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Research Article

Prevalence and Patterns of Congenital Malformations

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Abstract: Congenital anomalies have emerged as one of the leading cause of perinatal mortality and morbidity all over the world. Congenital malformations represents dysmorphogenesis occuring in early fetal life. We are in need of systematic data on the magnitude of congenital anomalies, their prevalence, and their impact on neonatal health. The aim of this study was to determine the prevalence of lethal and non lethal congenital anomalies observed during 2019 . The study was conducted in the Department of Radiology, Sree Balaji Medical College and Hospital, Chennai, from January 2019 – December 2019.

Keywords: Congenital anomalies, Malformations, Lethal and Non lethal anomalies.

INTRODUCTION

According to WHO, Congenital anomalies are defined as structural or functional anomalies, including metabolic disorders which are present at the time of birth (WHO. 2012; & National health Portal of India). Birth defects, congenital malformations, congenital abnormalities and congenital anomalies (CAs) are interchangeable terms used to describe developmental defects that are present at birth (UNICEF.). Congenital malformation represents dysmorphogenesis occurring in early fetal life. Congenital anomalies are a spectrum of disorders with prenatal origin that can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and or micronutrient deficiencies. Maternal infections such as rubella, CMV systemic illnesses like diabetes mellitus (DM), hypothyroidism and folic acid deficiency, exposure to medicinal and recreational drugs including alcohol and tobacco, certain environmental chemicals and doses of radiation are all other factors that cause birth defects (WHO.2009). The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections, whereas in developed countries they are cancer, accidents and congenital malformations. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.5,6 Congenital anomalies account for 11% of neonatal deaths globally and 9% in India. [3] The prevalence of birth anomalies in India is 6-7% (UNICEF; &WHO.2009). Patients with multiple congenital anomalies present a relatively infrequent but tremendously difficult challenge to the physician. Thus, we are in need of systematic data on the magnitude of congenital anomalies, their prevalence, and their impact on neonatal health. Prevalence studies give an idea about the pattern of occurrence of anomalies in different places, changes over a period of time and also give some clues to identify the aetiology. The present study the aim of this study was to determine the prevalence of lethal and non lethal congenital anomalies observed during the period of 2019.

MATERIALS AND METHODS:

It is an observational cross sectional study carried out at our hospital, Sree Balaji Medical College and Hospital from 1st January 2019 to 31st December 2019. This study was done, wherein all the women attending for their first antenatal checkup at our hospital Sree Balaji Medical College and Hospital are enrolled and followed till outcome. All the live neonates from newborn to 30 days of age irrespective of their general condition with Congenital malformation also comprised the study population. All the relevant informations regarding gender, weight, gestational age, mode of delivery, consanguinity, maternal age, antenatal visit record, and family history collected on a predesigned pro forma.

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OBSERVATIONS AND RESULTS:

During the study period the total number of pregnant patients who underwent ultrasound was 1095, of which the total number of congenital anomalies was 36. The prevalence rate is 3 %. The pattern of congenital malformations seen in neonates; most commonly affected musculoskeletal system 34 % followed by the central nervous system (32%), genitourinary system (30%), gastrointestinal system (8%), and syndromic (25%) [Lethal anomalies (66 .6 %), Non lethal anomalies (33.3%)]

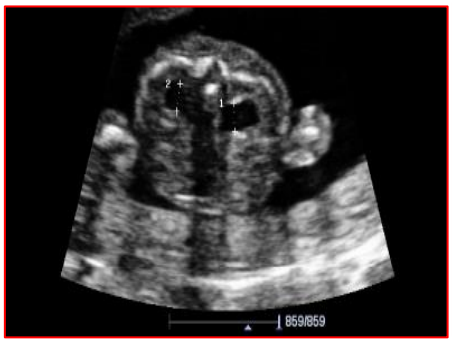
Multiparity was an important association among the risk factors studied. Among the maternal risk factors, gestational diabetes mellitus was found to be associated with birth of an anomalous baby, with nearly 24% of anomalous neonates being born to diabetic mothers . Regarding the maternal age, A higher proportion of anomalies were noted among teenage mothers and mothers less than 25 years with 5.5 % of anomalous babies being born to mothers less than 20 years and another 54.4 % to mothers between 20 to 25 years. Mothers between 26-30 years had 10 anomalous babies (27.7%) and between 31-35 years had 4 (10.8 %) babies with anomalies (Table 1). Distribution and pattern of lethal and non lethal anomalies were tabulated (Table 1).

