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Evaluation of the Incidence of Congenital Fetal Malformations by Sonography In The Second Trimester.

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ABSTRACT

OBJECTIVE: *TO EVALUATE THE INCIDENCE OF CONGENITAL FETAL MALFORMATIONS BY SONOGRAPHY IN THE SECOND TRIMESTER.*

ABSTRACT: *An early detection of fetal anomalies has become an important part of antenatal care, by helping in identifying the severity of disease and its impact, leading to either pregnancy termination or providing an opportunity for fetal therapy. This is a prospective follow-up study involving 3090 singleton pregnant women who were referred to the Department of Radiodiagnosis of Navodaya Medical College, Raichur for a complete second trimester antenatal ultrasound examination.*

MATERIAL AND METHODS: *The study included a total of 3090 singleton pregnant women, between the ages of 17 and 38, who were subjected to a second trimester complete antenatal ultrasound examination using gray scale & color duplex imaging. Complete information about the gestational age, placental location, fetal biometry & fetal anomalies (if any) was collected and tabulated. Antenatal sonographic findings were correlated with the pregnancy outcome in terms of normal fetus or fetus with malformations. This data was compared & appropriate statistical analysis was performed.*

RESULTS: *A total of 61 anomalies were detected in 51 fetuses. Prevalence of congenital malformations was 1.97%. CNS and renal abnormalities were the most common anomalies to be detected. The detection rate of facial, skeletal and cardiac defects was however less.*

CONCLUSION: *Ultrasonography provides an excellent tool for screening of congenital fetal anomalies. The priority goal in screening is early detection of major fetal anomalies. Routine anomaly screening improves perinatal outcome, especially in high risk cases, directly through termination of pregnancy for certain anomalies. Sonography has the advantage of being non-invasive, safe, fast, accurate and reproducible with real time display, causing no discomfort to the patient at any time of gestation.*

KEY WORDS: *USG; ANOMALIES; PREVALENCE*

INTRODUCTION

All pregnancies are at a risk of producing congenital malformations, though only some of them are at a greater risk. There is a need for routine and thorough screening for fetal congenital anomalies. The priority goal in screening is the early detection of major fetal anomalies, which are defined as malformations that affect fetal viability and quality of life. Congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India[1].

Ultrasonography has emerged as one of the most powerful tools for prenatal diagnosis of congenital malformations. A second trimester anomaly scan has been suggested in routine antenatal care to increase the prenatal detection rate of fetal defects[2]. Ultrasonography can identify at least 35 – 50% of major fetal malformations with a specificity of 90 – 100%[3]. Though other methods of screening like biochemical markers and karyotyping are available, ultrasonography has the advantage of being non-invasive, safe, fast, accurate and reproducible with real time display, causing no discomfort to the patient at any time of gestation.

Congenital anomalies are defined as structural defects, chromosomal abnormalities, inborn errors of metabolism and hereditary diseases diagnosed before, at, or after birth[4]. Any deviation from the normal range during morphogenesis constitutes an anomaly[5].

MATERIAL AND METHODS

The study included a total of 3090 singleton pregnant women, between the ages of 17 and 38, who were subjected to a second trimester complete antenatal ultrasound examination using gray scale & color duplex imaging. Complete information about the gestational age, placental location, fetal biometry & fetal anomalies (if any) was collected and tabulated. Antenatal sonographic findings were correlated with the pregnancy outcome in terms of normal fetus or fetus with malformations. This data was compared & appropriate statistical analysis was performed.

Study design: A prospective study of 3090 singleton pregnant women at Navodaya Medical College, Raichur.

Duration of study: The study was conducted over a period of one and a half year from January 2015 to June 2016.

Procedure for study: All pregnant women attending the antenatal clinic during the study period had routine ultrasound screening between 18-24 weeks gestation. All scans were performed as a standard level one ultrasonography. In cases of uncertain abnormal findings, the women were reviewed by a level two scan with repeated scans (if required).

Specific components of the examination include assessment of placental location, amniotic fluid volume, fetal number, presentation, cardiac activity, biometry (BPD, HC, AC, FL), and a fetal anatomy survey.

Antenatal sonographic findings were correlated with the pregnancy outcome in terms of normal fetus or fetus with malformations. This data was compared & appropriate statistical analysis was performed.

Equipment: Toshiba Nemio and Toshiba Famio ultrasound machines were used for the Ultrasonographic examination with curvilinear transducers, using gray scale & color doppler examination.

Inclusion criteria: All singleton pregnant women who were referred to the Department of Radiodiagnosis of Navodaya Medical College, Raichur for a second trimester complete antenatal ultrasound examination.

Exclusion criteria: Multiple gestations.

RESULTS

- A total of 61 anomalies were detected in 51 fetuses. Prevalence of congenital malformations was 1.97%.
- The most commonly detected anomalies were CNS at 43%, followed by genitourinary at 21%, skeletal at 8%, congenital heart diseases at 7%, abdominal wall defects, facial anomalies & hydrops fetalis at 5% each, thoracic at 3% and GI anomalies at 2%.
- The percentage of anomalies in primipara women (58.82%) was higher than in multipara (41.18%).

DISCUSSION

Congenital anomalies are important cause of still births, infant mortality and contributors to childhood morbidity. The great potential of ultrasonography for screening for morphological abnormalities throughout all trimesters of the pregnancy[6] has meant that its use with obstetric patients is becoming a routine part of prenatal care[7].

The literature describes the greatest frequency of prenatally diagnosed congenital anomalies occurring in the central nervous system, the genital and urinary systems and the musculoskeletal system[8]. Published data also indicates lower sensitivity for malformations of the circulatory and musculoskeletal systems when compared with other systems and organs[9].

Most of the pregnant women coming to our hospital for anomaly scan were in the age group of 21-25 years. It was noted in our study that the percentage of anomalies was also more in this age group (See Table 1).

