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Sonographic Evaluation of Congenital Fetalanomalies

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Abstract: *Sonography in Obstetrics was first introduced by Ian Donald & colleagues in 1958 at Glasgow University in Great Britain. An early detection of fetal anomalies has become an important part of antenatal care. This helps to identify the severity of disease and its impact, lead ing to either pregnancy termination or gives opportunity for fetal therapy and/or a better neonatal care. Ultrasound is non -invasive and safe and hence can be used repeatedly. It is quick, inexpensive, easily available & causes no discomfort to the patient at any time of gestation. Fetal anomaly scan is usually carriedout at 18 -24 weeks of gestation.*

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I. Materials And Methods

Study design:
A prospective study of 4090 singleton pregnant women at our hospital.

Duration of study:
The study was conducted over a period of one and half year from November 2015 to April 2017 .

Place of study:
Department of Radiodiagnosis,at our Institute of Medical Sciences, Hyderabad.

Aims and objectives :
i) To detect the prevalence and frequency distribution of various fetal malformations among the antenatal cases presenting at our Institute ,Hyderabad and
ii) To evaluate the efficacy of antenatal sonography in the detection of various fetal anomalies

Inclusion criteria:
All singleton pregnant women coming for antenatal sonographic examination at Department of Radiodiagnosis, at our institute Hyderabad.

Exclusion criteria:
Multiple gestations.

Procedure for study:
A complete second trimester antenatal ultrasound examination of 4090 pregnant women was done using gray scale & color duplex examination on PHILIPS HD7 machine with a transducer of frequency 3.5 to 5 MHZ. The information about the gestational age, location of placenta, fetal biometry & fetal anomalies was collected. The scans were performed as a standard level one ultrasonography. In casesof uncertain abnormal findings, the women were reviewed by a level two scan with repeated scans. This data was compared with the findings at delivery / termination ofpregnancy & appropriate statistical analysis was performed.

II. Results

A prospective study was conducted in which a total of 4090 singletonpregnancies were examined at Department of Radiodiagnosis, at our Institute ofMedical Sciences, Hyderabad over a period of one and half year from November 2015 to April 2017.. They were subjected to a second trimester complete antenatal ultrasound examination using gray scale & color duplex examination on PHILIPS HD 7 machine and the information about the gestational age, location of placenta, fetal biometry & fetal anomalies was collected. Antenatal sonographic findings were correlated with the pregnancy outcome in terms of normal fetus or fetus with malformations.

TABLE1: AGE-WISE DISTRIBUTION OF ANTENATAL WOMENWITH ANOMALOUS FETUS DETECTED ON ULTRASONOGRAPHY

MATERNAL AGE IN YEARS	NO. OF CASES
< 20 YRS	18
21- 25 YRS	26
26 – 30 YRS	6
>30YRS	1

Majority of women were in the age group between 21 – 25 years.

