Evaluation of the Efficacy of Antenatal Sonography in Detecting Various Fetal Congenital Anomalies in the Second Trimester of Pregnancy

ABSTRACT
OBJECTIVE: TO EVALUATE THE EFFICACY OF ANTENATAL SONOGRAPHY IN DETECTING VARIOUS FETAL CONGENITAL ANOMALIES IN THE SECOND TRIMESTER OF PREGNANCY.

Ultrasound has become an invaluable tool in the antenatal diagnosis of many fetal abnormalities, by allowing early and reliable identification of the severity of various fetal anomalies and their 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. The aim of this study was to assess the validity of ultrasound in prenatal detection of congenital anomalies.

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. This data was correlated with the pregnancy outcome and appropriate statistical analysis was performed.

RESULTS: A total of 61 anomalies were detected in 51 fetuses. Prevalence of congenital malformations was 1.97%. The sensitivity of the ultrasound scan was 88.46% and specificity 99.83%. The positive predictive value of ultrasound scans was 90.19% and the negative predictive value was 99.80%.

CONCLUSION: Ultrasonography is a highly effective and safe imaging modality for early detection of fetal malformations. 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.

INTRODUCTION
A second trimester ultrasound scan provides the pregnant woman and her doctor with information about multiple aspects of her pregnancy. It provides an opportunity to diagnose congenital anomalies as well as to detect any soft markers of aneuploidies. Congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India. 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.

Congenital anomalies are defined as structural defects, chromosomal abnormalities, inborn errors of metabolism and hereditary diseases diagnosed before, at, or after birth. Ultrasonography has a great potential in screening for morphological abnormalities throughout all trimesters of the pregnancy, being non-invasive, safe, fast, accurate and reproducible with real time display, causing no discomfort to the patient at any time of gestation. A second trimester anomaly scan has been suggested in routine antenatal care to increase the prenatal detection rate of fetal defects. Ultrasonography can identify at least 35 – 50% of major fetal malformations with a specificity of 90 – 100%.

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. This data was correlated with the pregnancy outcome and 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: Routine ultrasound screening was performed for all pregnant women between 18-24 weeks gestation, attending the ultrasound department of our hospital during the study period. The scans were performed as a standard level one antenatal ultrasonography. In cases of uncertain abnormal findings, the women were reviewed by a level two scan with follow-ups (if required).

Specific components of the ultrasonographic examination included assessment of placental location, amniotic fluid volume, fetal number, presentation, cardiac activity, biometry (BPD, HC, AC, FL), and a fetal anatomical 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 (See Table 4)
1. The total number of antenatal scans performed was 3090, out of which a total of 61 anomalies were detected in 51 fetuses. The prevalence of congenital malformations was (61/3090): 1.97%.
2. The sensitivity of ultrasonography scans was 88.46% and specificity 99.83%.
3. Number of cases with anomalies missed on antenatal scan (False negative): 6

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4. Number of normal cases misdiagnosed as anomalies on antenatal scan (False positive): 5
5. The positive predictive value of ultrasonography scans was 90.19% and the negative predictive value was 99.80%.

DISCUSSION

Any deviation from the normal range during morphogenesis constitutes an anomaly. Congenital anomalies are important causes of still births, infant mortality and morbidity. Ultrasonography has emerged as one of the most powerful tools for prenatal diagnosis of congenital malformations and its use with obstetric patients has become a routine part of prenatal care.

Most of the pregnant women referred to the Ultrasound department of our hospital for anomaly scans were in the age group of 21-25 years and it was noted in our study that the percentage of anomalies was also comparatively more in this age group (See Table 1).

In our study, we detected a total of 61 anomalies in 51 malformed fetuses, which indicated that some fetuses had more than one malformation. The most commonly detected fetal anomalies in our study were of CNS at 43%, followed by genitourinary at 21%, skeletal at 8%, congenital heart diseases at 7%, abdominal wall defects, facial anomalies & hydronephrosis at 5% each, thoracic at 3% and GI anomalies at 2% (See Table 2).

There were 6 cases which were missed during the second trimester antenatal scans. These included 3 cases of congenital heart diseases, 2 cases of club foot and 1 cleft lip.

The 2 cases of club foot were not diagnosed during antenatal scan as they were associated with severe oligohydramnios, which was a limitation for a detailed anomaly scan. The 1 case of cleft lip was missed antenatally, as it was also associated with severe oligohydramnios. Unfavorable fetal position and maternal obesity were also the limiting factors in the same case. The 3 cases of congenital heart disease were also missed on antenatal scans. This indicated the level of expertise and the quality of equipment required for detection of cardiac anomalies. It has been acknowledged in other studies that even in the best of hands and circumstances, the 18–22 week scan has limitations and cannot detect all fetal and maternal abnormalities.

The percentage of false positive cases was 8.1%. Out of the 5 false positive cases, we had 3 cases of mild hydronephrosis and 2 cases of mild ventriculomegaly. The 3 cases of mild (grade I) hydronephrosis were found to have been resolved on follow-up third trimester scans, along with postnatal confirmation of complete resolution. Feldman DM et al suggested that most cases of mild hydronephrosis will resolve before delivery. In his study out of the 88% fetuses that were detected with mild hydronephrosis, most had complete resolution before delivery.

The 2 cases of mild ventriculomegaly which were detected in the second trimester in our study were almost normal at term on follow up examination. This was further confirmed postnatal.

The sensitivity of ultrasonography scans in our study was computed to be 88.46%, which was comparable to other similar studies and the specificity was 99.98%, which was also comparable to other similar studies (See Table 3).

A retrospective study of Chittey et al (1998), who had a sample size of 8785 concluded with a 1.5% prevalence of anomalies, sensitivity of 71.5% and 99.98% specificity, which was comparable with our study. Levi et al (1991) did a large prospective study with a sample size of 15654. The prevalence of anomalies was 2.66%, with sensitivity of 40.4% and specificity 99.82%. Our study had better sensitivity and a comparable specificity. Shirley et al (1991) in their retrospective study had 6412 scans done at 19 weeks and obtained a prevalence of 1.4%. Their sensitivity was 57.3% and specificity 99.97%. Our study had the same prevalence and specificity but the sensitivity was more.

Another prospective study by Luck et al (1992) had a sample size of 8844, prevalence of 1.9%, sensitivity of 85.3% and specificity 99.90%. Their sensitivity was more than our study but specificity of our study was much comparable. Souka et al (2006) in Greece did another prospective study with a sample size of 1148 and prevalence of 1.21%. Their study concluded with a better sensitivity of 92.9% but the specificity of 99.74% was comparable to our study (See Tables 3 & 4).

The positive predictive value of second trimester antenatal Ultrasonographic scans was estimated at 90.19% and the negative predictive value was 99.80%.

TABLE 1: AGE-WISE DISTRIBUTION OF ANTEUNATAL WOMEN WITH ANOMALOUS FETUS DETECTED ON ULTRASONOGRAPHY.
TABLE 4: EFFICACY OF SECOND TRIMESTER ULTRASOUND IN DETECTING FETAL ANOMALIES

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REFERENCES