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Original Article

Neonatalogy Section

Prevalence and Spectrum of Congenital Malformations in a Tertiary Care Centre

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ABSTRACT

Introduction: Congenital anomaly is an abnormality of physical structure that is seen at birth or within few weeks after. These abnormalities largely contribute to neonatal and infant morbidity and mortality. The occurrence and pattern of presentation vary from region to region. The exact aetiology is not known and genetic and environmental factors may be implicated.

Aim: To determine the prevalence and types of congenital anomalies in newborns and to study the associated risk factors.

Materials and Methods: This observational hospital based study was conducted in Department of Obstetrics and Gynaecology MKCG Medical College and Hospital, Berhampur, Odisha. Total 100 cases of congenital malformation detected antenatally or after delivery were analysed within a study period of Jan-2011 to March 2012.

Result: Total 100 cases of anomalies were studied out of 7973 pregnancies. The prevalence of congenital malformation was 125 in 1000(12.5%). Major congenital malformations were 104 in 1000(10.4%) and minor malformations were 21 in 1000(2.1%). Most of the anomalous babies were born to women of fourth gravida or more (42%). Commonest system involved was central nervous system (43%). In the postpartum period maximum (56%) babies were diagnosed to have anomalies. Amniotic fluid abnormalities were the most commonly (18%) associated maternal conditions.

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

Keywords: Antenatal woman, Birth defects, Cystourethrography

INTRODUCTION

Congenital anomalies are defined as structural or functional anomalies, that occur during intrauterine life and can be identified antenatally, at birth or later in life. According to WHO congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. The aetiology of congenital abnormality may be genetic (30–40%) or environmental (5–10%) [1] It accounts for 8–18% of perinatal deaths and 10–15% of neonatal deaths in India [2]. The birth prevalence of congenital anomalies in the developing countries is actually underestimated due to lack of diagnostic techniques and their reliability [3]. Birth Defects Registry of India (BDRI) was instituted in 2001 by Foetal Care Research Foundation to document incidences of congenital abnormalities in India. Literature search reveals that India has the highest numberof children with birth defects [4]. The present study was carried out with the aim to determine the prevalence of congenital malformations, the associated risk factors and type of anomaly affecting various organ systems.

MATERIALS AND METHODS

It was a prospective observational hospital based study conducted in Department of Obstetrics and Gynaecology MKCG Medical College and Hospital, Berhampur, Odisha. After obtaining ethical committee approval, total 100 cases of congenital malformation detected antenatally or after delivery were analysed within a study period of Jan-2011 to March 2012. Those women who had diagnosed to have anomalous fetus either in antepartum period or after delivery 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. The screening for congenital malformation was done by antenatal ultrasound, blood tests, amniocentesis in selected cases. 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. CT, MRI, Cystourethrography were

performed in selected cases only. All the foetuses and babies having congenital malformation were taken into account and were analysed for the study. Data was entered into excel data sheet and appropriate statistical analysis was performed. Percentage was calculated for all the variables.