Prospective studies by Amar Taksande etal[10] and Singh etal[11] found that the percentage of anomalies in primipara women was higher than in multipara, which corresponded with the results in our study (See Table 2).

In our study, we observed that more anomalies (61%) were detected in women with consanguineous marriages whereas 39 % of detected fetal anomalies had no history of consanguinity (See Table 3).

On evaluating the amniotic fluid index, it was found in our study that most of the anomalies were associated with a normal amniotic fluid. The percentage of anomalies associated with

polyhydramnios was slightly higher than those associated with Oligohydramnios (See Table 4).

In our study, the most commonly detected anomalies were CNS at 43%, followed by genitourinary at 21%, skeletal at 8%, congenital heart diseases at 7%, abdominal wall defects, facial anomalies & hydrops fetalis at 5% each, thoracic at 3% and GI anomalies at 2% (See Tables 5 & 8).

Among CNS anomalies, neural tube defects were the most commonly detected anomalies with anencephaly being the commonest, 26% in subgroup and 11.4% overall. The second most common CNS anomaly was spina bifida & ventriculomegaly with a prevalence of 22% each in subgroup and 9.8% each overall. During our study we found that most of the cases of ventriculomegaly were associated with spina bifida, with only three cases of isolated ventriculomegaly. Further in our study we observed encephalocoele to be at 18 % in the subgroup and 8.1% overall. Among the five cases detected, three were isolated and two were associated with dandy walker variant and bilateral multicystic kidney disease. We detected one case of holoprosencephaly and microcephaly, which was associated with multicystic kidney disease (See Table 7).

Among the anomalies of the genitourinary system, in our study, the prevalence of hydronephrosis was 9.8%. Few cases which showed dilated renal pelvis during late second or third trimester appeared normal at term or postnatally, on follow up scans. The incidence of renal cystic disease was 9.8% in our study. Among them, bilateral renal cystic diseases were associated with severe oligohydramnios and incompatible with life (See Table 8).

In the Musculoskeletal system, we got a prevalence of 1.6% for Short Limb dwarfism and 3% for club foot (See Table 9).

Abdominal wall defects constituted 5% of the anomalies detected. The incidence of Omphalocele in our study was 3.2%, whereas the incidence of gastrochisis, congenital diaphragmatic hernia, duodenal atresia, congenital cystic adenomatoid lung malformation was found to be 1.6% each (See Tables 10 and 11).

Cardiac defects accounted for 7% of the anomalies detected. The prevalence of Hydrops fetalis came to be 4.9 % in our study. Among the three cases of hydrops fetalis, two presented in late second trimester and were associated with still births. Among facial anomalies, the detection rate of cleft lip was less in our study. Fetal position, oligohydramnios and maternal obesity may obscure proper visualization of fetal parts and hence considered limiting factors for a complete scan (See Table 12).

On comparing the period of gestation at which the malformations were detected, it was observed that 47 out of 61 total anomalies were detected before 22 weeks of gestation, constituting 77% of the anomalies (See Table 13).

On comparing the pregnancy outcome, 45% of the women with a detected major anomaly had opted for termination of pregnancy, which mainly included CNS anomalies and abdominal wall defects. Also, 14% of detected anomalies were associated with still births, seen in fetal hydrops, bilateral renal cystic disease and short limb dwarfism. Live births were seen in 41% of the detected anomalies which included mainly PUJ obstruction, cleft lip and club foot (See Table 14).

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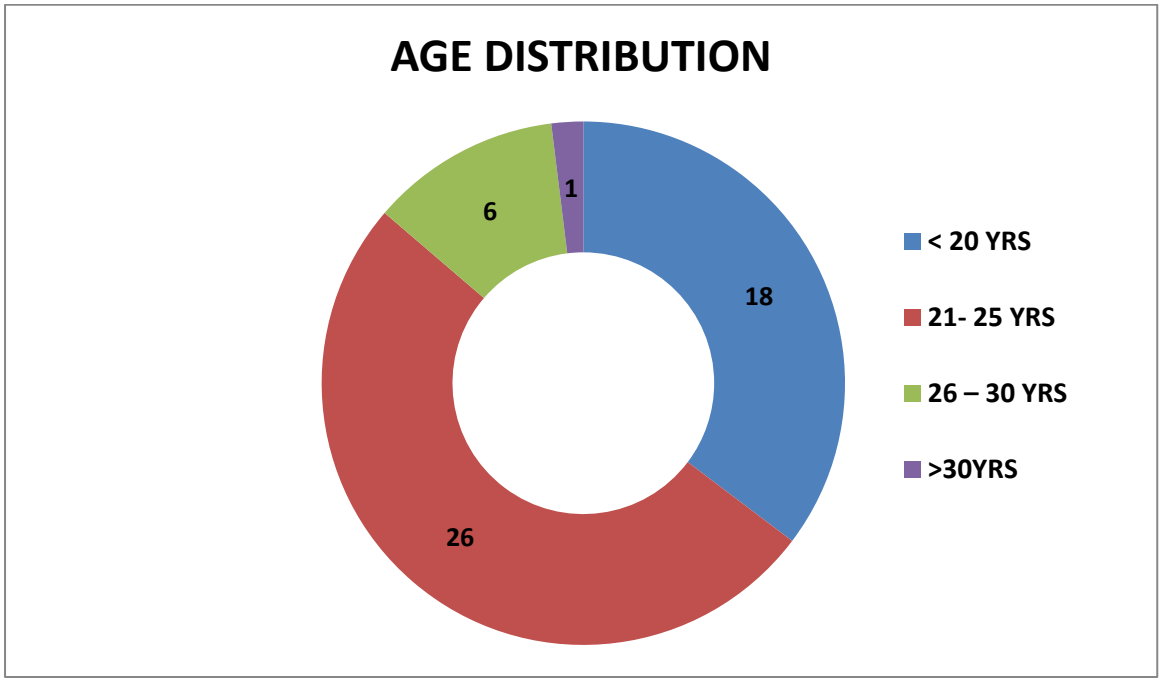

TABLE 1: AGE-WISE DISTRIBUTION OF ANTENATAL WOMEN WITH ANOMALOUS FETUS DETECTED ON ULTRASONOGRAPHY.