TABLE - 1

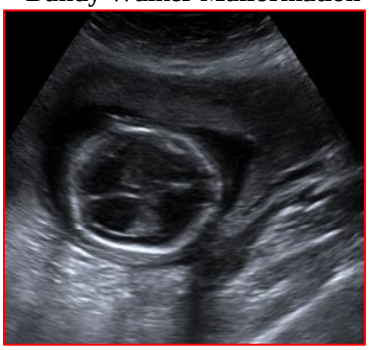
S.NO	AGE (Yrs.)	GESTATIONAL AGE IN WEEKS	CONGENITAL ANOMALIES / SYSTEM INVOLVED	CONGENITAL ANOMALIES
1	21	20 WEEKS	CNS GUT	ANENCEPHALY BILATERAL HYDRONEPHROSIS
2	30	10-11 WEEKS	CNS MSK	ANENCEPHALY CYSTIC HYGROMA CAUDAL REGRESSION SYNDROME
3	32	21-22 WEEKS	CNS SPINE	ARNOLD CHIARI MALFORMATION WITH MENINGOCELE
4	24	22 WEEKS	CNS SPINE CVS	ARNOLD CHIARI MALFORMATION WITH MENINGOCELE CARDIAC ECHOGENIC FOCUS
5	22	23-24 WEEKS	CNS SPINE	ARNOLD CHIARI MALFORMATION WITH MENINGOCELE
6	24	18-19 WEEKS	GIT RS MSK	GASTROSCHISIS NARROW THORAX ABSENT RIGHT LOWER LIMB PHOCOMELIA
7	24	21-21 WEEKS	GUT MSK	BILATERAL DILATED RENAL PELVIS CLUB FOOT
8	23	27-28 WEEKS	CNS	BILATERAL DILATED LATERAL VENTRICLES
9	24	21 -22 WEEKS	MSK	TALIPES EQUINO VARUS
10	26	21 WEEKS	CVS	EXOPHYTIC ECHOGENIC CARDIAC FOCUS
11	30	20-21 WEEKS	GUT FACE	MULTICYSTIC KIDNEY , RIGHT CLEFT PALATE AND CLEFT LIP
12	23	35-36 WEEKS	FACE	RIGHT CLEFT PALATE AND CLEFT LIP , SINGLE UMB ARTERY
13	30	20-21 WEEKS	MSK	TALIPES EQUINO VARUS IN RIGHT SIDE
14	32	35 WEEKS	MSK CVS	BILATERAL TALIPES EQUINUS , ECHOGENIC CARDIAC FOCUS
15	25	19 WEEKS	RS ABDOMEN	CHAOS CPAM ASCITES
16	24	20 WEEKS	RS ABDOMEN	CHAOS CPAM ASCITES
17	20	19 WEEKS	RS ABDOMEN	CPAM CONG.LOBAR EMPHYSEMA MESENTRIC CYST
18	30	35 WEEKS	GUT	MULTICYSTIC KIDNEYS

19	23	12 WEEKS	CNS	ACRANIA EXCENCEPHALY
20	24	18 WEEKS	GIT RS	GASTROSCHISIS NARROW THORAX
21	23	19 WEEKS	GUT	PELVIC KIDNEY
22	27	38 WEEKS	GUT	BILATERAL PUJ NARROWING
23	23	18 -19 WEEKS	CNS SPINE CVS CNS	ARNOLD CHIARI MALFORMATION WITH MENINGOCELE CARDIAC ECHOGENIC FOCUS
24	22	13 -14 WEEKS	GUT MSK	MECKEL GRUBER SYNDROME
25	28	23 WEEKS	GUT	BILATERAL DILATED RENAL PELVES
26	25	24 WEEKS	GUT CNS	MULTICYSTIC DYSPLASTIC KIDNEYS HOLOPROSENCEPHALY
27	22	20-21 WEEKS	SPINE MSK	ABSENT SACRUM BILATERAL CLUB FOOT AND HAND
28	28	20 WEEKS	CNS	VENTRICULOMEGALY
29	20	31 WEEKS	CNS	SCHIZENCEPHALY
30	25	21-22 WEEKS	GUT	POSTERIOR URETHRAL VALVE
31	22	22 -23 WEEKS	MSK	SINGLE UMBILICAL ARTERY
32	32	35 WEEKS	MSK CVS	TALIPES EQUINUS IN RIGHT SIDE TALIPES EQUINO VARUS IN LEFT SIDE ECHOGENIC CARDIAC FOCUS
33	27	15-16 WEEKS	MSK	ACHONDROGENESIS ASPHYXIATING THORACIC DYSTROPHY
34	21	21-22 WEEKS	CVS	SINGLE ARTERIAL TRUNK TRUNCUS ARTERIOSUS
35.	26	21 WEEKS	MSK	CLUB FOOT TALIPES EQUINO VARUS
36	32	13 WEEKS	MSK	CYSTIC HYGROMA

