



PROFILE OF CONGENITAL BIRTH DEFECTS IN NEWBORN: A HOSPITAL BASED STUDY

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ABSTRACT

BACKGROUND: Birth defects are abnormalities in body structure and function they can range from minor to life threatening abnormalities. They can cause long term morbidity, disability and increased mortality in newborn and infancy.

AIMS AND OBJECTIVES: To study the incidence, various patterns, and presentations of Congenital Birth defects.

MATERIALS AND METHODS: This hospital based descriptive Cross-sectional study was conducted in a tertiary care hospital. All the newborns presenting to newborn unit with congenital abnormalities were included. Data was collected and was entered into Excel sheet and analyzed.

RESULTS: Total 222 babies had congenital birth defects with incidence of 4.9%. Of total 208 congenital birth defects babies 165 were having congenital heart defects. Central nervous system defects were next common defects in babies after cardiovascular defects with 21 babies having defects.

CONCLUSION: Cardiovascular defects were common than any other system involved followed by central nervous system defects. A system should be put in to screen all babies with pulse-oximetry to detect cardiovascular defects before discharge and plan for further investigations.

KEYWORDS : Congenital, Birth defect, Cardiovascular, Central nervous system.

1. INTRODUCTION

Birth defects (also called congenital anomalies) are defined as abnormalities of body structure or function that are present at birth and are of prenatal origin. Birth defects contribute to a significant proportion of perinatal, neonatal and child mortality (1,2). Birth defects are a significant but under-recognized cause of mortality and morbidity among infants and children under five years of age and especially for foetal (including stillbirth) and neonatal deaths (3).

Birth defects are ranging from minor to serious. There are more than 4,000 different birth defects, in which some of the defect is treated and cured and other are not treated (4). Globally, about 7.9 million children are born annually with a serious birth defect of genetic or partially genetic origin, as per the estimates provided by March of Dimes Report (2006). WHO estimates that out of 1 million neonatal deaths in 2012 in the South-East Asia Region, about 46 000 (4.6%) were caused by birth defects (2). Congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India (5). A major congenital anomaly affects 2-3% of new born babies (4).

The life threatening congenital malformations must be identified through clinical examination because early diagnosis and surgical correction or palliation of these infants offers the best chance for survival (6). Prevalence studies of congenital anomalies are useful to establish baseline rates, to document changes over time, and to identify clues to etiology. They are also important for planning and evaluating antenatal screening for congenital anomalies, particularly in high risk populations (7). The purpose of the present study is to present the spectrum of congenital malformations in the newborns in our institute.

2. MATERIALS & METHODS

This hospital based descriptive Cross-sectional study was conducted in a tertiary care hospital in from September 2016 to august 2017.

Inclusion Criteria: Newborns in labour room, postnatal wards and also all newborns presenting to our hospital with congenital birth defects. Exclusion Criteria: Mothers not willing to participate in the study. Newborns presenting with congenital birth defects were thoroughly examined, the data detailing the antenatal history, birth time and postnatal period were either interviewed from mothers or were taken from case records. Necessary investigations were done to diagnose the defect and data was entered into pro forma and later into Excel sheet.

3. RESULTS

All newborns were examined thoroughly for birth defects. Total admission during study period was 13750 babies. Total 222 babies (1.6%) have congenital birth defects. Males babies were 108 (49%) and females were 114 (51%) with congenital birth defects. Of total 222 congenital birth defects babies, 133 (60%) were term babies and 89 (40%) were preterm babies. There were total of 104 (47%) low birth weight (<2500 grams) as seen in Table 1.

Table 1 shows Newborn characteristics of study group.

GENDER	MALES	108 (49%)
	FEMLES	114 (51%)
GESTATION	TERM	133 (60%)
	PRETERM	89(40%)
BIRTH WEIGHT	>2500 GRAMS	118 (53%)
	<2500 GRAMS	104 (47%)

In newborns with congenital birth defects cardiovascular defects were common than any other system involved. Of total 222 congenital birth defects babies 165 were having congenital heart defects. Central nervous system defects were next common defects in babies after cardiovascular defects with 21 babies having defects. (Table 2)

Table 2: Systems involved and birth defects with ICD 10 classification.

