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Prevalence and pattern of congenital anomalies at tertiary health care hospital

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Abstract

Background: Congenital abnormality is an abnormality of structure, function or body metabolism that is present at birth and results in physical or mental disability or is fatal. Congenital anomalies are the important cause of perinatal, early neonatal and infant morbidity and mortality. The long term disability caused by congenital anomalies may have a significant impact not only on the child's wellbeing and development but also on families, health care systems and societies

Material & Method: It was an observational hospital based study conducted in Department of Obstetrics and Gynaecology & Department of Paediatrics, NSCB Medical College and Hospital, Jabalpur. Total 64 cases of congenital malformation detected antenatally or at birth were analysed within a study period of one year

Result: Total 64 cases of anomalies were found out of 5885 deliveries. The prevalence of congenital malformation was 1.08%. Majority were central nervous system anomaly 56.25%. Renal anomalies were 10.93%, multiple system involvement were in 7.8%, gastro-intestinal in 7.8%, skeletal anomalies were 6.25%, followed by respiratory, cardiovascular, lymphatic & soft tissue in 4.68%, 3.12%, 1.56%, 1.56% respectively.

Conclusion: Congenital anomalies are important cause of still births and infant mortality, and are also contributors to childhood morbidity. Hence prenatal diagnosis by proper antenatal care and targeted scan is mandatory.

Keywords: Congenital anomaly, prevalence, malformation, pattern

Introduction

Congenital abnormality is an abnormality of structure, function or body metabolism that is present at birth and results in physical or mental disability or is fatal. Congenital anomalies are the important cause of perinatal, early neonatal and infant morbidity and mortality worldwide [1]. In the Global Burden of Disease study 2013 congenital anomalies are among the top ten causes for mortality in children less than five years of age [2]. In the World Health Organization (WHO) document of 1972, the term congenital malformations were confined to structural defects at birth [3]. The more recent WHO fact-sheet of September 2016, congenital anomalies are defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy [4,5]. Approximately 1 in 33 infants are affected by birth defects and result in 3.2 million birth defect related disabilities every year. An estimated 270,000 newborns die during the first 28 days of life every year from birth defects. Literature search reveals that India has the highest number of children with birth defects. Besides, birth defects may result in long-term disability, which may have significant impacts on individuals, families, healthcare systems and societies. Worldwide about 7.9 million children (6%) annually are born with a serious birth defect [8]. Most of the available Indian studies, including the data available from Birth Defect Registry of India (BDRI) [9] show that the common systems involved in birth defects are central nervous system, musculoskeletal system and cardiovascular system, with neural tube defects being the commonest [10].

Although birth defects may be genetic, infectious or environmental in origin, most often it is difficult to identify the exact cause. Many birth defects can be prevented. Penrose (1961) stressed that major advances in the prevention of defects may be achieved by attention to environmental factors rather than by attempting to improve heredity [11]. Increased use of irradiation, alkylating agents, antimetabolites, self-drugging, smoking, alcohol consumption has contributed to increased incidence of congenital malformation. Vaccination, adequate intake of folic acid (preconceptional and antenatal) and iodine, avoidance of consanguineous marriage,

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control of diabetes and avoidance of aforementioned risk factors, such as radiation exposure and antimetabolites and adequate antenatal care are keys for prevention. This study looked at the various congenital anomalies at birth and organ systems involved and their prevalence in a tertiary care hospital.

Material & Method

It was an observational hospital based study conducted in Department of Obstetrics and Gynaecology & Department of Paediatrics, NSCB Medical College and Hospital, Jabalpur, Madhya Pradesh. Total 64 cases of structural congenital malformation detected antenatally or at birth were analysed within a study period of Jan-2017 to Dec 2017. Those women who had diagnosed to have anomalous fetus either in antepartum period or at birth were included in study after informed consent. Detailed information regarding maternal age, order of pregnancy, gestational age, and consanguinity was documented. Antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. All the new born babies were examined thoroughly by the paediatrician to detect the congenital malformation. If any internal congenital malformation were suspected further investigation like Ultrasonography, ECHO, X-ray etc. were done in live babies. Congenital anomalies were diagnosed through physical examination, or through various investigations (ultrasonography, 2D echocardiography).