RESULT

Total 100 cases of anomaly were studied out of 7973 pregnancies. The prevalence of congenital malformation was 125 in 1000 (12.5%). Major congenital malformations were 104 in 1000(10.4%) whereas minor malformations were 21 in 1000(2.1%). Majority of antenatal women (39%) having anomalous foetus were in 21-30yr age group followed by 30-35yr (23%), >35yr (21%) and <20yr (17%) age group [Table/Fig-1]. Most of the anomalous babies were born to women of fourth gravida or more (42%), but second and third gravida were 33% and primigravidas were 25%. Among all the babies 56% were male, 41% were female and 3% babies had ambiguous genitalia. In the postpartum period 56% babies were diagnosed to have anomaly, 30% babies were diagnosed during 3rd trimester, 13% in 2nd trimester and only1% was diagnosed during 1st trimester [Table/Fig-2].Amniotic fluid abnormalities were associated with 18% cases of anomalous babies followed by consanguinity in 6%, severe preeclampsia in 4%, multiple pregnancies in 2% and uncontrolled diabetes mellitus, hypothyroidism, ante partum eclampsia, chicken pox each in 1% of babies [Table/Fig-3]. Majority were central nervous system anomaly (43%). Skeletal anomalies were 18%, multiple anomalies and abdominal wall defect each were 7% followed by renal 6%, sensory 5 %, gastrointestinal tract 4%, genital 3%, cardiovascular, neck and hydrops each 2% and respiratory anomalies were 1% [Table/Fig-4]. Among the central nervous system, hydrocephalous were 15%, anencephaly 10%, hydrocephalous with meningomyelocele 2%, meningomyelocele 6%, open spina-bifida and microcephaly each 3%, iniencephaly and Dandy Walker syndrome each were 2%. Conginital Talipes Equine Varus (CTEV) were the most frequent anomaly (9%) among skeletal system followed by syndactyly (3%) and cleft lip (3%), polydactyly, achondroplasia, and syrenomelia each were 1%. Atrial and ventricular septal defect both were found in one baby and pericardial effusion was also in one baby. Ascites was found in 6% of babies and one had congenital diaphragmatic hernia. Only one baby had tracheo-oesophageal fistula whereas, two babies had cystic hygroma. Among the sensory system orbital haemangioma, anophthalmos, epidermal nevus, congenital blister of skin, albinism each were 1%. Gastroschisis was there in 2 babies, one baby had duodenal atresia and one had oesophageal atresia. Four babies had hydronephrosis, 1 had polycystic kidney disease and pelviureteric junction obstruction was there in one baby. Only 3 babies had pseudo hermaphroditism [Table/Fig-5].

Maternal age	No of anomalous babies	Percentage
Age (n=100)		
<20	17	17%
21-30	39	39%
30-35	23	23%
>35	21	35%
Order of pregnancy (n=100)		
G 1	25	25%
G 2-3	33	33%
G 4 or more	42	42%
Sex variation		
Sex of baby	**No of malformed babies**	**Percentage**
Male	56	56%
Female	41	41%
Ambiguous	3	3%

[Table/Fig-1]: Demographic features.

Trimesters	Total cases(n=100)	Percentage (%)
1st trimester	1	1
2nd trimester	13	13
3rd trimester	30	30
After delivery	56	56

[Table/Fig-2]: Time of detection of anomaly.

Risk Factors	No of cases	Percentage (%)
Uncontrolled diabetes mellitus	1	1
Hypothyroidism	1	1
Severe PET	4	4
Consanguinity	6	6
Multifetal gestation	2	2
Ante partum eclampsia	1	1
Chicken pox	1	1
Abnormalities of AFI	18	18

[Table/Fig-3]: Maternal risk factors.

Type of abnormality	No of cases	Percentage (%)
CNS	43	43
CVS	2	2
Abdomen	7	7
Renal	6	6
Skeletal	18	18
Sensory	5	5
Multiple	7	7
Neck	2	2
Hydrops	2	2
Gastrointestinal	4	4
Genital	3	3
Respiratory	1	1

[Table/Fig-4]: Organ systems involved.

Systems Involved	Malformation	Percentage (%)
CNS (n=43)	Hydrocephalus	15
	Anencephaly	10
	Hydrocephalus and meningomyelocele	2
	Menigomyelocele	6
	Microcephaly	3
	Iniencephaly	2
	Open spina Bifida	3
	Dandy Walker Syndrome	2
Skeletal system (n=18)	Cleft lip	3
	CTEV	9
	Polydactyly	1
	Syndactyly	3
	Achondroplasia	1
	Syrenomelia	1
CVS (n=2)	ASD+VSD	1
	Pericardial effusion	1
System	**Malformation**	**Percentage**
Abdomen (n=7)	Ascites	6
	Congenital diaphragmatic hernia	1
Respiratory (n=1)	Trachea oesophageal fistula	1
Neck (n=2)	Cystic hygroma	2
Multiple (n=9)		7
Sensory (n=5)	Orbital haemangioma	1
	Anophthalmos	1
	Epidermal nevus	1
	Congenital blister of skin	1
	Albinism	1
Gastrointestinal (n=4)	Gastroschisis	2
	Duodenal atresia	1
	Oesophageal atresia	1
Renal (n=6)	Polycystic kidney disease	1
	Pelviureteric junction obstruction	1
	Hydronephrosis	4
Genital (n=3)	Pseudohermaphroditism	3

[Table/Fig-5]: Type of congenital anomalies.

