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Study on the Spectrum of Congential Anamoly of Fetus in a Tertiary Care Centre in South India

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Abstract
Background: *According to WHO, Congenital anomalies are structural/ functional anomalies which includes metabolic disorders that are present during the time of birth. In India, those anomalies are a main health problem reckoning for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths. This study was accomplished to perceive the frequency, pattern of congenital anomalies and various presentations. The knowledge gained in this study may guide to evolve strategies for patient consulting and management. The objective of study was to present the spectrum of various congenital anomalies, epidemiological features of pregnant women with anomalous fetus.*
Methods: *Retrospective, analytical hospital based study of 102 antenatal patients with fetal congenital anomaly for a period of 4 months (October, November, December 2018 & January 2019). The appropriate information concerning maternal age, parity, gestational age, birth weight, sex, and consanguinity antenatal ultrasound was recorded.*
Results: *During the study period 102 antenatal women with fetal congenital anomalies are seen, of which this 48 (47%) are terminated. 56 (55%) patients are primi, 53 (52%) patients are in the age group of 20 to 25 years. Most of them are female fetus 43 (42.1%) Central nervous system 15 (37%) was the most common system involved.*
Conclusions: *Early diagnosis, antenatal ultrasonography, proper counseling for this pregnancy and subsequent pregnancy in needed for proper management of the problem.*
Keywords: *Congenital malformations, New-born, Still born, antenatal ultrasound, Central nervous system.*

Introduction

Congenital anomalies are substantial agents of health problem leading to childhood death, chronic illness, and disability in many countries. Congenital anomalies are also referred to as birth defects, congenital disorders/ congenital malformations[1]. According to WHO fact sheets, an estimated 3,03,000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies[2]. Figure 1. shows the causes of 2.68 million deaths during the neonatal period in 2015, worldwide.

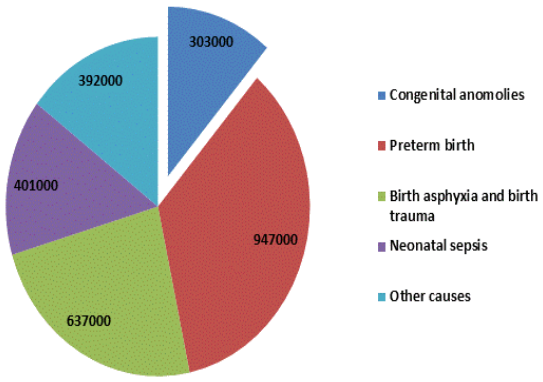

Figure 1. Causes of 2.68 million deaths during the neonatal period in 2015, worldwide (Source adapted from WHO 2000-2015 child causes of death)

Congenital anomalies are structural/ functional anomalies which includes metabolic disorders that result at the time of intrauterine life and can be diagnosed prenatally, during birth/ later in life/ detected thereon in infancy, i.e., hearing defects. The genetic/ environmental factors including errors of morphogenesis, infection, epigenetic modifications on a parental germline/ a chromosomal abnormality may be the causes for the defects during birth. The consequence of the defect may rely upon complicated relationship between the prenatal deficit and the postnatal environment[3]. Also these anomalies persists in long-haul, that produces serious impingement on persons, siblings, health-care organizations, and communities. The consequence of children with congenital anomalies in developing countries is poor than in developed countries for the reason that the deficiency of proper provisions for their control. Congenital anomalies reckons for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths in India[4]. In addition, infections and nutritional deficiencies, the major causes of infant mortality are brought under control, congenital anomalies/ disorders are emerging as one of the primary worldwide problems[5,6]. The causes for 40% to 60% of congenital anomalies are of obscure etiology, 20% to 25% of anomalies are multifactorial, 10 % to 13% are because of environment and 12% to 25 % are attributed to genetic causes. Among the risk factors are advanced maternal and paternal age, consanguinity, teratogenic agents and nutritional deficiencies. Low socioeconomic status and poorantenatal care prevents early diagnosis of the malformations. Our hospital is a tertiary care medical college hospital receiving patients from nearby districts and PHC. It is important to know the frequency, pattern of congenital anomalies and various presentations. This in turn will help to develop strategies for patient counseling and management. Patient should be seen early in pregnancy and second trimester ultrasound scan should be performed. Neonatal management along with medical and surgical intervention counseling needs to be a part of the strategy. This study was aimed at presenting the spectrum of various congenital anomalies, epidemiological features of pregnant women with anomalous fetus. The current study was performed with a motivation to study the percentage of different congenital anomalies among the patients admitted over a period of October, November, December 2018 & January 2019 in our centre.

Aim and Objectives

1) To determine the frequency of different structural congenital anomalies in our hospital.
2) To identify the possible risk factors responsible for these anomalies.
3) To evaluate the fetal outcome.