Results

Total 64 cases of anomaly were studied out of 5885 deliveries. The prevalence of congenital anomaly was 1.08%. Majority of women (82.81%) having anomalous foetus were in 21-30yr age group followed by <20yr (7.8%), 31-35yrs (7.8%) and >35yr (1.56%) age group [Table/Fig-1]. Most of the anomalous babies were born to primi gravida (51.56%), followed by second gravida (32.81%), third gravida (7.8%) and fourth gravida or more were (7.8%). Among all the babies 54.68% were male & 45.31% were female. 93.75% babies were diagnosed during 3rd trimester & 6.25% diagnosed during 2nd trimester. Only 6.25% of diagnosed cases have previous history of anomalous babies (table-2). Majority were central nervous system anomaly (56.25%), Renal anomalies were 10.93%, Multiple system anomalies were in 7.8%, gastro-intestinal in 7.8%, skeletal anomalies were 6.25%, followed by respiratory, cardiovascular, lymphatic & soft tissue in 4.68%, 3.12%, 1.56%, 1.56% respectively. Among the central nervous system, hydrocephalous were 28.12%, anencephaly 17.18%,

meningomyelocele 7.81%, and occipital encephalocele 3.1%. Hydronephrosis were the most frequent anomaly (7.8%) in renal system followed by polycystic kidney & multicystic dysplastic kidney each in 1.56%. 3 babies had congenital diaphragmatic hernia. Atrial septal defect were found in 2 babies. One baby diagnosed with Prune-Belly syndrome, one with down syndrome. 2 babies delivered as Hydrops foetalis, whereas one twin affected as cranio-thoracopagus. Among gastro-intestinal system foetal ascitis, oesophageal atresia, Gastroschisis, duodenal atresia & ileal atresia each of them found in 1.56% of case. One baby had Sacrococcygeal teratoma, one had lobster claw deformity, one affected with polydactyly & one had cleft lip. Cystic hygroma & cystic swelling found in 1.56% each (Table-3 & figure 1).

Table 1: Demographic profile

Maternal age	No of anomalous babies (n=64)	Percentage
≤20	5	7.8%
21-30	53	82.81%
31-35	5	7.8%
>35	1	1.5%
Locality		
Rural	38	59.37%
Urban	26	40.62%
Education of mother		
Illiterate	6	9.37%
Primary school	38	59.37%
Higher secondary	15	23.43%
Graduation	5	7.8%

Table 2: Obstetrics factors

Order of pregnancy	No of anomalous babies	Percentage
G1	33	51.56%
G2	21	32.81%
G3	5	7.8%
G4 or more	5	7.8%
Sex of baby		
Female	29	45.31%
Male	35	54.68%
Ambiguous	0	0%
Trimester of pregnancy		
1 st	0	0%
2 nd	4	6.25%
3 rd	60	93.75%
Previous H/O of anomalous baby		
Yes	4	6.25%
No	60	93.75%

Table 3: Spectrum of abnormality

System Involved	Malformation	number	Percentage
CNS	Hydrocephalous	18	28.12%
	Anencephaly	11	17.18%
	Meningomyelocele	5	7.81%
	Occipital encephalocele	2	3.1%
Skeletal	Cleft lip	1	1.56%
	polydactyly	1	1.56%
	Lobster claw deformity	1	1.56%
	Sacro-coccygeal teratoma	1	1.56%
CVS	ASD	2	3.12%
Gastro-intestinal	Foetal ascitis	1	1.56%
	Oesophageal atresia	1	1.56%
	Gastroschisis	1	1.56%
	Duodenal atresia	1	1.56%
	Ileal atresia/Hirschsprung disease	1	1.56%
Renal	Hydronephrosis	5	7.81%

	Multicystic dysplastic kidney	1	1.56%
	B/L Polycystic kidney	1	1.56%
Respiratory	Congenital Diaphragmatic hernia	3	4.68%
Lymphatic	Cystic hygroma	1	1.56%
Soft tissue	Cystic swelling in neck	1	1.56%
Multiple	Prune-Belly syndrome	1	1.56%
	Hydrops foetalis	2	3.12%
	Down syndrome	1	1.56%
	Cranio-thoracopagus	1	1.56%