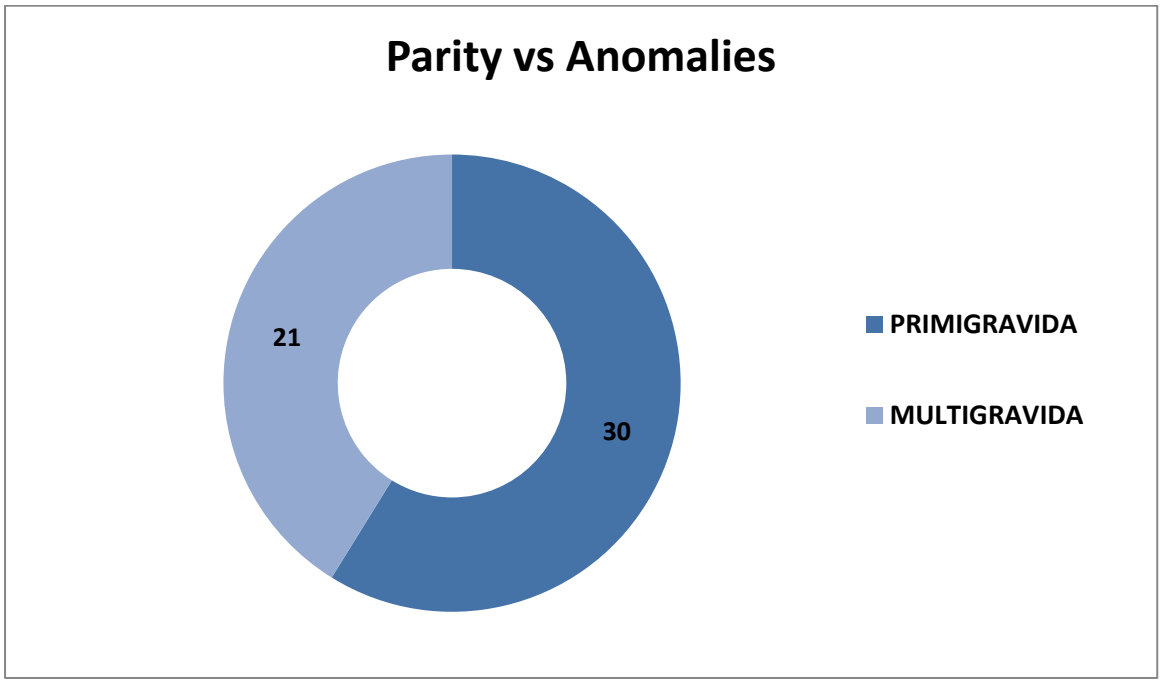

TABLE 2: DISTRIBUTION OF ANTENATAL WOMEN WITH ANOMALOUS FETUS ACCORDING TO PARITY.

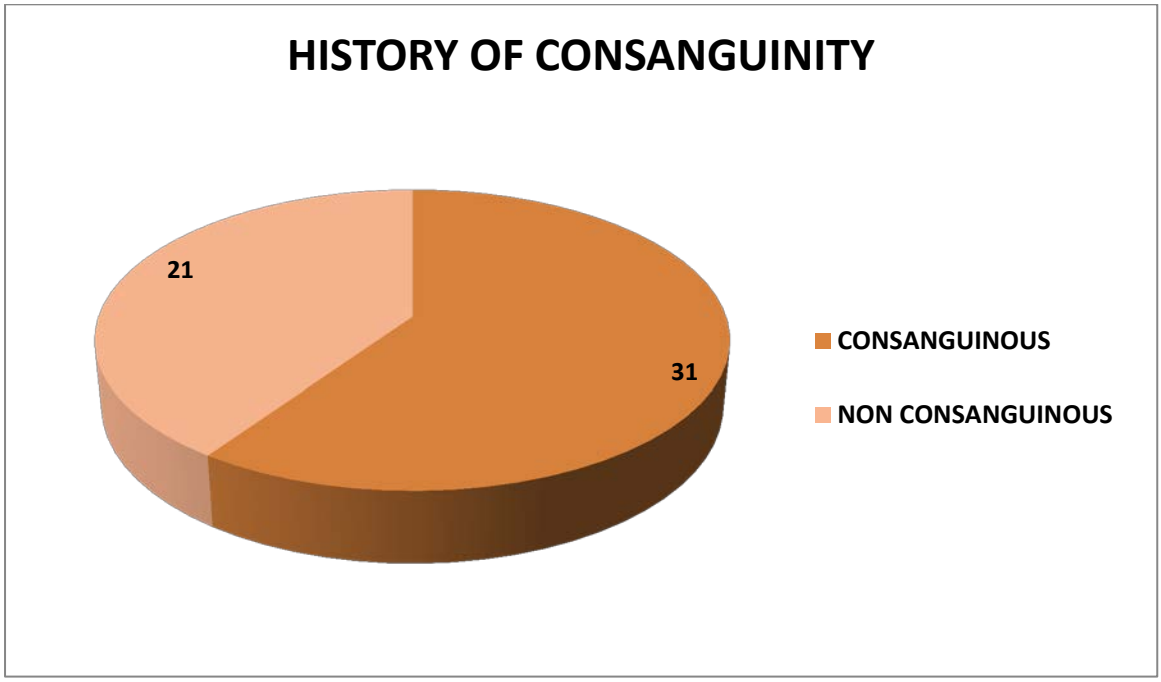

TABLE 3: DISTRIBUTION ACCORDING TO HISTORY OF CONSANGUINITY.