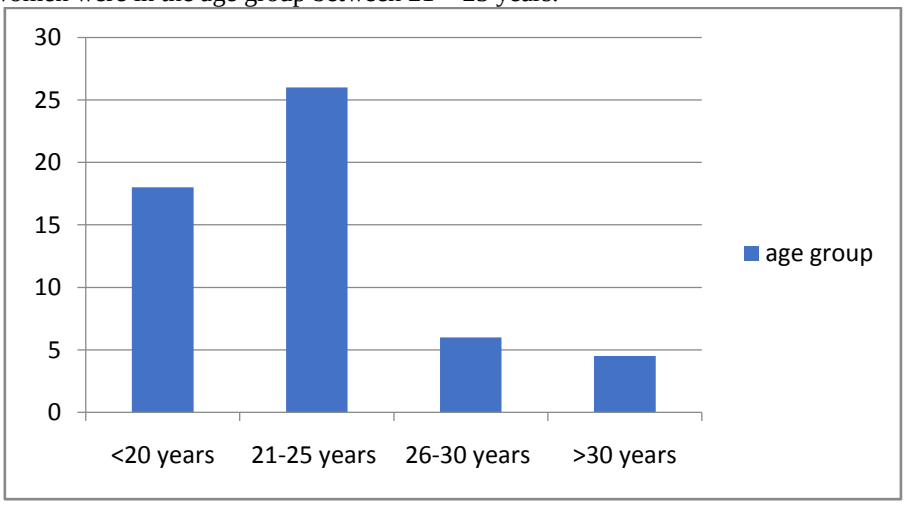
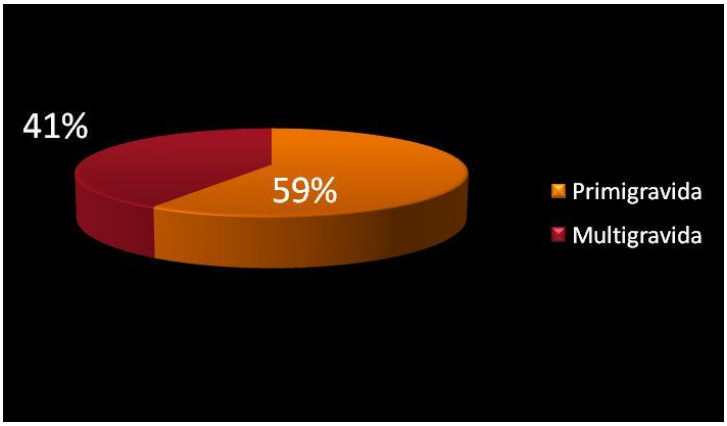


TABLE 2: DISTRIBUTION OF ANTENATAL WOMEN WITH ANOMALOUS FETUS ACCORDING TO PARITY OF WOMAN

PARITY OF WOMEN	NO. OF CASES
PRIMIGRAVIDA	30
MULTIGRAVIDA	21

It was found that the percentage of anomalies in primipara women was higher than in multipara by 18 percent.



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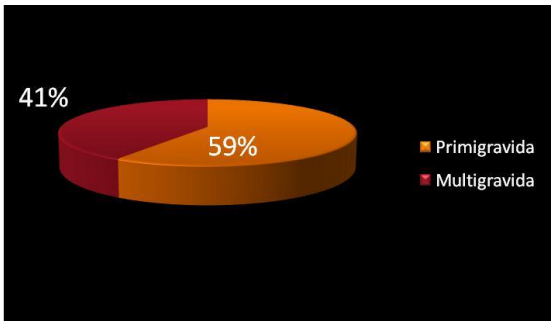


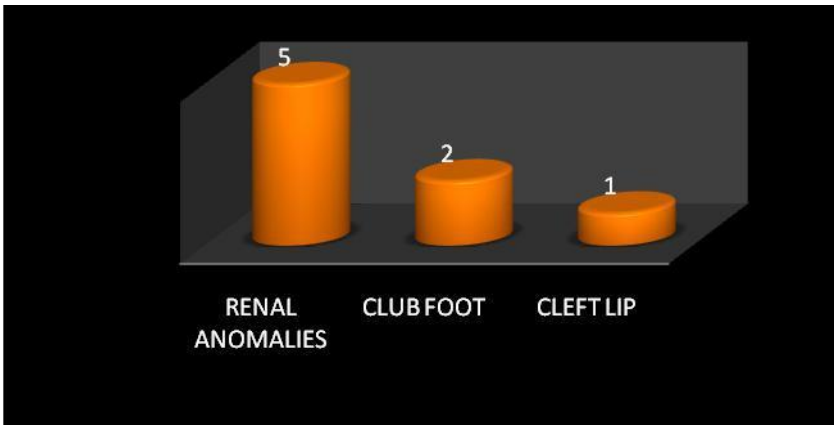
TABLE 4: SYSTEM - WISE DISTRIBUTION OF ANOMALIES ASSOCIATED WITH POLYHYDRAMNIOS

SYSTEM	NO.OF ANOMALIES
CENTRAL NERVOUS SYSTEM	7
ABDOMINAL WALL DEFECTS	2
HYDROPS FOETALIS	2
DUODENAL ATRESIA	1

Polyhydramnios was mostly associated with central nervous system anomalies.