Methods of Study

The study is a cross sectional analytical type and includes all mothers with ultrasound detected anomalous fetus regardless of gestational age and risk factors from the period October, November, December 2018 & January 2019. The thorough history of age, consanguinity, folic acid intake, teratogenic drug exposure, medical disorders namely diabetes, hypothyroid are taken. And an expert ultrasound is carried out at radiology department. Retrospective, analytical hospital based study of 102 antenatal patients with congenital anomalous baby from a period of october 2018 to January 2019. Relevant information regarding maternal age, parity, gestational age, birth weight, sex, and consanguinity was documented. Important antenatal history such as maternal illness, ingestion of drugs, exposure to radiation and complications of labor was documented. Whether the patient had an antenatal ultrasonography (USG) scan and findings are noted. All the aborted fetus and newborns are examined for congenital malformations soon after delivery.

Results

During the study period 102 antenatal patients with congenital anomalies of fetus are seen of which 48 (47%) are terminated as they are lethal anomaly, 10 (10%) patients are in the age group of teenage, 20 to 25yrs are 53 (52%), 26 to 30yrs are 29 (28%) >30 years are 10(10%). It is shown in table 1. 56 (55%) patients are primigravida and 46 (45%) patients are multigravida .It is shown in table 2. Majority of patients 85% had only middle school education and are from low socio economic status and rural areas. Most of structural congenital anomalies are diagnosed by antenatal ultrasound before 20 weeks of gestation and terminated in our centre. Central nervous system was the most common system involved 38 (37.2%). Anencephaly was the common malformation in CNS. Some of chromosomal anamolies (down syndrome) are found which requires additional investigations. Despite all antenatal investigations in current pregnancy some congenital anomalies are found after birth. Cardiovascular anamoly 23(22.5%) are detected after 20 weeks of gestation which are still under follow up. Multisystem involvement is seen in 6.8% of cases which are lethal and requires termination. Some of them are pentology of Cantrell, body limb wall complex, craniorachischisis totalis. Despite all antenatal investigation in current pregnancy some congenital anomalies are found only after birth. They are admitted in neonatal ward and corrective surgeries are offered for cases such as cleft lip and palate, congenital talipes equinovarus, congenital diaphragmatic hernia and bladder extrophy. It is shown in table 4. The anomalies categorized as lethal pregnancy is terminated as suggested according to the gestational age and for non lethal pregnancy, expert opinion from department of neonatology, pediatric surgery are obtained and pregnancy was carried till term. Lethal and Non-Lethal Survey shown in table 5 indicates that 48(48.2%) patients are Anomaly Terminated before 20 Weeks, (46%) patients are Delivered at Term and 38(37.2%) patients are on Follow Up.

Table 1 Maternal Age of the patients and their distribution

Maternal Age	Distribution
< 19 YRS	10 (10%)
20 – 25 YRS	53 (52%)
26 – 30 YRS	29(28%)
>30YRS	10 (10%)

Table 2 Gravida Analysis of the patients

Primigravida	56 (55%)
Multigravida	46 (45%)

Table 4 Systems involved in the study

System Involved	Number	Percentage
CNS	38	37.2
CVS	23	22.5
Renal	6	5.8
Musculoskeletal	9	8.8
Chromosomal	6	5.8
Multisystem	7	6.8
Gastro Intestinal Tract (GIT)	2	1.9
Congenital Diaphragmatic Hernia (CDH)	9	8.8

Table 5 Lethal and Non-Lethal Survey

Anomaly Terminated Before 20 Weeks	48(48.2%)
Delivered At Term	54(54.4%)

Fetal Non Immune Hydrops

Non-immune fetal hydrops is diagnosed due to the accumulation of fluid in more than one extravascular space including soft tissue edema in the skin or scalp, or fluid in body cavities such as ascites pleural effusion, pericardial effusion, or hydrocele. Non-immune fetal hydrops is easily diagnosed by antenatal ultrasound. The figure 2 shows the condition of fetal non Immune Hydrops.

Figure 2. Fetal Non Immune Hydrops

Occipitoencephalocele

Occipitoencephalocele is a congenital malformation portaid by protrusion of meninges and/or brain tissue due to a skull defect (higher incidence) or due to etiology, pathogenesis and environmental factors[7,8]. It is one form of neural tube defects as the other two, anencephaly and spina bifida[1]. The figure 3 shows the condition of fetal Occipitoencephalocele.