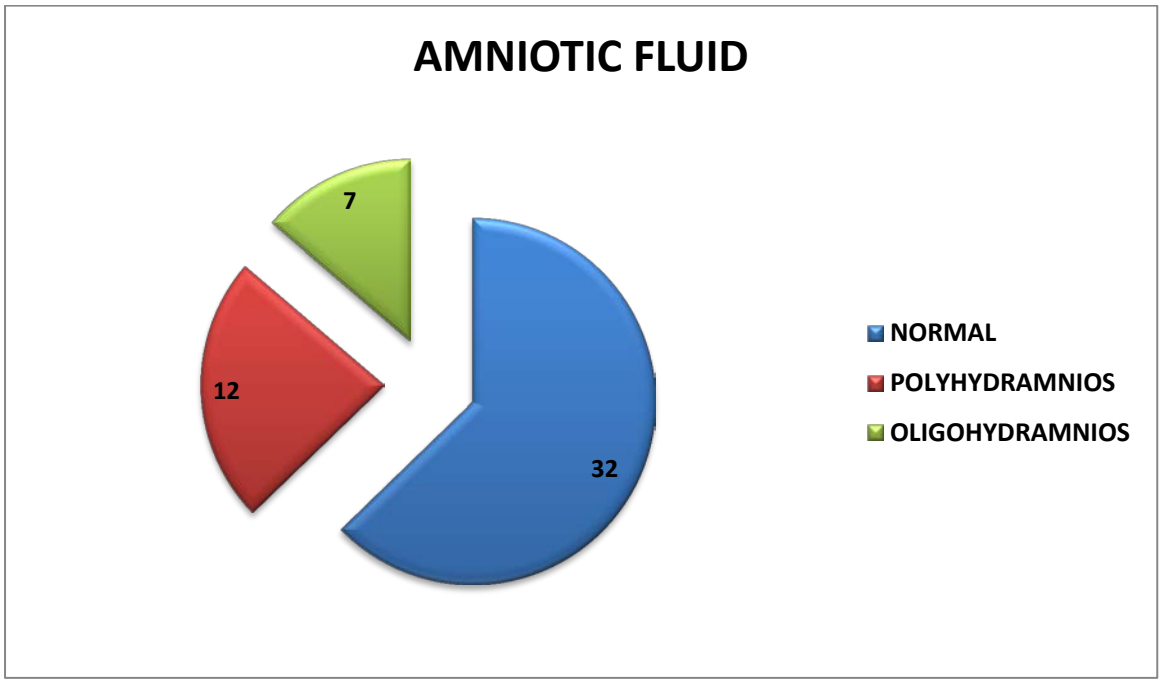

TABLE 4: DISTRIBUTION ACCORDING TO AMNIOTIC FLUID VOLUME.

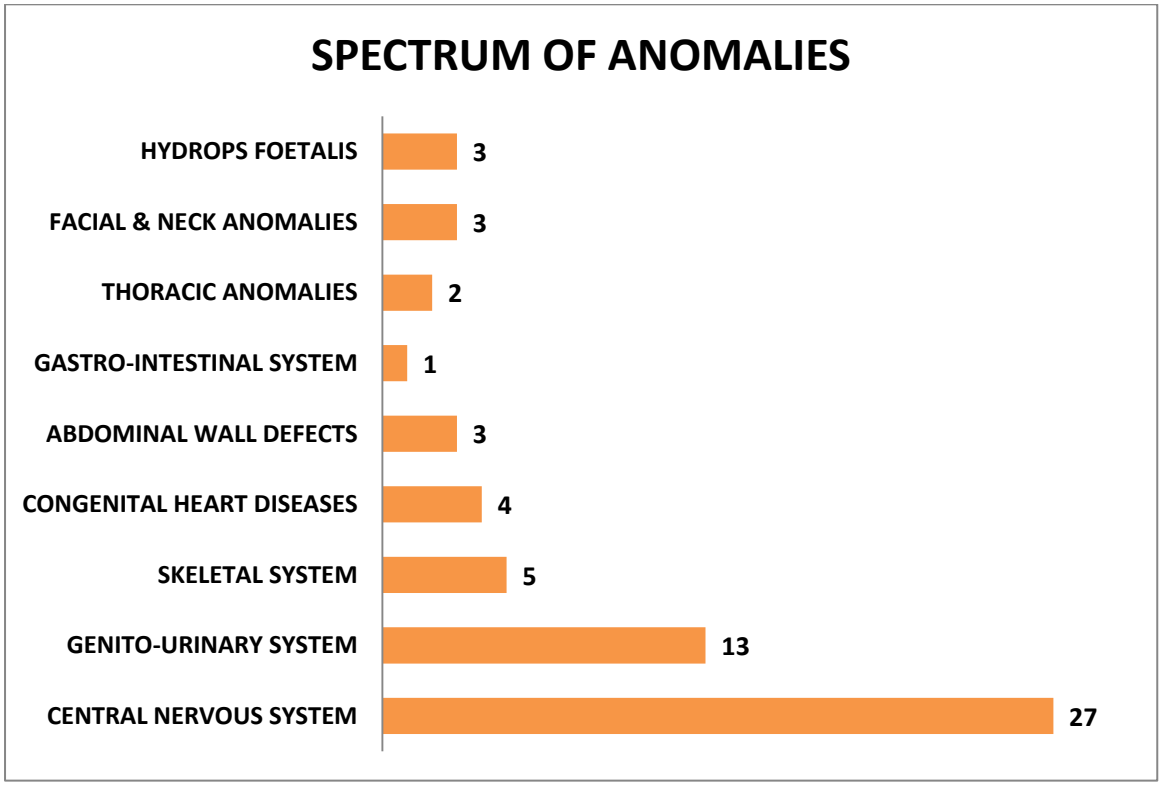

TABLE 5: SPECTRUM OF VARIOUS ANOMALIES DETECTED DURING THE STUDY.