TABLE 5: SYSTEM - WISE DISTRIBUTION OF ANOMALIES ASSOCIATED WITH OLIGOHYDRAMNIOS

SYSTEM	NO.OF ANOMALIES
RENAL ANOMALIES	4
CLUB FOOT	2
CLEFT LIP	1



In our study, oligohydramnios was mostly associated with renal anomalies. Oligohydramnios in most cases is associated with fetal renal anomalies or severe growth restriction (ref Callen).

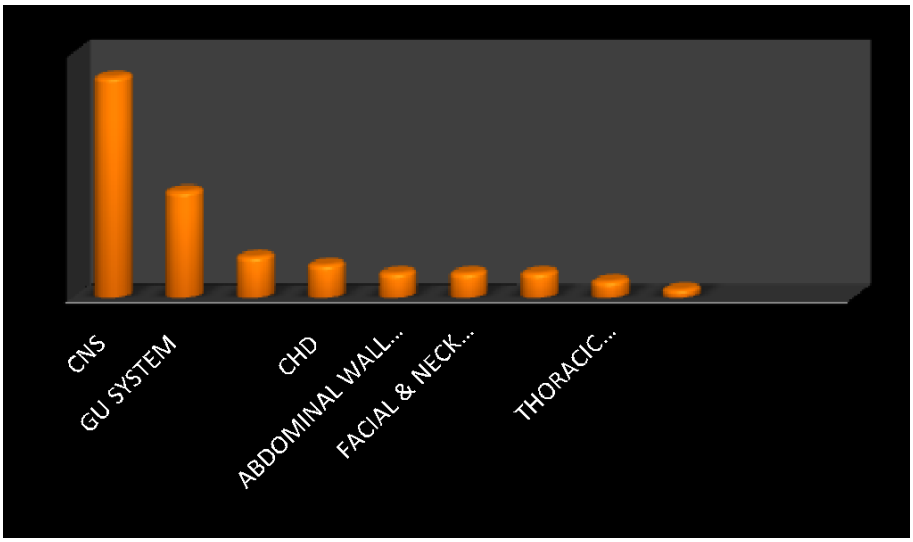


TABLE 6: SPECTRUM OF VARIOUS ANOMALIES DETECTED DURING THE STUDY

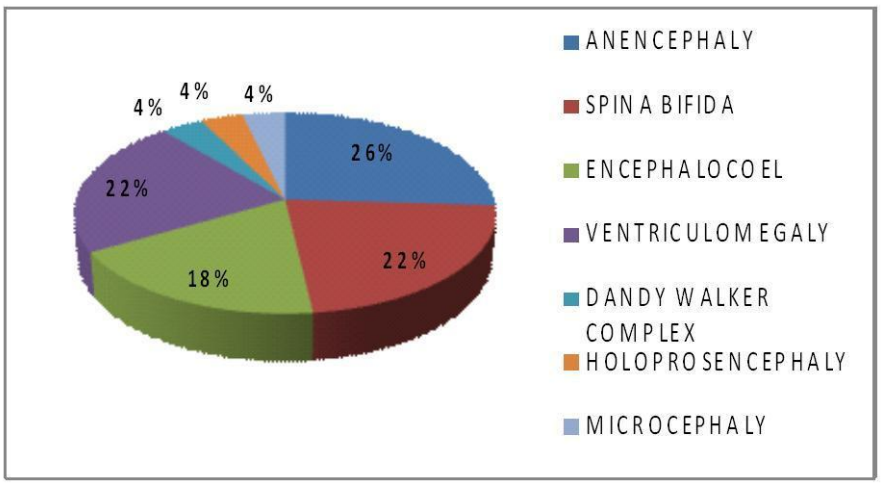
S.NO.	SPECTRUM OF ANOMALIES	NO.OF CASES
1	CENTRAL NERVOUS SYSTEM	27
2	GENITO-URINARY SYSTEM	13
3	SKELETAL SYSTEM	5
4	CONGENITAL HEART DISEASES	4
5	ABDOMINAL WALL DEFECTS	3
6	GASTRO-INTESTINAL SYSTEM	1
7	THORACIC ANOMALIES	2
8	FACIAL & NECK ANOMALIES	3
9	HYDROPS FOETALIS	3