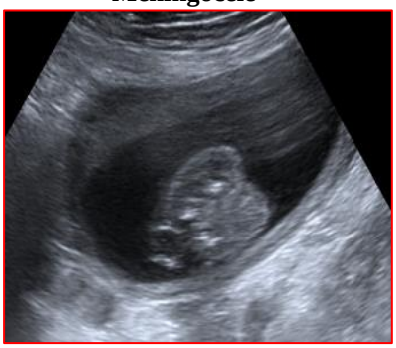
Dilated Renal Pelvis



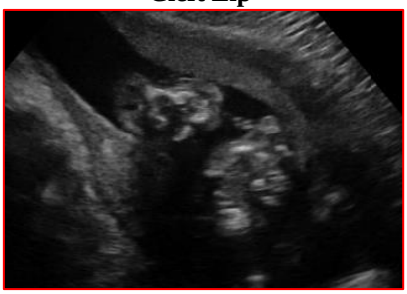
Dandy Walker Malformation



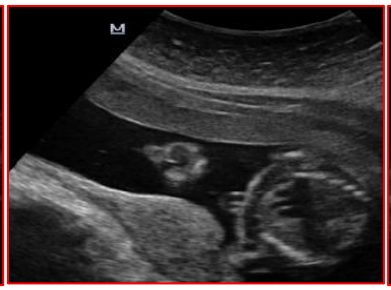
Meningocele



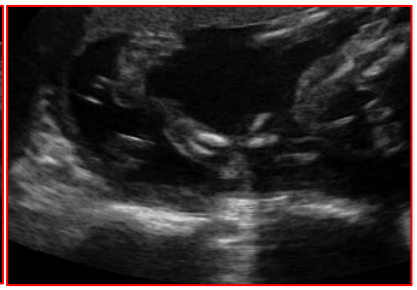
Cleft Lip



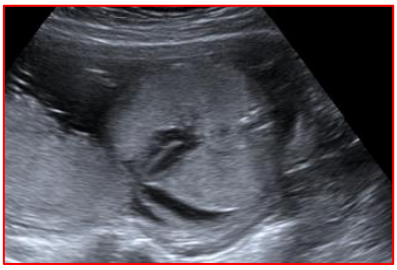
Cleft Palate



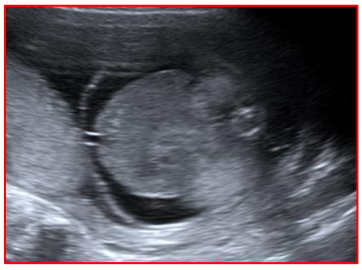
Cleft Foot



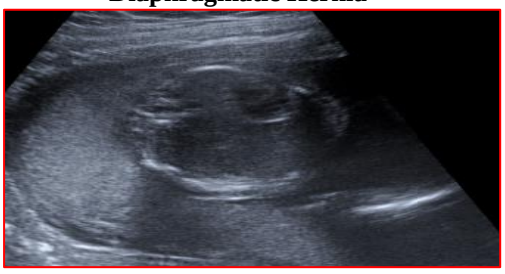
CCAM



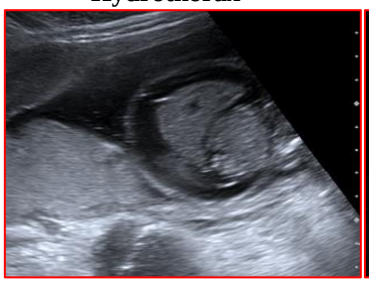
Fetal Ascites



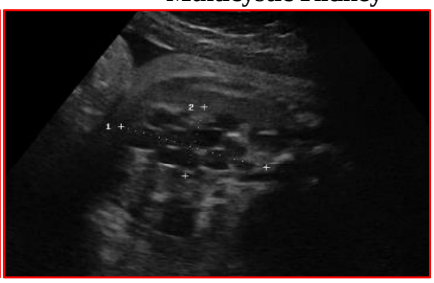
Diaphragmatic Hernia



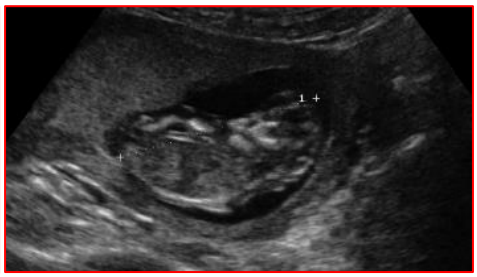
Hydrothorax



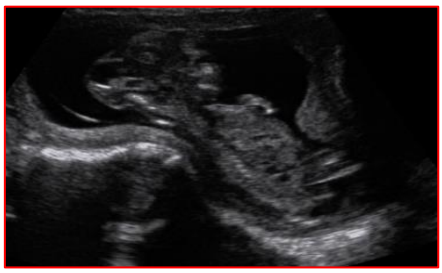
Multicystic Kidney



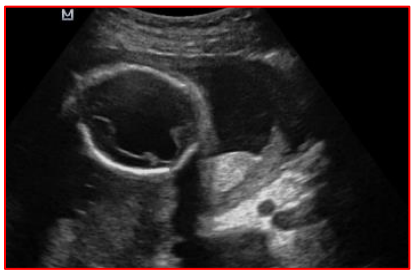
Acrania



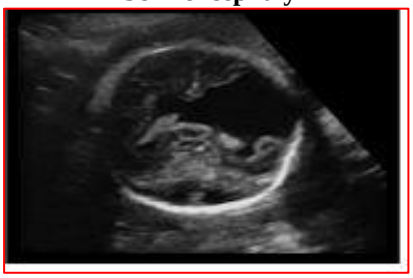
Encephalocele



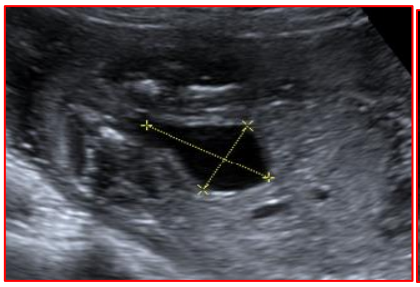
Holoprosencephaly



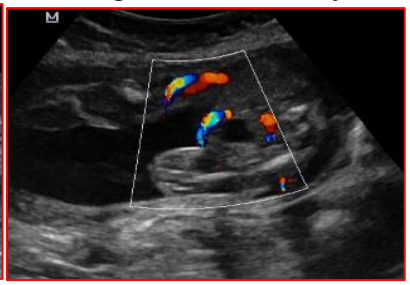
Schizencephaly



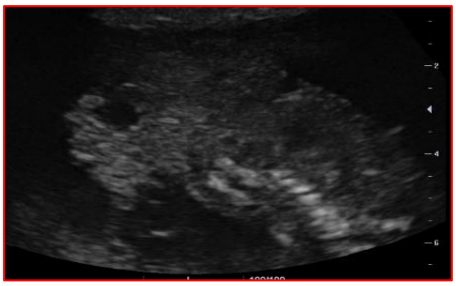
Posterior Urethral Valve



Single Umbilical Artery



Gastroschisis



DISCUSSION

There are several factors that determine the incidence, pattern and prevalence of congenital malformations. Genetic, ethnic and racial background are the key factors and the other factors include socio economic, cultural and environmental factors and their interaction with the genetic component which determines the occurrence of an anomaly.

The incidence of anomalies detected in our study was 3% which is slightly comparable to studies done by Swain *et al.,* Baht BV *et al.,* in South India (3.7%), Jehangir *et al.,*,, Chathurvedi *et al.,* and Bhide *et al.,* (Chaturvedi, P., & Banerjee, K. S. 1989; Jehangir, W. *et al.,* 2009; Bhide, P. *et al.,* 2016).The present study results are also compared to the study by Dolk H *et al.,* in Europe (2.39%) and also with the western data from the EUROCAT surveillance and found to be higher prevalence (Swain, S. *et al.,* 1994; Bhat, B. V., & Babu, L. 1998; Dolk, H. *et al.,* 2010).