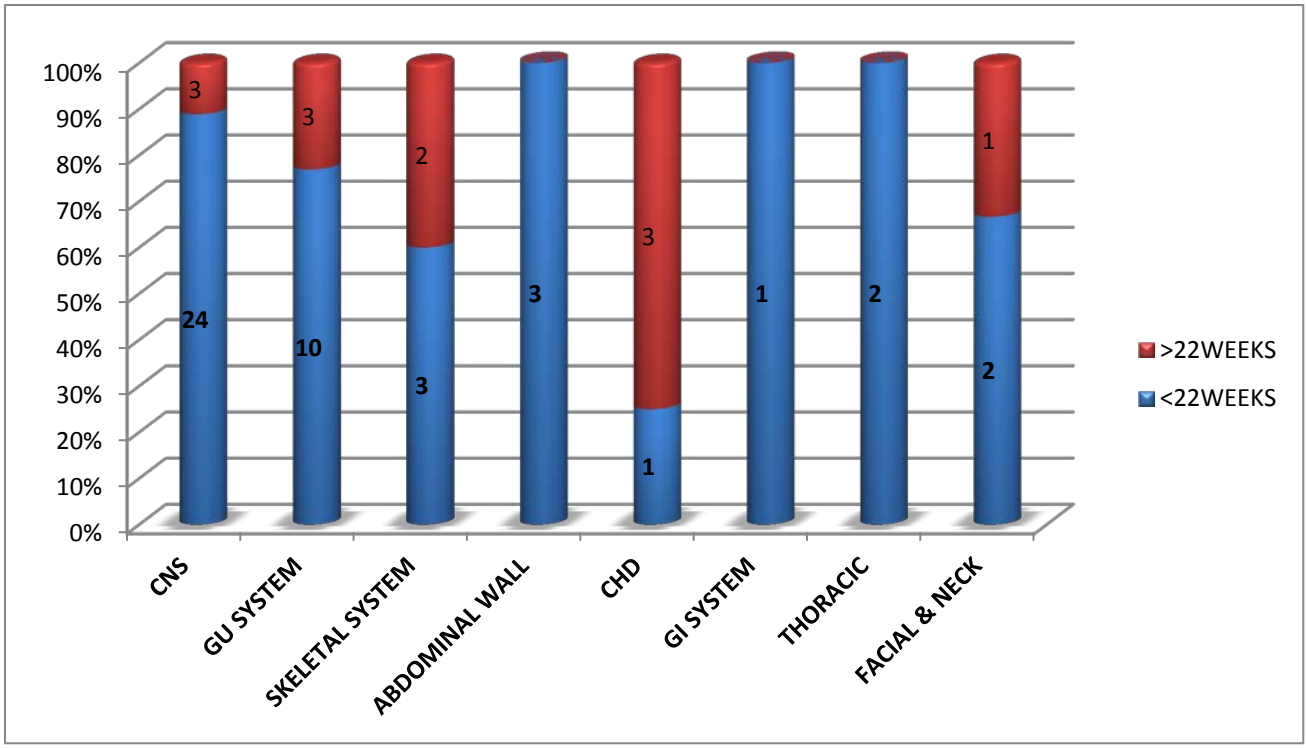

TABLE 6: SUMMARY OF THE GESTATIONAL AGE -WISE DISTRIBUTION OF ANOMALIES.

