Original Research Article

Evaluation of congenital malformations at birth in a tertiary centre

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Abstract

Introduction: Congenital malformations are one of the important causes of still births and infant mortality. The incidence of congenital malformations in infants is on rise in antenatal and during neonatal period due to advanced diagnostic technology, especially sonography and fetal echocardiography. **Objective:** To determine the incidence and type of congenital malformations among neonates delivered at Krishna hospital and research centre. **Methodology:** This prospective study was conducted in Krishna Institute of Medical Sciences from 1st January 2013 to 31 December 2014 (2 years). During the study period, all newborns delivered at Krishna hospital were examined clinically and investigated further, if required. **Results:** Out of the total 9651 deliveries conducted during study period, 9439 were live births and 212 were stillbirths. The total number of babies with congenital malformations was 246 (2.54%). Out of the 9472 singleton births, 192 (2.02%) were malformed. 71 of the 246 malformed babies (16.67%) were still born. The maternal tobacco chewing and smoking was found to have a higher risk of congenital anomalies. CNS malformations were most common in our study followed by cardiovascular malformations. **Conclusion:** Congenital malformations are a major cause of perinatal mortality at present. Clinical evaluation of various systems of high risk newborns to rule out congenital anomalies is the important factor to be considered.

Keywords: congenital malformations, musculoskeletal, screening, cleft lip/palate.

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INTRODUCTION

The WHO defines Congenital anomalies as structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth or later in life. Congenital anomalies may be genetic, infectious, nutritional or environmental in origin; most often it is difficult to identify the exact causes. Antenatal detection of congenital anomalies gives the opportunity to prepare parents for the birth of a child with a congenital anomaly, to plan postnatal management or to consider termination of pregnancy.[1] Birth defects are a group of disorders of antenatal origin that can be caused by gene defects, chromosomal abnormalities, multifactorial inheritance, environmental teratogens and iatrogenic. Maternal infections such as rubella, toxoplasmosis, maternal illnesses like diabetes mellitus (DM), iodine and folic acid deficiency, exposure to medicinal and other addictive substances like tobacco chewing or smoking and higher doses of radiations are the factors that cause birth defects.[2]

MATERIALS AND METHODS

This prospective study was conducted at Krishna Institute of Medical Sciences from 1st January 2013 to 31 December 2014 (2 years). During the study period, all newborns delivered at Krishna hospital were examined clinically and investigated further, if required. The newborns with structural congenital malformations were identified from the antenatal case records and on clinical examination. The functional anomalies (e.g. metabolic disorders) were excluded from our study. All the babies were routinely examined within 1 hour of birth. Intrauterine death (foetal death before the onset of labor), stillbirth (death during the process of delivery), and death immediately after live birth without any physical stress were all considered as stillbirth in this study. The parity

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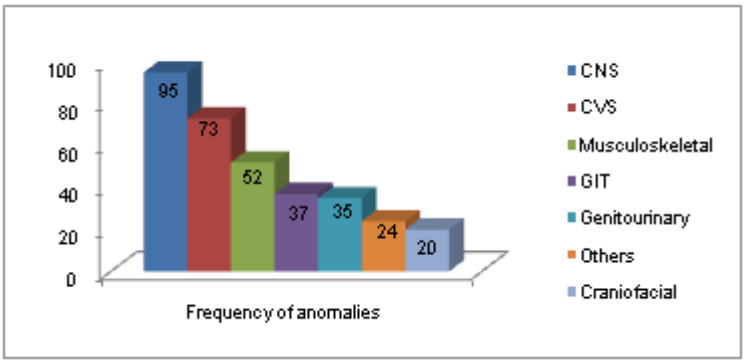
and age of mother of malformed newborns were noted and analysed. The paternal habits of smoking, chewing tobacco and alcohol consumption were also recorded. Mortality was defined as death during hospital stay. The diagnosis of congenital malformations is based on analysis of antenatal records, clinical examination and further investigations, if required.