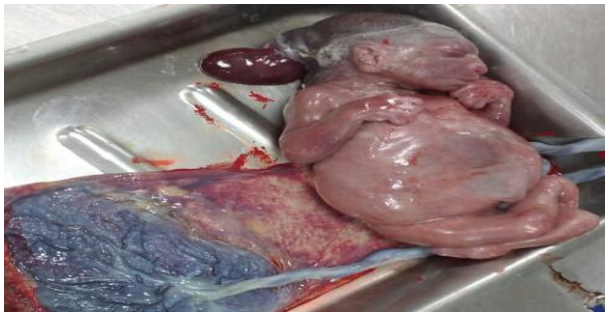

Figure 3. Occipitoencephalocele

Multiple Congenital Anomaly

Congenital anomalies are structural/ functional anomalies which includes metabolic disorders that are present during the time of birth. These disorders are of prenatal origin that results from defective embryogenesis/ intrinsic abnormalities in the development process[9,10]. In most of the cases, the origin of congenital anomalies are unclear[11,12]. However, several factors are associated in these disorders such as genetic factors, maternal infections like rubella, cytomegalovirus, toxoplasmosis and syphilis, drugs (thalidomide, streptomycin, tetracycline, phenytoin), smoking, irradiation, maternal age, health, geographical factors and dietary factors. The figure 3 shows the condition of multiple congenital anomaly.

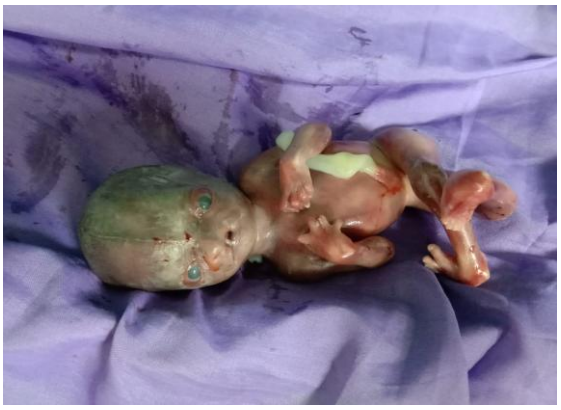

Figure 4. Multiple Congenital Anomaly

Discussion

The occurrence of congenital anomalies are more in primi gravida 56 (55%) and in the age group of 20 to 25 years 53 (52%) patients which is different than other studies where higher incidence was noted in babies born to mothers with age above 30 years. Both these results could be explained on the basis of the number of patients from rural area was 87%, belonging to low socioeconomic status.

The prenatal ultrasound at 18 to 20 weeks can detect major structural anomalies in approximately 60% of cases. Prenatal diagnosis of congenital anomalies provide information for decision on pregnancy and labour management. In our study 48% patients had anomalies diagnosed on antenatal ultrasound before 20 week and are terminated.

In our study 37.2% of cases involved central nervous system. Multisystem anomaly are also presented in our study. The low pervasiveness of cardiovascular irregularity at birth is by cause of the most congenital heart defects (CHDs) turn into symptomatic by 2 months to 4 months of age. In chromosomal anomalies Down's syndrome 15% was most frequently seen. Congenital talipes equinovarus was the commonest musculocutaneous abnormality observed. Table 6 shows the anomalies diagnosed during the period of study.

Table 6 Anomalies Diagnosed

Anomaly	Number	Percentage
Diagnosed at late gestational age	23	22.5%
Central Nervous System (CNS)	9	39%
cardiovascular System (CVS)	5	21.7%
Congenital Diaphragmatic Hernia	6	26%
GIT	1	0.04
Musculoskeletal system	1	0.04
Renal	1	0.04

In our study, out of 102 pregnant women with malformation, 23 (22.5%) are diagnosed after 24 weeks. Condition which tend to be diagnosed late in gestational like hydrocephalus, corpus callosal agenesis, various cardiac structural malformation like TOF, hypoplastic left heart syndrome, duodenal atresia, horse shoe kidney.

Out of 102 congenital anomalies, 32 (31.2%) are from consanguineous marriage, in that 20 from 3rd degree, 12 from 2nd degree consanguineous marriage. Consanguinity may plays an important role in high rates of major malformation and must be taken into account for genetic counselling [13,14].

Conclusions

This study is accomplished to evaluate the incidence of structural congenital anomalies and to predict the attributes which contribute in the incidence of congenital anomalies so that the related perinatal morbidity and mortality can be reduced. Early detection and termination of congenital anomaly will reduce the birth of babies with congenital anomalies. It will also ease the economic burden, psychological trauma to the parents and family. Collaboration between Obstetrician, Pediatricians, Geneticist and Sonologist is required for management of viable congenital anomalies. Increasing awareness of maternal care, use of Folic acid, early diagnosis, antenatal ultrasonography, proper counselling for this pregnancy and subsequent pregnancy can take care of the couple to face this dreaded complication of pregnancy

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