Total 51foetuses were found to have 61 congenital malformations indicating some foetuses had more than one malformation.

most commonly detected anomalies were central nervous system with 44%, followed by genitourinary at 21%, skeletal at 8%,congenital heart diseases at 7%, abdominal wall defects, facial anomalies & hydrops foetalis at 5% each , thoracic at 3% and GI anomalies at 2%.

TABLE 7: INCIDENCE OF CENTRAL NERVOUS SYSTEM ANOMALIES

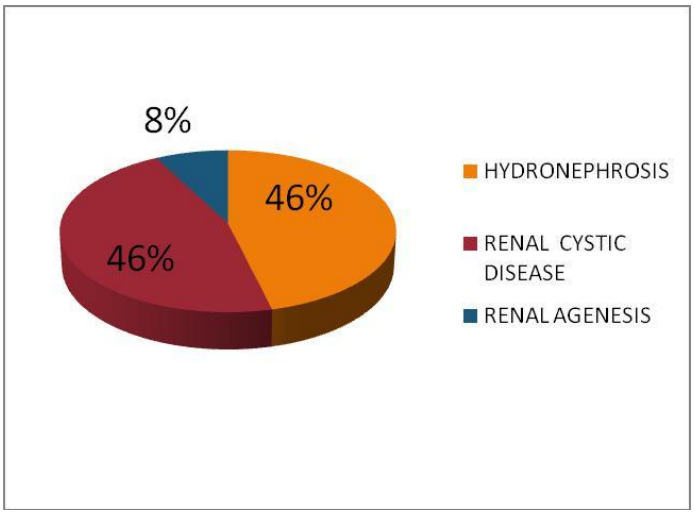
S.NO.	CNS ANOMALIES	NO.OF CASES	<22WEEKS	>22WEEKS
1	ANENCEPHALY	7	7	--
2	SPINA BIFIDA	6	6	--
3	VENTRICULOMEGALY	6	5	2
4	ENCEPHALOCELE	5	5	---
5	DANDY-WALKER VARIANT	1	1	---
6	HOLOPROSEANCEPHALY	1	1	---
7	MICROCEPHALY	1	---	1



Among CNS anomalies, most common was anencephaly at 26% and it was also the most common anomaly overall at 11.4%(7/61). The second most CNS anomaly was spina bifida &ventriculomegaly with a prevalence of 22% each in the subgroup and 9.8% each overall.

TABLE 8: INCIDENCE OF GENITOURINARY ANOMALIES

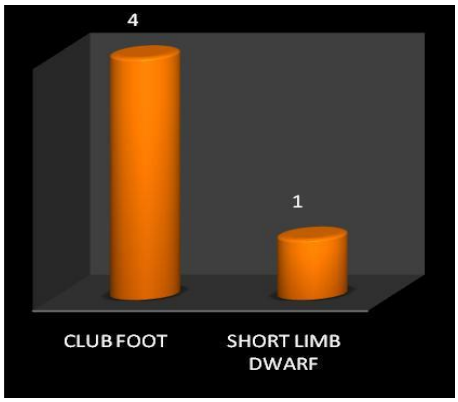
S.NO	RENAL ANOMALIES	NO. OF CASES	<22 WEEKS	>22 WEEKS
1	HYDRONEPHROSIS	6	4	2
2	RENAL CYSTIC DISEASE	6	5	1
3	RENAL AGENESIS	1	1	---



Renal anomalies were the second most common group of anomalies detected. The renal cystic disease and hydronephrosis were the commonest renal anomalies witha prevalence of 46% each in the subgroup and 9.8% overall.