OBSERVATIONS AND RESULTS

Total 9651 consecutive newborns including both live and still born babies during study period (1st January 2013 to 31 December 2014) were examined for visible structural anomalies and required investigation was conducted to determine the overall incidence of congenital malformations. In our study total numbers of malformed babies were 246, so the point incidence of congenital anomalies is 2.54%. Out of these, 177(72%) had a single congenital malformation and the 69 (28%) had multiple malformations. Thus, there were a total of 336 anomalies among 246 malformed babies. **Systemic Distribution of Congenital Malformed Babies:** As far as systemic distribution of congenital malformations is analysed, in our study, it was observed that out of total 336 malformations, 95 were central nervous system malformations making most common.



Congenital malformations in relation to maternal age and consanguinity: Our study has found that congenital malformations were more common in babies born out of consanguineous marriage. This is in agreement with results published by M L Kulkarni *et al* in his study.[3] Prenatal counselling should be done for such couples as consanguineous marriage are very much prevalent in Maharashtra.

Table 1

Factors	Total (N-9651)	Malformed Newborns (N - 246)
Maternal Age (Years)		
<20	1266(13.12%)	42(17.07%)
20-35	7552(78.25%)	164(66.67%)
>35	833(8.63%)	40(16.30%)
Consanguinity		
Non Consanguineous Marriage	6996(72.5 %)	176(71.54%)
Consanguineous Marriage	2655(27.5%)	70(28.45%)

Congenital malformations in relation to maternal nutrition and tobacco use: Those babies born to under-nourished were more prone to have congenital malformations. Maternal tobacco chewing and smoking was associated with higher percentage of congenital malformations as compared to general population.[9] American Heart Association (AHA) also concluded that maternal smoking (active or passive) early in their pregnancies are more likely to have babies with certain types of CHD.[4]

Table 2

Factors	Total (N-9651)	Malformed Newborns (N - 246)
Maternal Nutrition		
Well Nourished	4562(47.30%)	54(22.0%)
Undernourished	4577(47.42%)	151(61.40%)
Obese	512(5.30%)	41(16.70%)
Addictions		
Tobacco Chewing	856	31(3.62%)
Smoking	74	3(4.05%)

Congenital malformations in relation to gender and weight of newborns: Among 246 babies, 132 babies were male, and 111 babies were female. The low birth weight category babies were more prone to develop congenital malformations. This association of LBW and malformations has been well documented in study by Singh A *et al*.[5] Total 19 newborns in our study had a family history of a congenital anomaly. It was also observed from the records of neonates with congenital malformations that they had higher incidence of neonatal seizures as compared to normal newborns in maternity

ward. This aspect need to be further studied as neonatal seizures are one of the most important neurologic events which contribute to both mortality and morbidity in neonatal period.[6]

Table 3

Factor	Malformed Newborns (N - 246)	Percentage
Gender		
Male	132	51.56%
Female	111	45.12%
Amibigious Genitalia	3	1.21%
Birth Weight		
< 2500 G	138	56.09%
≥ 2500 G	108	43.90%
Anamoly In Family		
Present	19	7.72%
Absent	227	92.27%

Systemic Distribution of Congenital Malformed Babies: In our study, the incidence of CNS malformations was higher than other systemic anomalies. This observation is comparable with the results of other studies by Mohan *et al*[7] and Sankar and Phadke.[8] Neural tube defects and congenital cardiac malformations both have an incidence of around 10 per 1000 live births which is in agreement with our findings.[9] Myelomeningocele is one of the most common neural tube defects in our study; various literatures have also shown same results.[10] Total number of still births in our hospital during study period were 149 and out of it 71 presented with various malformations. Out of 336 congenital malformations, 209 were diagnosed on antenatal investigations.