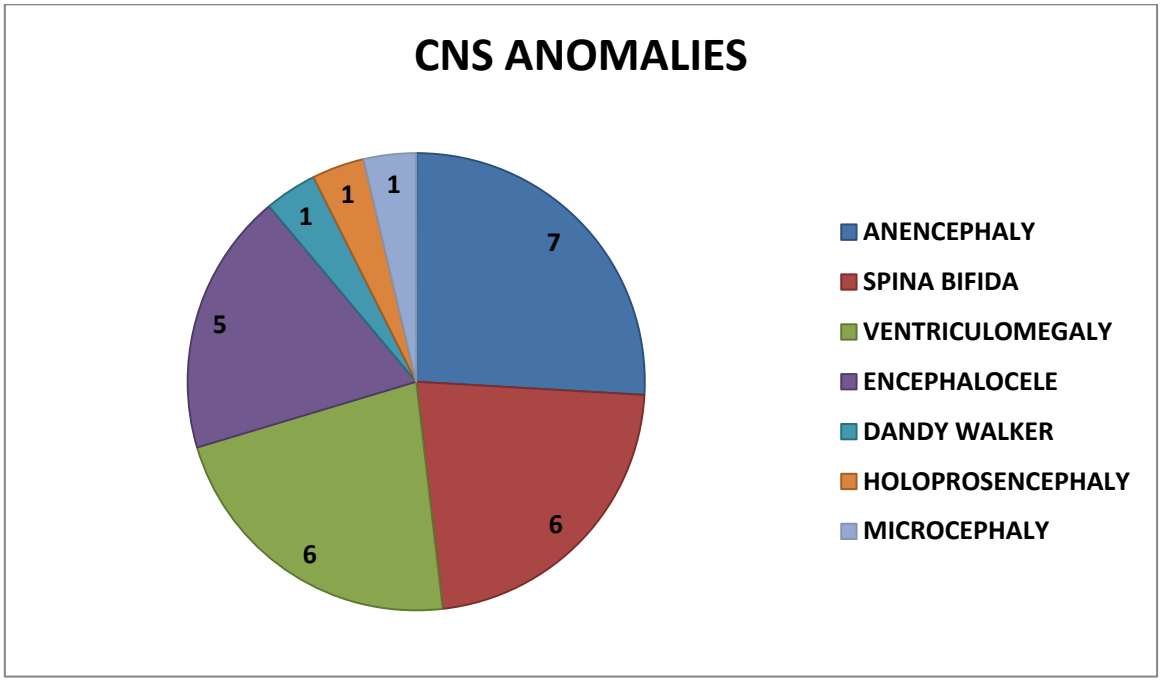

TABLE 7: INCIDENCE OF CENTRAL NERVOUS SYSTEM ANOMALIES

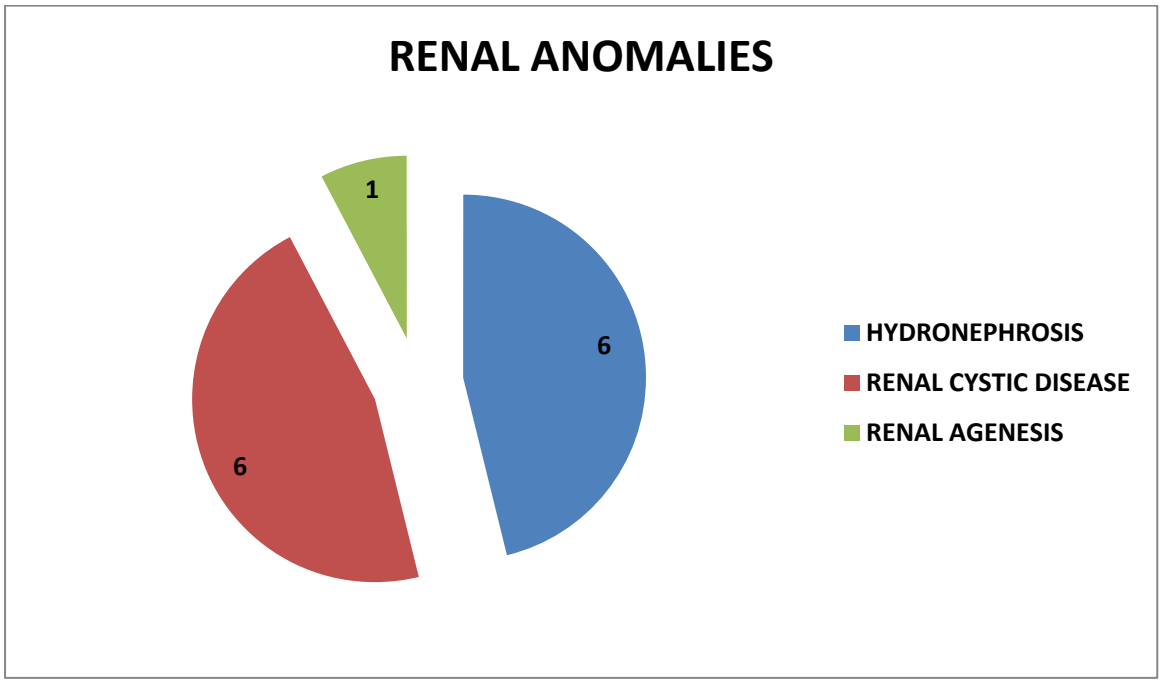

TABLE 8: INCIDENCE OF GENITOURINARY ANOMALIES

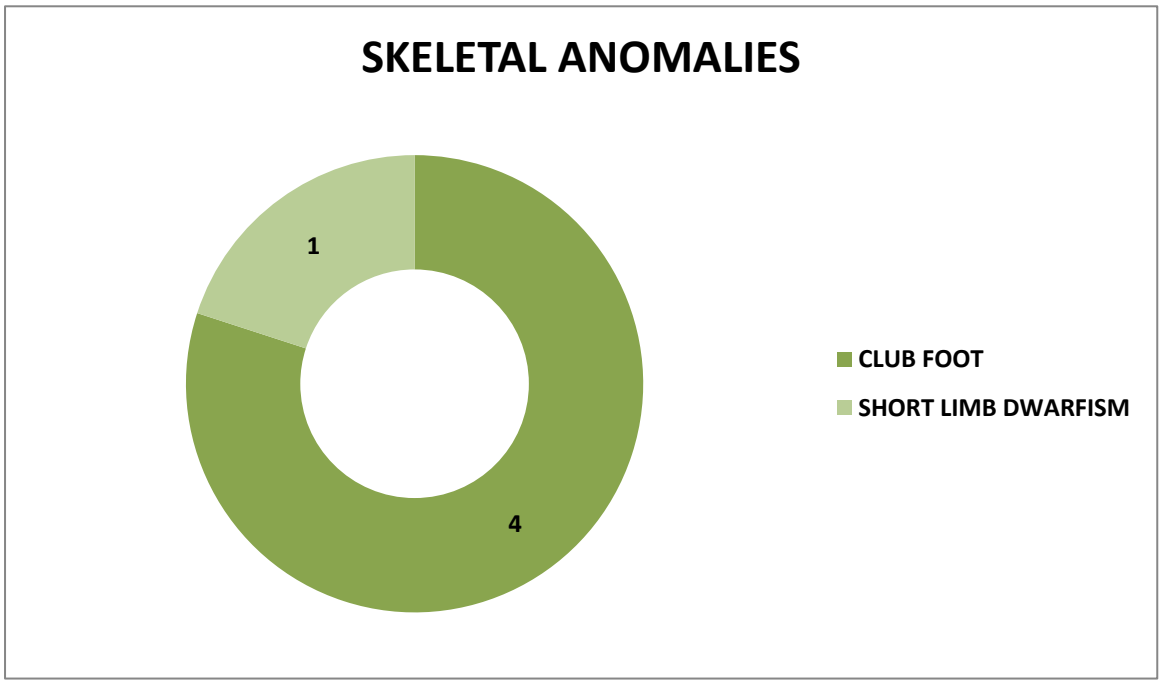

TABLE 9: INCIDENCE OF SKELETAL ANOMALIES

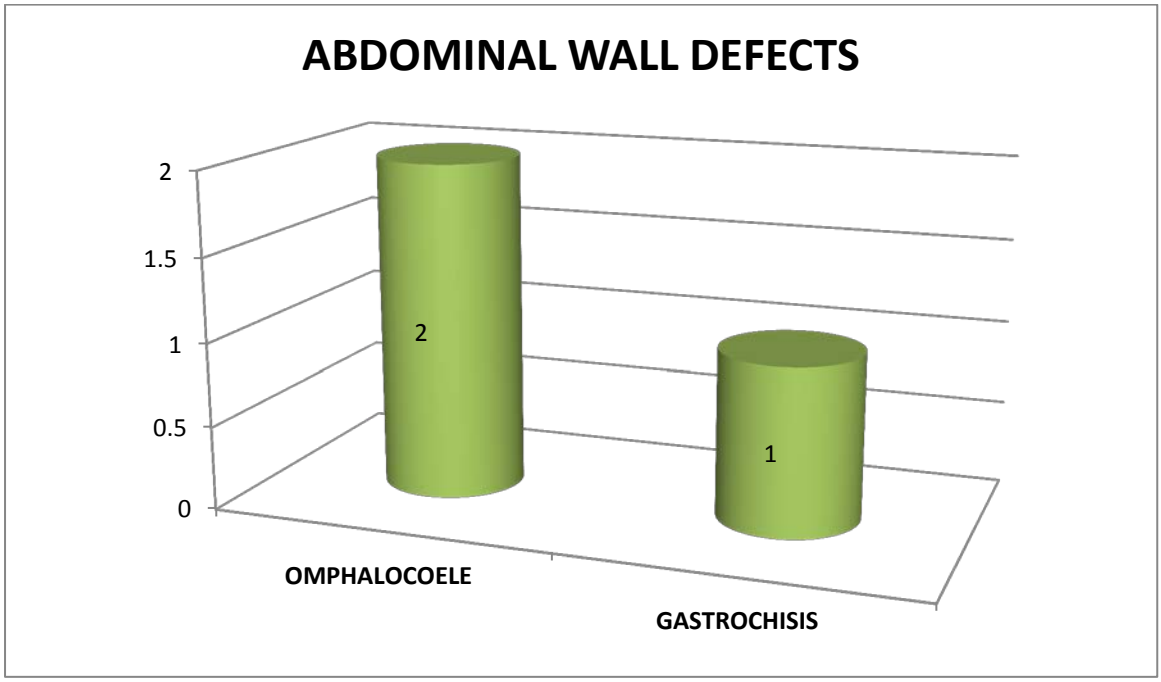

TABLE 10: INCIDENCE OF ABDOMINAL WALL DEFECTS

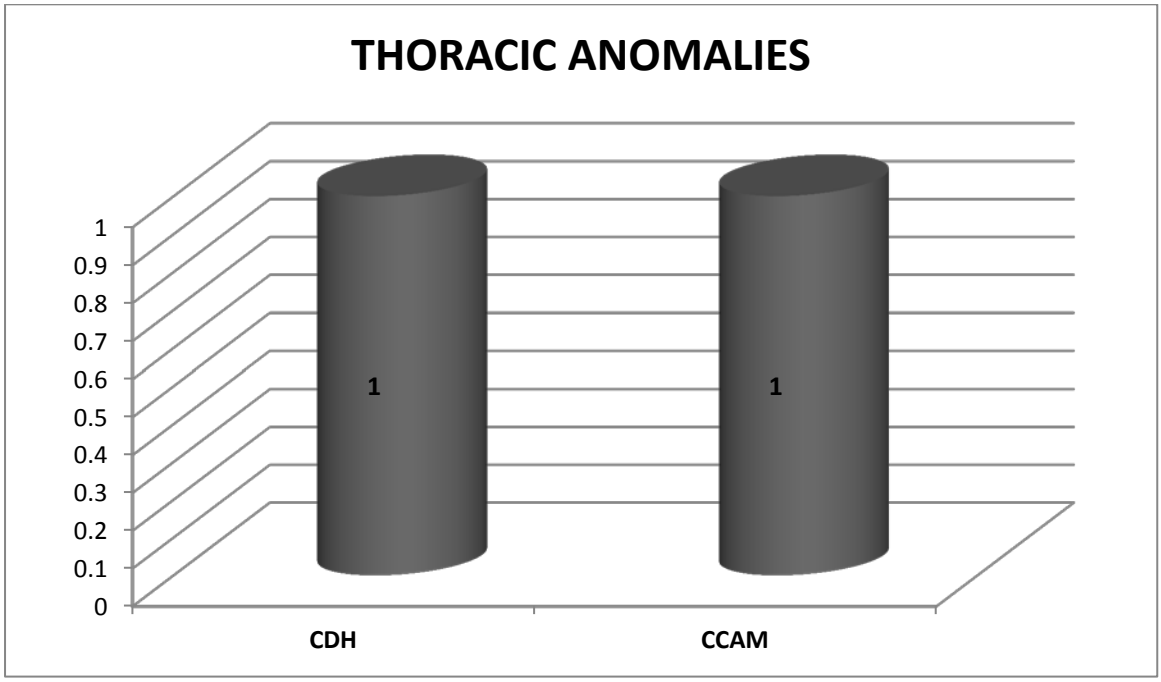