System	Birth defect	ICD 10 Classification	Number of cases
Congenital malformations of the nervous system (Total n=21)	Anencephaly	Q00.00	3
	Craniorachischisis	Q00.1	1
	Congenital hydrocephalus, unspecified	Q03.9	2
	Congenital malformations of corpus callosum	Q04.0	3
	Congenital cerebral cysts	Q04.6	1
	Cervical spina bifida with hydrocephalus	Q05.0	1
	Lumbar spina bifida with hydrocephalus	Q05.2	1
	Sacral spina bifida with hydrocephalus	Q05.3	1
	Lumbar spina bifida without hydrocephalus	Q05.7	5
	Sacral spina bifida without hydrocephalus	Q05.8	2
	Arnold-Chiari syndrome	Q07.0	1
Congenital malformations of eye, ear, face and neck (n=2)	Anophthalmos	Q11.1	2
Congenital malformations of the circulatory system (n= 165)	Double outlet right ventricle	Q20.1	1
	Discordant ventriculoarterial connection	Q20.3	1
	Atrial septal defect	Q21.1	55
	Ventricular septal defect	Q21.0	31
	Other specified atrioventricular septal defect	Q21.28	1
	Tetralogy of Fallot	Q21.3	3
	Congenital tricuspid stenosis	Q22.4	1
	Ebstein's anomaly	Q22.5	2
	Hypoplastic left heart syndrome	Q23.4	2
	Dextrocardia	Q24.0	4
	Patent ductus arteriosus	Q25.0	57
	Coarctation of aorta	Q25.1	4
	Total anomalous pulmonary venous connection	Q26.2	3
Congenital malformations of the respiratory system (n=3)	Congenital (cystic) adenomatoid malformation of the lung	Q33.80	2
	Other congenital malformations of respiratory system	Q34	1
Cleft lip and cleft palate (n= 8)	Cleft hard palate with cleft soft palate, unspecified	Q35.5	1
	Cleft hard palate with cleft soft palate, unilateral	Q35.50	1
	Cleft hard palate with cleft lip, bilateral	Q37.0	3
	Atresia of oesophagus with tracheo-oesophageal fistula	Q39.1	3
Other congenital malformations of the digestive system (n=4)	Congenital absence, atresia and stenosis of duodenum	Q41.0	1
	Congenital absence, atresia, stenosis of anus without fistula	Q42.3	3
Congenital malformations of genital organs (n=1)	Undescended testicle, bilateral	Q53.2	1
Congenital malformations of the urinary system (n=5)	Renal agenesis, bilateral	Q60.1	1
	Congenital hydronephrosis	Q62.0	3
	Ectopic kidney	Q63.2	1
Congenital malformations and deformations of the musculoskeletal system (n=13)	Longitudinal reduction defect of radius	Q71.4	2
	Congenital diaphragmatic hernia	Q79.0	7
	Exomphalos	Q79.2	1
	Gastroschisis	Q79.3	2
	Prune belly syndrome	Q79.4	1

4. DISCUSSION

Congenital anomalies are not only major cause of morbidity and mortality in newborn period, they also are contributes a significant infantile and childhood morbidities (4). The congenital malformations prevalence varies all around the globe ranging from 100 to over 450 per 10,000 births (7). In our study incidence of congenital birth defects was 1.6% compared to other studies like Jain SR et al had incidence of 2.69% (6), Kokate P et al had incidence of 0.9% (8), and study done by Shah et al incidence of congenital malformations was found to be 2.38% (9).

In our study we have observed that cardiovascular system defects were more common followed by the central nervous system defects but various studies have varied presentation in most common system involved. Our study had similar observation to other studies like Keerti Singh et al, and Jain SR et al (6,7) where cardiovascular system was most common followed by other systems. Kokate P et al and Shah et al studies have shown central nervous system defects were most common found (8,9). In study done by El Koumi et al found musculoskeletal system was the most commonly involved followed by the central nervous system (10). There is lot of variability observed in prevalence and presentation of the congenital birth defects due to difference in collection of the data and also methodology in collecting the data (7).

There was rapid decline in deaths caused by the infection and perinatal asphyxia which were major causes of mortality in newborn as there was improvement in health care and quality of the newborn care in developed and developing countries over past few years, whereas the morbidity and mortality due to major congenital defects remained same. This led to impression that there was increase in mortality due to birth defects have increased over the decade time (3).

Birth defects can be caused by various etiologies like single gene defects, chromosomal disorders, and multifactorial inheritance, or from Maternal infections such as rubella, maternal illnesses like diabetes mellitus, iodine and folic acid deficiency, exposure to various drugs, chemicals or radiations during pregnancy can lead to varying presentation of congenital birth defects (6).

Birth defects can range from minor abnormalities to major life threatening malformations. These babies not only have increased mortality in newborn period they also have increased morbidity and long term disabilities. Birth defects can cause lifelong impairment which can effect productivity of the person and also burden to family and society as these babies need lifelong support for drugs and treatment. A simple cost effect programmes like folic acid fortification have found to prevent 13% of neonatal deaths due to major congenital

anomalies. Such preventive programmes can decrease mortality and also economic burden to society. (3)

5. CONCLUSION

Incidence of congenital birth defects was 1.6%. Cardiovascular system malformations were most common compared to other system defects. Early antenatal and postnatal diagnosis and treatment could decrease morbidity and mortality in newborn and infancy.

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