Table 4

System	Malformations	No. Of cases (n-336)	%
CENTRAL NERVOUS SYSTEM (N-95) (28.27%)	Anencephaly	18	5.35
	Myelomeningocele	18	5.35
	Hydrocephalus	14	4.16
	Encephalocele	13	3.86
	Microcephaly	12	3.57
	Meningocele	06	1.78
	Spina bifida occulta	06	1.78
	Sacral agenesis	05	1.48
	Corpus callosum agenesis	03	0.89
CARDIOVASCULAR (N-73) (21.72%)	Ventricular septal defect	26	7.73
	Patent ductus arteriosus	18	5.35
	Atrial septal defect	14	4.16
	Tetralogy of Fallots	06	1.78
	TGV	04	1.19
	Dextrocardia	04	1.19
	Acardia	01	0.29
MUSCULOSKELETAL (N-52) (15.47%)	Talipes equinovarus	12	3.57
	Polydactyly	11	3.27
	Bone malformations	08	2.38
	Rib malformations	06	1.78
	Syndactyly	06	1.78
	DDH	04	1.19
	Osteogenesis imperfecta	03	0.89
	Infantile scoliosis	02	0.59
GASTROINTESTINAL (N-37) (11.01%)	Tracheo-esophageal fistula	12	3.57
	Imperforate anus	07	2.08
	Diaphragmatic hernia	04	1.19
	Omphalocele	04	1.19
	Gastrochesis	03	0.89
	Duodenal atresia	03	0.89
	Intestinal stenosis	02	0.59
	Inguinal hernia	02	0.59
GENITOURINARY (N-35) (10.41%)	Hypospadiasis	09	2.67
	Unilateral undescended testis	07	2.08
	Hydronephrosis and ureterocele	06	1.78
	Bilateral undescended testis	05	1.48
	Hydrocele	03	0.89
	Amibigious genitalia	03	0.89
	Exstrophy of bladder	02	0.59
CRANIOFACIAL (N-20) (5.95%)	Hypoplasia DAOM	06	1.78
	Cleft lip	06	1.78
	Cleft palate	03	0.89
	Microphthalmia	02	0.59
	Congenital cataract	02	0.59
	Anophthalmia	01	0.29
OTHERS (N-24) (7.14%)	Single umbilical artery	12	3.57
	Pre-auricular tag	05	1.48
	Hemangioma	05	1.48
	Pulmonary hypoplasia	02	0.59

DISCUSSION

Birth defects are common - about 2% of babies are born with medically significant malformations which may be recognized on first day of life and 4% of infants will have their malformations diagnosed by 1 year of age.[11] Congenital malformations are traumatizing conditions that results in physical or mental disability. Many birth defects are incompatible with life and therefore result in spontaneous or induced abortion and early neonatal mortality. In this prospective study, we have made attempts to find the total and individual systemic incidence of anomalies in our hospital and to find causal relationship and association, if any, between various

etiological factors and congenital anomalies. Due to effective prevention and treatment of neonatal sepsis and nutritional deficiencies, congenital malformations have now become an important cause of perinatal mortality and morbidity in developed countries and will very soon become an important determinant of perinatal mortality in developing countries.[12] The main aim of maternal and child health intervention in India, as well as other developing countries is reduction of child mortality due to preventable causes. Until now, congenital malformations had remained a low public health priority with no programs for the prevention and care for the affected[13]. A worldwide assessment of the prevalence of congenital malformations was conducted by Christianson A *et al*[14]. India had ranked 38th among all countries in terms of the prevalence of birth defects (India 64.3/1000). In terms of the absolute number of affected children, India had the highest estimated numbers of live or stillbirths due to birth defects (estimated 1,613,502 births). This figure was one and a half times more than China, which had the second highest estimated global burden of children born with birth defects.[15] The nature of congenital malformation in a newborn will decide the investigation to be done. The neurosonography, MRI and CT brain (plain or contrast) is required to access CNS malformations like absent corpus callosum. Echocardiography should be done in patients with abnormal cardiovascular examination or to find out associated anomaly. Chromosomal analysis and gene studies will rule out abnormal genetic patterns. Medical field has now entered the molecular era. The problem of congenital malformations has to be solved with combined efforts of scientists, doctors and society.[16]

CONCLUSION

The genetic disorders may be prevented with appropriate genetic counselling as these conditions are extremely traumatic to both, patients and families. The development of a congenital malformation yojana with aim of prevention, care, surveillance, research and treatment is need of hour, as congenital malformations account for a significant burden on patient, parents and family. The diagnosis of life threatening congenital malformations in early newborn period can give way for surgical correction or palliation of anomalies of these infants.

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