Several factors like the study population, duration of the study, period and place where the study is conducted determines the incidence to a great extent. Therefore comparison of incidence with the present study is relatively difficult. We had a slightly higher incidence compared to previous studies done in the Indian settings. This could be possibly explained by the fact that the study was conducted in a referral hospital which caters high risk pregnancies with higher percentage of consanguinity, low socioeconomic class, nutritional and maternal problems.

There are plenty of studies to support the increased incidence of congenital anomalies in advanced maternal age (Prajapati, V.J. *et al.,* 2015; Swain, S. *et al.,* 1994; Savaskar, S.V. *et al.,* 2014; & Parmar, A. *et al.,* 2010). But as the maximum number of deliveries occur in the age group 20-30 years, more anomalies were detected in babies born to mothers of this age group. In our study 76.8% of the anomalies were from this maternal age group.

Swain, Savaskar and Padma observed that congenital anomalies were more in multigravidae than in primigravidae (Swain, S. *et al.,* 1994; Savaskar, S. V. *et al.,* 2014; & Padma, S. *et al.,* 2011). It was significantly seen to be higher in mothers of gravidity 4 or more (Mohanty, C. *et al.,* 1989; & Kulshreshtha, R. *et al.,* 1982). This study showed that 52.9% of the anomalies were in multigravidae. Congenital malformations are usually associated with low birth weight. Studies by Prajapati, Patel and Aman Taskade showed a significantly higher incidence of anomalies in preterm babies than term babies (Patel, Z. M., & Adhia, R. A. 2005; Taksande, A. *et al.,* 2010; & Prajapati, V.J. *et al.,* 2015). In present study 30.8% of the babies were born before 37 weeks of gestation. 47.9% of them were below 2.5kg.

The risk of congenital anomalies (excluding terminations) for gestational diabetes is 1.2 times higher than in the total population (Sharpe, P. B. *et al.,* 2005).

Statistically significant association was found between congenital malformation and consanguineous marriage. Agarwal SS and Desai N *et al.,* found highly significant correlation between congenital malformation and consanguinity (Agarwal, S. S. *et al.,* 1991; & Desai, N., & Desai, A. 2006). In our study statistically significant association was found between congenital malformation and previous child with malformation. Similar findings was also obtained in the study of Agrawal *et al.,* while Anand *et al.,* and Sagunabai *et al.,* did found such significant association between congenital malformation and previous child with malformation (Agarwal, S. S. *et al.,* 1991; & Anand, J. S. *et al.,* 1988).

In our study the pattern of congenital malformations seen in neonates are as follows; most commonly affected musculoskeletal system 34 % followed by the central nervous system (32%), genitourinary system (30%), gastrointestinal system (8%), and syndromic (25%).

Among the musculoskeletal anomalies, Congenital talipes equino varus was the commonest musculocutaneous abnormality observed in our study, followed by phocomelia, caudal regression, cleft lip, and cleft palate etc. (Table 1)

With reference to the central nervous system anomalies, anencephaly (most common) followed by Arnold chiari malformations, ventriculomegaly, meningocele, holoprosencephaly, schizencephaly etc.

Regarding the genito urinary anomalies, multicystic dysplastic kidneys, polycystic kidneys, pelvicalyceal system dilatations of all sorts like pelvi ureteric junction narrowing, low placed kidney, posterior urethral valve are seen.

In case of gastrointestinal system anomalies, two cases gastroschisis and mesenteric cyst are seen.

In cardiac anomalies, echogenic focus, cardiac myxoma and single arterial trunk -truncus arteriosus seen.

In respiratory system malformations we have detected congenital airway malformation (CPAM OR CCAM) and congenital lobar emphysema.

With reference to the syndromes that we encountered are the Arnold chiari malformations, Dandy walker malformation, Meckel gruber syndrome, caudal regression syndrome, prune belly syndrome, posterior urethral valve, Greenberg syndrome, phocomelias etc.

CONCLUSIONS:

In developing countries like India the important cause for the perinatal mortality and morbidity are the congenital malformations. Routine Antenatal surveillance and prenatal diagnosis are recommended to detect all the CNS, MSK, GIT, AND GUT anomalies for effective prevention, early intervention and planned termination and appropriate treatment planning..

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