TABLE 11: INCIDENCE OF THORACIC ANOMALIES

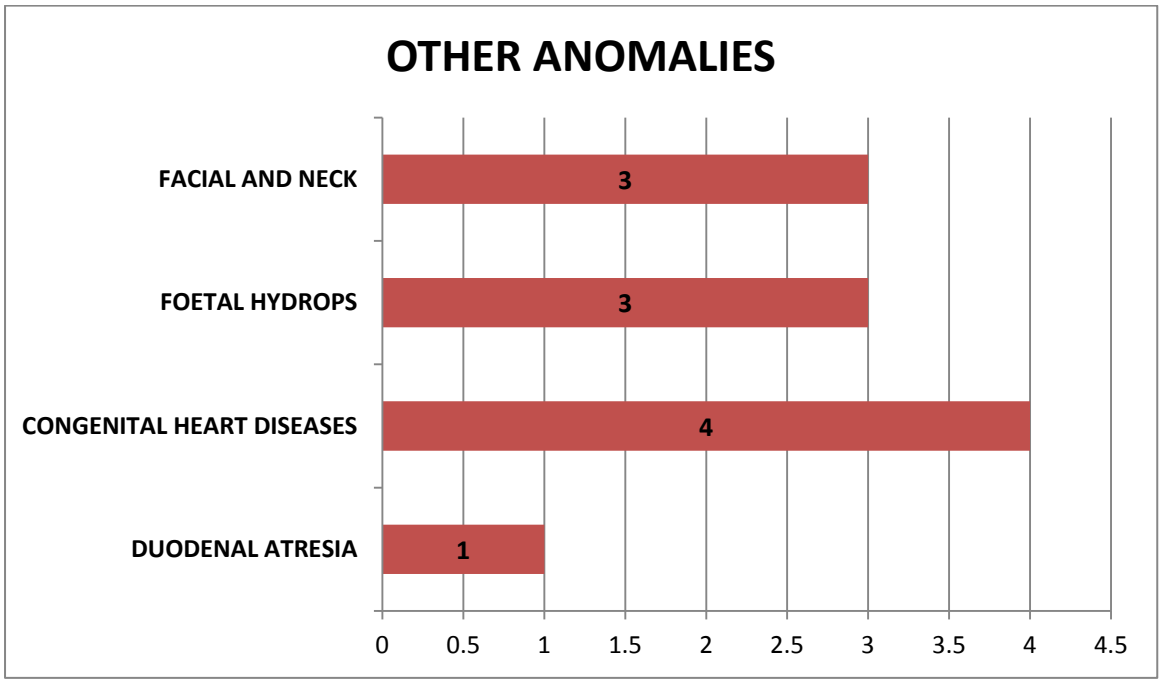

TABLE 12: INCIDENCE OF OTHER ANOMALIES

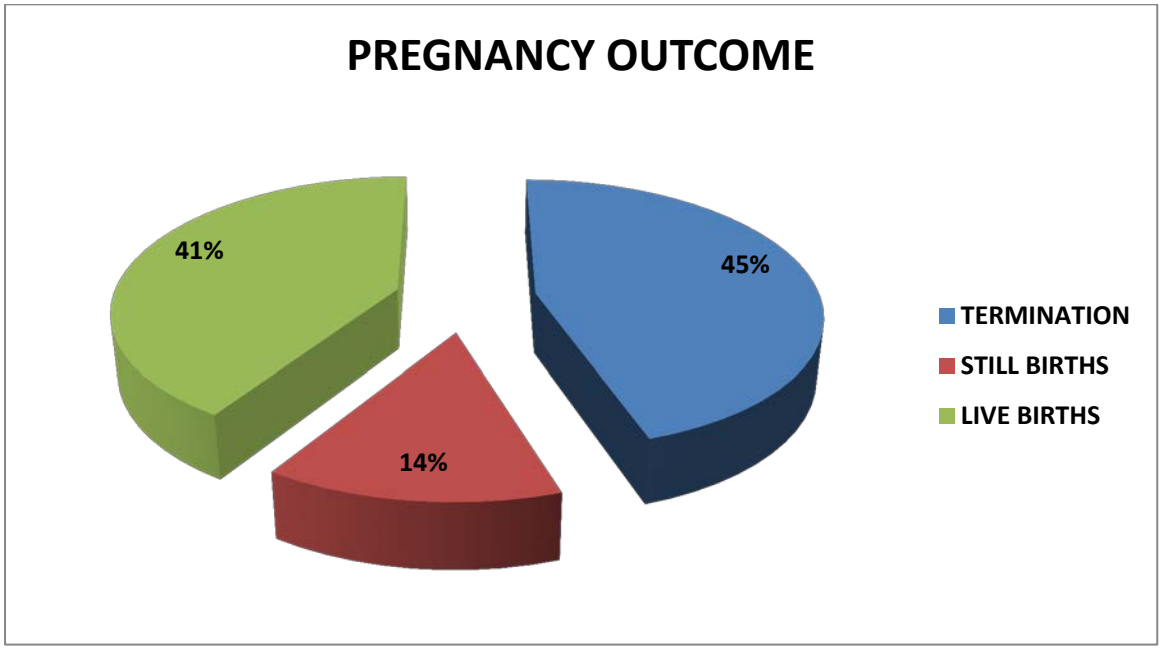

TABLE 13: OUTCOME OF THE PREGNANCIES.

Study name	Country	No. of pregnancies screened.	No. of anomalies in the screened population.	Prevalence of anomalies per 1000 screened patients.
Brocks & Bang	Denmark	14297	81	5.67
Levi etal	Belgium	16072	259	25.95
RADIUS	USA	7765	187	23.10
Helsinki	Finland	4073	45	11.05
Luck	UK	8523	67	7.80
Shirley etal	UK	6183	84	14.30
Roberts etal	New Zealand	12909	249	19.29
Chittey etal	UK	8432	125	14.82
Anderson etal	New Zealand	7880	157	19.80
S Singh etal	India (Punjab)	10890	124	11.40
Present study	India (Karnataka)	3090	61	19.74

TABLE 14: PREVALENCE OF FETAL ANOMALIES IN PUBLISHED STUDIES