TABLE 9: INCIDENCE OF SKELETAL ANOMALIES

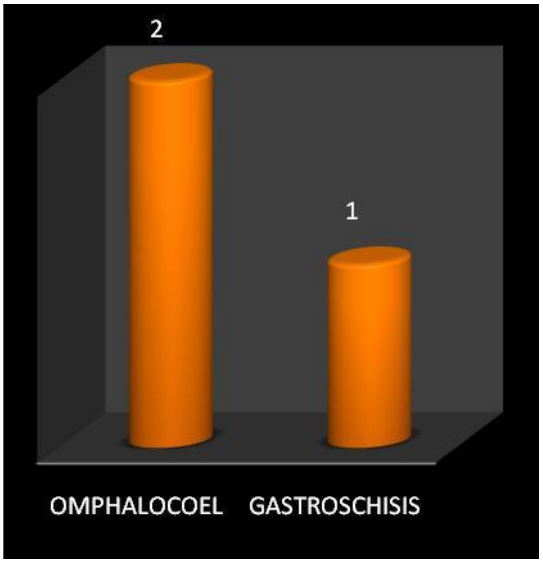
S.NO	SKELETAL ANOMALIES	NO.OF CASES	<22 WEEKS	>22 WEEKS
1	CLUB FOOT	4	2	2
2	SHORT LIMB DWARF	1	1	---



In skeletal system, club foot was the most common anomaly withprevalence of 6.5 overall.

TABLE 10: INCIDENCE OF ABDOMINAL WALL DEFECTS

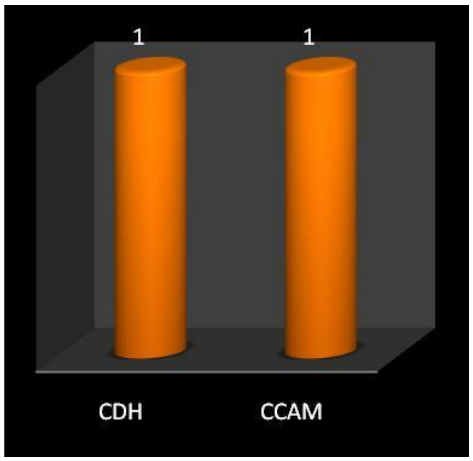
S.NO	ABDOMINALWALL DEFECTS	NO. OF CASES	<22WEEKS	>22WEEKS
1	OMPHALOCOEL	2	2	----
2	GASTROSCHISIS	1	1	----



Among abdominal wall defect anomalies ,omphalocoel and gastroschisis weremost commonly detected

TABLE 11: INCIDENCE OF THORACIC ANOMALIES

S.NO.	THORACIC ANOMALIES	NO. OF CASES	<22 WEEKS	>22WEEKS
1	CDH	1	1	----
2	CCAM	1	1	----



Congenital diaphragmatic hernia and congenital cystic adenomatoid malformation were most commonly detected thorasic anomalies.

TABLE 12: INCIDENCE OF GASTROINTESTINAL TRACT ANOMALIES

S.NO.	GI ANOMALIES	NO. OF CASES	<22WEEKS	>22WEEKS
1	DUODENAL ATRESIA	1	1	-----

TABLE 13: INCIDENCE OF OTHER ANOMALIES

S.NO	ANOMALIES	NO. OF CASES	<22 WEEKS	>22 WEEKS
1	CONGENITAL HEART DISEASES	4	1	3
2	FOETAL HYDROPS	3	1	2
3	FACIAL AND NECK	3	2	1