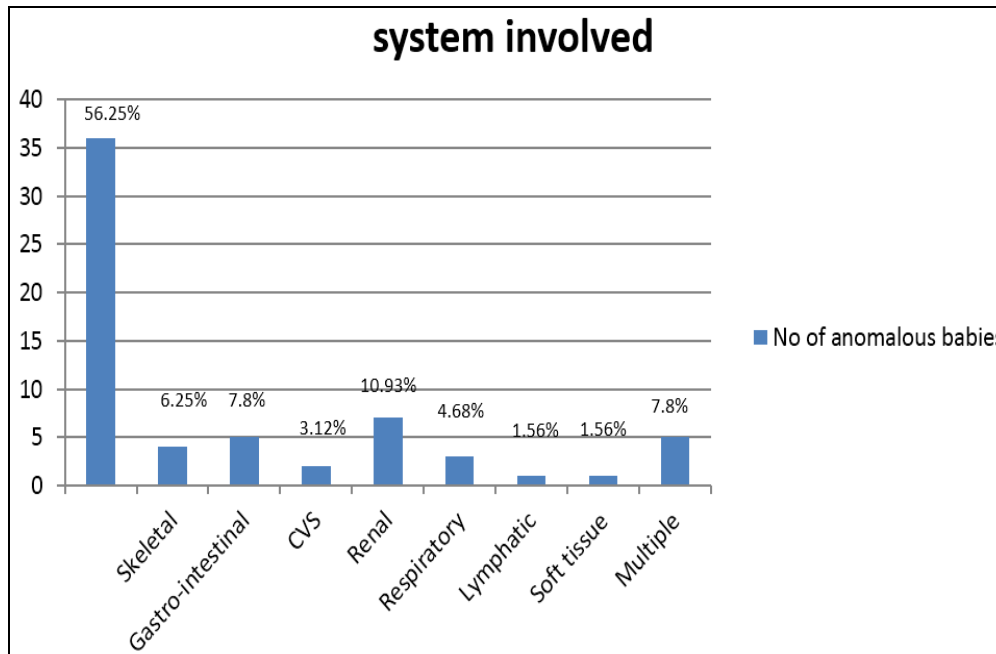


Fig 1

Discussion

Congenital malformation, one of the important cause of infant mortality and morbidity. Prevalence of congenital anomaly in our study is 1.02%.The prevalence and type of congenital anomalies are varying depending on geographical location and over different period of time. It reflects various genetic and environmental factors. This is similar to the findings of Pattanaik T *et al.* [12], 25%, Hossein Mashhadi Abdolahi *et al.* [13] 1.13% & Taksande A, 1.9% [14]. According to Shatanik Sarkar *et al.* [15] study conducted in a tertiary care hospital in Eastern India prevalence was 2.22% while Bhide P [9] reported 2.30% prevalence. the incidence of birth defects are much higher in hospitals where autopsies are performed [16, 17] Our study does not reflect the exact prevalence in the population as it was a hospital based study & in our study maximum number of women was within 21-30 yr of age. In present study only structural anomalies detected antenatal period or during deliver were included, women having first trimester abortions likely to have anomalies were not included as the maximum number of deliveries occur in the age group 20-30 years of age group, more anomalies are detected in babies born to mothers of this age group. In our study majority of antenatal women (82.81%) having anomalous foetuses were in 21-30yr age group which was same as Tapan Patnaik *et al.* [12]. There are many studies which support the increased incidence of congenital anomalies in advanced maternal age [18].

Most of the mothers in present study were primi gravida (51.56%), followed by second gravida (32.81%), third gravida (7.8%) and fourth gravida or more were (7.8%) whereas in Jayasree S *et al.* [5]. Maximum cases mothers were multigravida, followed by primigravida and then in grand multigravida. In S.

Lakshmi Vinodh [19] congenital anomaly was more in primigravida then in multigravida. In present study 93.75% babies were diagnosed during 3rd trimester& 6.25% diagnosed during 2nd trimester but first trimester abortions were not included. In Jayasree S *et al.* less than 20 weeks of pregnancy were not included and most of the case were detected in third trimester. Tapan *et al.* [12] maximum cases were detected during third trimester and during delivery. In the present study it is observed that congenital anomalies are more prevalent in male babies compared to females, 54.68% were male & 45.31% were females. This was similar to study of Jayasree S *et al.* [5] & Dutta V *et al.* [16]. It may be because of the fact that the females were affected with more lethal congenital malformations and could not survive up to an advanced gestation²⁰ In this study 6.25% mothers had history of previous baby with congenital anomaly. In Sawsan Sahib *et al.* [21] there was positive family history of congenital anomalies.

With regard to pattern of congenital anomalies in the study, the most common system involved was CNS 56.52%, followed by renal system (10.93%), gastrointestinal (7.8%), skeletal system (6.25%) respiratory (4.68%), cvs (3.12%) etc. Kalra *et al.* [22] & Pattanaik T *et al.* [12]. Reported that the CNS defects had the highest incidence, whereas Francine *et al.* [23]. Found the commonest anomalies as congenital cardiovascular disease (16.6%) and limb anomalies (16.6%). In our study hydrocephalus and anencephaly were the commonest malformation in CNS. In our study one baby of Down syndrome found, Down syndrome is the most common chromosomal malformation in newborns. Throughout the world [24], the overall prevalence of Down syndrome is 10 per 10,000live births.

Conclusion

Congenital malformation, one of the important causes of infant mortality and morbidity can be reduced by proper preconception care and antenatal anomaly scan. Even the treatment and rehabilitation of these anomalous children is a challenging task. So Regular antenatal visits and prenatal diagnosis by early trimester screening test are recommended for prevention, timely intervention and even planned termination. We found neurological defect as commonest malformation which can easily be prevented by pre-conceptional folic acid and vit-B12.

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