DISCUSSION

Congenital malformation is a very common cause for neonatal morbidity and mortality. This study was an attempt to find out the most common system affected and to find out the causal relationship of different aetiological factors. Routine antenatal ultrasonography can detect majority of structural abnormality of foetus. Advanced diagnostic technology can also detect a large number of anomalies in neonatal period.The prevalence of congenital abnormalities was 1.25% in this study. This was similar to the findings of Amar Taksande [5]. According to Obu et al., the prevalence of congenital abnormalities was 2.8% in their study in the neonatal unit of a tertiary hospital in Enugu [6]. The incidences of birth defect are three times higher in the hospitals where autopsies are performed. Another centres reported a higher incidence of congenital malformations due to more autopsy rates [7,8]. Our study does not reflect the exact prevalence in the population as it was a hospital based study. In our study maximum number of patients was within 21-30 years of age. Regarding the order of pregnancy 42% of cases are fourth gravida or more. Dutta et al., documented statistically insignificant association of increased maternal age and congenital anomalies and in contrary in Turkey, studies show that 5.2% of the mothers are elderly (>35 years of age) [7,9]. The relationship between maternal age and babies born with congenital malformations, in our study, revealed that a majority of malformed babies were born of mothers aged 21-30 years but it was statistically insignificant. Previous studies have reported significantly higher incidence of malformation among multiparous women. Our study also consistent with this finding [5,8]. In our study total 6% cases had consanguineous marriage. Like previous studies our study also showed a higher incidence of congenital malformation in parents having consanguineous marriage [10,11]. Male: female ratio was 1.37:1 in our study. Male preponderance was similar to the other studies [7,8]. Out of 100 cases 56 cases were detected after delivery. This was due to poor rate of antenatal screening. With regards to the associated conditions along with congenital malformation most common condition associated was abnormalities of amniotic fluid volume. Other condition like diabetes, hypothyroid, severe preeclampsia was also associated with increased incidence of congenital malformation. Ordonez et al., showed positive association of diabetes mellitus, hypertension, and hypothyroidism with congenital malformation [12]. Amar Taksande also concluded similar pattern of association [5]. The annual report of Indian Council of Medical Research shows that the commonest congenital malformations are cardiac in nature (0.57%) [13]. With regard to pattern of congenital anomalies in the study, the most common system involved was CNS (43%), followed by skeletal system (18%), genitourinary (6%), cardiovascular system (2%) etc. Kalra et al., reported that the CNS defects had the highest incidence, whereas Francine et al., found the commonest anomalies as congenital cardiovascular disease (16.6%) and limb anomalies (16.6%); polydactyly (12.5%) and abnormal palmar creases (4,16%) [14,15]. Mathur et al., reported that the musculoskeletal abnormalities were the commonest [10]. In our study hydrocephalus and anencephaly were the commonest malformation in CNS. CTEV was the commonest malformation in the musculoskeletal system in our study. This was a hospital based study so the prevalence may be higher compared to population

based studies. Another limiting factor of our study was that autopsy was not routinely done due to moral and ethical issues.

CONCLUSION

This study has focused on the prevalence and types of congenital anomalies seen in our region. Congenital malformation, one of the important causes of infant mortality and morbidity can be reduced by proper preconception care and level two anomaly scan. Even the treatment and rehabilitation of these anomalous children is a challenging task. Parents are also likely to be anxious and guilt on learning of the existence of a congenital anomaly and require sensitive counseling 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. Emergence of neural tube defect as the highest category of congenital malformation indicates maternal malnutrition (especially folic acid) that needs significant attention and management.

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