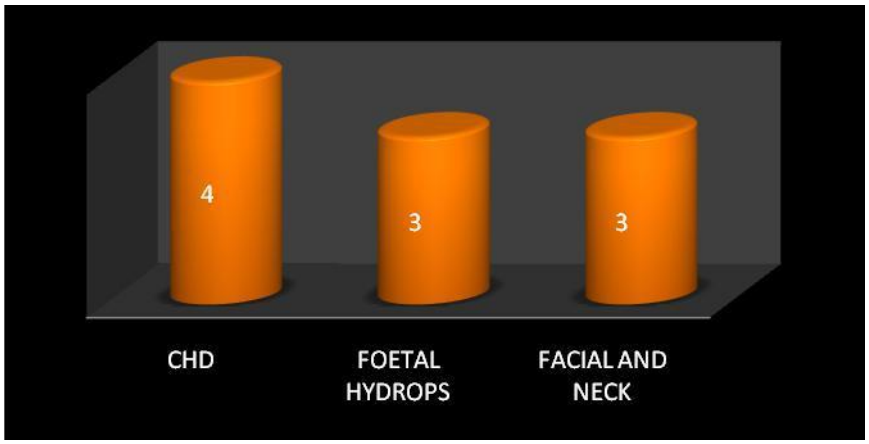


TABLE 14: EFFICACY OF SECOND TRIMESTER ULTRASOUND IN DETECTING FETAL ANOMALIES

Total number of antenatal scans done: 4090

No. of pregnant women with fetal anomaly: 51

No. of cases detected on antenatal ultrasound: 45

No. of cases missed on antenatal scan overall (False negative): 6

No. of false positive cases: 5

Total no. of anomalies detected: 61

No. of anomalies detected before 22 weeks: 47

No. of cases detected after 22 weeks: 8

ULTRASOUND SCAN	ANOMALY PRESENT	ANOMALY ABSENT
POSITIVE	45 TRUE POSITIVE	5 FALSE POSITIVE
NEGATIVE	6 FALSE NEGATIVE	4034 TRUE NEGATIVE

PREVALENCE OF CONGENITAL MALFORMATIOS (61/4090): 1.49 %

SENSITIVITY OF SCAN: 88.2%

SPECIFICITY OF SCAN: 99.8%

III. Discussion

Congenital anomalies are important cause of still births, infant mortality and are contributors to childhood morbidity. In countries where routine prenatal ultrasound examination is practiced, it is noted that ultrasound scanning is beneficial in detecting congenital malformations at a stage where intervention is an option.

Women who delay child bearing are at an increased risk of having an adverse outcome of pregnancy[1].Increasing maternal age is independently linked with definite adverse pregnancy as well as fetal abnormalities and multiple gestation[2]. Bobrowski R et al have statistically shown that mothers between 25 to 30 years of age stand at a higher risk of producing malformed babies[3]. Sugunabai[4] reported a higher incidence of malformation in babies born to mothers aged over 35 years, whereas Datta etal[5] documented statistically insignificant association of increased maternal age and congenital anomalies.

Since our study did not have any women with detected fetal anomaly above 35 years, we could not correlate and infer anything when we compared our study with that of Sugunabai et al. Most of the pregnant women coming to our hospital for anomaly scan were between the age group of 21-25 years. It was also noted that the percentage of anomalies was more in the age group 21-25 years.

Another maternal factor that was studied was obstretic history. Amar Taksande etal[6] found that the percentage of anomalies in primipara women was higher than in multipara by 1 to 3 percent. A prospective study conducted by S.Singh etal[7] also found most of the fetal anomalies in primigravida. This corresponds to the results in our study wherein the percentage of anomalies in primigravida was more than in multigravida. Previous study by Mohanty C, Mishra OP, Das BK etal[8] have reported significantly higher incidence of malformation among multigravida. Our results differ with this finding.Amar Taksande et al evaluated that the factors which significantly increase the risk of congenital malformations were presence of hydramnios, maternal febrile illness in first trimester, past history of abortions, diabetic mother, eclampsia, history of previous child with congenital anomalies or malformed babies. We in our study found that nearly 15 percent women had a history of previous child with an anomaly among the total anomalies detected during the study.

