia	2 (7%)

these anomalies are apparent at birth and can be easily detected, this may be the reason for their increased incidence¹⁷. In some developed countries the incidence of neural tube defects has

during the peri conception period¹⁸.

Other systems involved in our study after CNS in descending orders were GIT 19.95%, CVS 16.44%, Respiratory 9.43% and Genito-Urinary

system 4.85%. Study by Tuncbilek et al¹⁶ showed urinary system 14.4%, musculo skeletal system 11.70% and cardiovascular systems 8.28% to be the most common systems involved after CNS. While Himmetoglu et al¹⁹ reported CNS, Genito-Urinary, facial and musculo skeletal systems to be involved in descending order of frequency. Study done in Pakistan at Ayub Teaching Hospital, Abbottabad²⁰ showed CNS involvement in 31% cases with NTD's making 77% of the CNS anomalies. That study showed the incidence of congenital heart defects to be 16% and urogenital anomalies to be 6%. These results are comparable to our study. The incidence of Cleft lip/Palate in our study was 4.58%. Cleft lip/palate is seen in every 600-800 live birth. In 60% of the cases they occur with other anomalies²¹.

It has been reported in some literatures that about 50% of the congenital anomalies are preventable²². To decrease the incidence of birth defects in our country we should have population based registry of congenital anomalies, regular antenatal checkup and to ensure that female in childbearing age should take food supplements having multi vitamins.

CONCLUSION

Congenital anomalies are an important cause of admission to NICU. The prevalence of congenital anomalies was found more in males than in females. In our set up the most common system involved in congenital anomalies is central nervous system followed by gastrointestinal tract and cardiovascular systems.

CONFLICT OF INTEREST

This study has no conflict of interest to declare by any author.

REFERENCES

- Ruth Kohut RN, Rusen ID. Health Canada. Congenital anomalies in Canada a perinatal health report 2002.
- Sipek A. Incidence of congenital defects in selected areas and countries 1998-1998. *Ceska Gynekol* 2002; 67(4): 202-09.
- Penchaszadeh VB. Preventing congenital anomalies in developing countries. *Community Genet* 2002; 5(1): 61-9.
- Tomatir AG, Demirhan H, Sorkun HC, Koksak A, Ozerdem F, Cilengir N. Major congenital anomalies: a five-year retrospective regional study in Turkey. *Genet Mol Res* 2009; 8(1): 19-27.
- Temtam SA, Meguid NA, Ismail SI, Ramzy MI. A new multiple congenital anomaly, mental retardation syndrome with preaxial brachydactyly, hyperphalangism, deafness and orodental anomalies. *Clin Dysmorphol* 1998; 7(4): 249-55.
- Prsa M, Saroli T, Correa JA, Asgharian Mackie AS, Dancea AB. Birth prevalence of congenital heart disease. *Epidemiology* 2010; 21(2): 275-77.
- Aksoy F. Congenital Anomalies: Identification, Classification, Terminology and Anomalyed Fetus Investigation. *Türk Patol Dergisi* 2001; 17: 57-62.
- Czeizel AE. Reduction of urinary tract and cardiovascular defects by periconceptional multivitamin supplementation. *Am J Med Genet* 1996; 62: 179-83.
- WHO. Control of genetic diseases (Executive Board EB116/3). World Health Organization, Geneva 2005.
- WHO. Primary Health Care Approaches for Prevention and Control of Congenital and Genetic Disorders (WHO/HGN/WG/00.1). World Health Organization, Geneva 2000.
- Khaskheli M, Baloch S, Khushk IA, Shah SS. Pattern of fetal deaths at a university hospital of Sindh. *J Ayub Med Coll Abbottabad* 2007; 19(2): 32-4.
- Miliaras D, Meditskou S, Ketikidou M. Increased male proportion in fetal deaths and in fetuses with congenital malformations in Greece. *Hum Reprod* 2008; 23(10): 2385-86.
- Golalipour MJ, Ahmadpour-Kacho M, Vakili MA. Congenital malformations at a referral hospital in Gorgan, Islamic Republic of Iran. *East Mediterr Health J* 2005; 11(4): 707-15.
- Jones KL. Smith's recognizable patterns of human malformation. 5th ed. Philadelphia: WB Saunders; 1997. p. 608.
- De Galan-Roosen AE, Kuijpers JC, Meershoek AP, Van Velzen D. Contribution of congenital malformations to perinatal mortality. A 10 years prospective regional study in the Netherlands. *Eur J Obstet Gynecol Reprod Biol* 1998; 80: 55-61.
- Tunçbilek E, Boduroglu K, Alikasifoglu M. Results of the Turkish congenital malformation survey. *Turk J Pediatr* 1999; 41: 287-97.
- Göynüner FG, Kepkep K, Yetim G, Tuncay Y. Retrospective analysis of major congenital anomalies in dogs. *Perinatol Magazine* 2005; 13: 31-4.
- Health Canada. Canadian perinatal health report 2000. Ottawa: Minister of Public Works and Government Services Canada; 2000.
- Himmetoglu O, Tiras MB, Gursoy R, Karabacak O. The incidence of congenital malformations in a Turkish population. *Int J Gynaecol Obstet* 1996; 55: 117-21.
- Kazmi NH, Najeeb S, Hussain S, Raza A. Frequencies of congenital anomalies among newborns admitted in nursery of ayub teaching hospital Abbottabad, Pakistan. *J Ayub Med Coll Abbottabad* 2001; 23(1): 117-21.
- Biri A, Onan A, Korucuoglu Ü, Tiras B. The frequency and distribution of congenital malformations in a university hospital. *Perinatol Magazine* 2005; 13: 86-90.
- Czeizel AE. Prevention of congenital abnormalities by periconceptional multivitamin supplementation *BMJ* 1993; 306: 1645-8.