We had 3 women with gestational diabetes amongst 51 antenatal cases with detected fetal anomalies, which is approximately 7.8%. Ordonez et al[12] stated that diabetes mellitus , arterial hypertension and hypothyroidism show a positive association with congenital malformation .This was inconsistent with our study in which the incidence of congenital anomalies in women who had history of diabetes mellitus was lower than in women who did not have a positive history..

When we look at the amniotic fluid index, most of the anomalies were associated with normal amniotic fluid. The percentage of anomalies associated with polyhydramnios was slightly higher than those associated with oligohydramnios. Oligohydramnios, in most cases is associated with fetal renal malformations or severe growth restriction (Callen). Polyhydramnios is seen mostly associated with CNS anomalies. The diagnosis of polyhydramnios, according to Callen , although seeming to be less serious , in many cases may be associated with significant complications to the mother and the fetus which can result in development of fetal anomalies.

We detected 61 anomalies in 51 malformed oetuses indicating that some oetuses had more than one malformation. We obtained different types of anomalies. 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 foetalis at 5% each , thoracic at 3% and GI anomalies at 2%. There have been previous studies like that of Anderson etal, Kalraet al[13], Kook Lee et al that are in line with our studies.

Contrary to our study, 45th annual report of Indian Council of Medical Research[57], 2002-03 says that the commonest congenital malformations are cardiac in nature (0.57%). Nevertheless in our hospital study the incidence of CNS related malformations was the highest.

Singh et al detected 124 malformed oetuses with 150 malformations. In their study the most common anomaly was ventricolomegaly followed by anencephaly and neural tube defects accounted for >50% of the anomalies with associated spinal deformities. This was comparable with our study.

Among CNS anomalies , neural tube defects were the most commonly detected anomalies with anencephaly being the commonest, 26% in subgroup and 11.4%(overall 7/61). The second most common CNS anomaly was spina bifida &ventriculomegaly with a prevalence of 22% each in subgroup and 9.8% each overall. The previous studies like the Eurofetus study, Agarwal SS etal, Shamaoetuses l also showed CNS anomalies as the most common type of fetal anomalies detected on routine antenatal sonography.

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 was seen associated with multicystic kidney disease.

On comparing the genitourinary system, in our study the prevalence of hydronephrosis was 9.8%, which had less variance when compared with study by Shirley et al (7.5%).We have seen during the study that few cases which showed dilated renal pelvis during late second or third trimester ,on follow up scans appeared normal at term or postnatally. In a previously mentioned study by Kook Lee, the incidence of renal cystic disease was 2% which when compared to our study was less. In our study the prevalence was 9.8%. Among them, bilateral renal cystic diseases were associated with severe oligohydramnios and incompatible with life.

In the Musculoskeletal system,we got a prevalence of 1.6% for Short Limb dwarfism. In a study by Katherine Fong, the incidence of clubfoot was 3% which had a little variance with our study with 4.9% incidence.

Abdominal wall defects constituted 5% of the anomalies detected.The incidence of Omphalocele in a study carried out by Kook Lee was 6% whereas in our study it was 3.2%. The prevalence of gastroschisis, Congenital Diaphragmatic hernia, duodenal atresia, congenital cyst adeomatoid malformation of lung was 1.6% each.

Cardiac defects accounted for 7% of the anomalies detected. The detection rate for cardiac anomalies was less in our study. A study by Kirk JS, Comstock CH et al[15] concluded that the sensitivity of sonographic screening to defect fetal cardiac anomalies varies with the type of lesion. The highest detection rates involve hypoplastic ventricles and atrioventricular septal defects, followed by lesions of the great arteries. Isolated septal defects were the most difficult lesions to detect.

The prevalence of Hydrops fetalis came to be 4.9 % in our study. However, in a study by Kook Lee the incidence was calculated to be 10%.among the three cases of hydrops foetalis, two of them had 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. Fetalposition, oligohydramnios and maternal obesity may obscure proper visualization of fetal parts and hence considered limiting factors for a complete scan. Maarse

W, Bergé SJ et al[16] stated that the diagnostic accuracy of 2D ultrasound screening for cleft lip and palate in a low-risk population has a relatively low detection rate. 3D ultrasound can achieve a reliable diagnosis, but not of cleft palate only.

On comparing the period of gestation at which the malformations were detected, it was observed that 47 out of 61 anomalies were detected before 22 weeks of gestation constituting 77% of the anomalies. In our study, the false negative for a mid trimester scan was higher (ie) 22.9 % than when compared to overall detection (ie) 9.8%. The percentage of false positive cases was 8.1% .This was comparable to study by Anderson et al with 10% of false positive cases. Out of the 14 anomalies which were detected after 22 weeks, there were two cases of fetal hydrops which presented in late second trimester, two cases of hydronephrosis which on followup scan in third trimester showed an increased renal pelvis AP diameter. One case of renal cystic disease with associated microcephaly and cccipitalencephalocele was detected in late second trimester because of the non compliance of the women for a mid second trimester scan. There was one case of ventriculomegaly which on follow up scans at third trimester showed an increase in atrial width. There were six cases which were missed during antenatal scans overall. These included three congenital heart diseases, two club foot and one cleft lip.

Two cases of club foot were not diagnosed during antenatal scan as they were associated with oligohydramnios which was limitation for a detailed scan. The use of prenatal ultrasonography for the detection of club foot has increased steadily during the 90"s. Its accuracy is related to several factors such as skill of the examiner, quality of the ultrasonographic equipment, gestational age, amount of amniotic fluid, fetal position and the methodology used.

According to a study by D.Keret et al, three types of isolated idiopathic club foot were identified based on the age of prenatal detection.

Early club foot: detected between 12^{th} to 17^{th} week of gestation on TVS

ii) Late onset club foot: detected between 18^{th} and 24^{th} weeks.

iii) Very late onset club foot: seen between 25^{th} and 32^{nd} weeks of gestation.

Bar- Hava I, BronshteinM et al[17] have shown that a fetus can temporarily turn the foot into a position simulating club foot, but this aberrant positioning is usually expected to readjust during a standard examination of 30 minutes.

One case of cleft lip was missed antenatally, as it was associated with oligohydramnios and also the fetal position, maternal obesity were the limiting factors. Three cases of congenital heart disease were missed on antenatal scan. This indicates the level of expertise and quality of equipment required for detection of cardiac anomalies.

Out of the five false positive cases, we had 3 cases of mild hydronephrosis and 2 cases of mild ventriculomegaly. The three cases of mild (grade I) hydronephrosis resolved in the later gestational weeks with postnatal confirmation of complete resolution. Feldman DM et al[61] suggested that most cases of mild hydronephrosis will resolve before delivery. In his study out of the 88% oetuses that were detected with mild hydronephrosis, most had complete resolution before delivery.

The two cases of mild ventriculomegaly which were detected in the second trimester were almost normal at term on follow up examination. This was further confirmed postnatally.

IV. Conclusion

Obstetric ultrasonography has become an important part of routine antenatal care. Routine anomaly screening improves perinatal outcome directly through termination of pregnancy for certain anomalies. Fetuses with conditions that required immediate postnatal pediatric surgery like urinary tract anomalies were referred to an appropriate center. Some parents who were confronted with the diagnosis of a lethal or severely impairing fetal disease frequently opted termination of pregnancy. In conclusion, with these diagnostic capabilities, the advantages of mid-trimester routine ultrasonographic screening for major fetal anomalies was useful in the given study population although the detection rate was low for cardiac anomalies and few facial defects. Almost all lethal and life threatening anomalies could be diagnosed antenatally to allow the options of counseling, pregnancy termination, or selective referral. Also, the prevalence of anomalies and the sensitivity in the present study is comparable to other studies.

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