ISSN: 2581 – 3633
PubMed - National Library of Medicine - ID: 101745081

International Journal of Medical Science and Advanced Clinical Research (IJMACR)

Available Online at:www.ijmacr.com

Volume – 7, Issue – 3, June - 2024, Page No. : 102 – 109

A prospective study to evaluate the efficacy of early anomaly scan in detecting fetal structural anomalies versus traditional anomaly scan

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How to citation this article: Dr.Hema K R, Dr.Yashasvini K, Dr. Rithika P, Dr. Nidhi B R, Dr. Dwarakanath L, "A prospective study to evaluate the efficacy of early anomaly scan in detecting fetal structural anomalies versus traditional anomaly scan", IJMACR- June - 2024, Volume – 7, Issue - 3, P. No. 102 – 109.

Type of Publication: Original Research Article

Conflicts of Interest: Nil

Abstract

Introduction: Every pregnant woman has a desire to have a healthy child who is free of anomalies. The incidence of major malformation in newborn infants is around 3-5%.[1]The early first trimester scan was initially introduced with the intention of measuring the fetal crown rump length to achieve accurate dating of pregnancy. Attempts to detect fetal disorders during the first trimester have been confined to high risk & selected populations.

Objectives: To identify serious fetal abnormalities either incompatible with life or associated with morbidity and to evaluate the influence of various fetal anomalies in obstetric decision making.

Method: It was a prospective study conducted for duration of 18 months.150 antenatal mothers who attended antenatal clinic were recruited for the study.

Results: 150 antenatal mothers underwent the second trimester scan and were analyzed based on the distribution of age, gravida, risk factors, no of anomalies detected and the outcome of their pregnancies. 68% of

participants belonged to 18-24 years of age, 50% of participants were primigravida and 2% of participants had previous anomalies, 3.3% had family history of anomaly and 3.3% had history of drug intake. 8% of participants had medical disorders. Anomalies detected at 11-13 weeks scan were anencephaly, cystic hygroma and anomalies detected at 18-22 weeks scan were right limb amelia, hypoplastic right ventricle with pulmonary atresia, congenital diaphragmatic hernia.

Conclusion: The ultrasound examination at 11-14 wks to screen for fetal abnormalities is effective and can be an adjunct to the routine 18-22 weeks anomaly scan. Hence first trimester ultrasound (11-14wks) must be made mandatory, not only for the detection of major fetal anomalies, but also for diagnosing abnormal pregnancy like missed abortion, molar pregnancy and ectopic pregnancy. Towards the end of the first trimester, the scan additionally offers an opportunity to identify gross fetal irregularities and, in health systems that offer first trimester aneuploidy screening, the measuring of the nuchal translucency thickness (NT) is also available.

Keywords: Nuchal Translucency Thickness, Aneuploidy, Imaging Technology

Introduction

Every pregnant woman has a desire to have a healthy child who is free of anomalies. The incidence of major malformation in newborn infants is around 3-5%.[1] Ultrasound plays a central role in the provision of prenatal screening and also in diagnosis. Since Professor Ion Donald introduced ultrasound into obstetrics in the late 1950's vast improvement has been made in the ultrasound technology. These revolutionary improvements and the use of high frequency transvaginal scanning have allowed the resolution of ultrasound imaging in the first trimester to evolve to the stage where detailed early fetal development can be well visualized.

The early first trimester scan was initially introduced with the intention of measuring the fetal crown rump length to achieve accurate dating of pregnancy. Moreover, imaging technology also made it possible to accurately diagnose chromosomal and structural anomalies before the second trimester. Based on the Radius Trial, the routine anomaly scan was done at 18-22 weeks but now the emphasis for screening for fetal abnormalities have been shifted to 11-13^{+6}weeks.[2]

Until recently, attempts to detect fetal disorders during the first trimester have been confined to high risk & selected populations. Evidence has shown that routine early ultrasound in screening for low risk pregnancies for fetal abnormalities is beneficial .[3]

Advantages of early ultrasound screening

a) Provides reassurance to the patient as well as to the obstetricians.

b) Provides information regarding specific abnormalities or aneuploidy.

c) Provides opportunity for prenatal invasive/non invasive testing.

d) Helps in deciding for early termination of anomalous fetuses.

e) Provides knowledge of the condition that may affect future pregnancies, provides essential counseling.

The 11-13^{+6} weeks of gestation are the best period to assess for gestational age, measurement of Nuchal translucency thickness and also for a potentially detailed anatomic survey. Whenever there is patient's history of an abnormal finding on a screening examination a more thorough examination of the fetal anatomy is performed. Many of the recent studies supports the utility of

Sonographic detection of major fetal structural abnormalities in the first trimester .Unfortunately, few anomalies will not be evident until later part of pregnancy because of ongoing fetal anatomical development. Detection rates (DRs) of various anomalies vary according to the organ system being examined, equipment settings, and radiologist experience. Screening for structural anomalies by ultrasound examinations, at 11–13^{+6} and 18–22 weeks, is gaining acceptance due to the detection of an increasing number of anomalies in the first trimester.[4]

As there is dearth of evidence in this area of research, this study was undertaken to add to the evidence about the outcomes of early anomaly scan in pregnancy.

Methodology

The aim of the study was to evaluate the efficacy of 11-13+6 weeks anatomy scan in detecting fetal structural anomalies compared to traditional 18 to 22 weeks scan". The objectives of study were to identify serious fetal abnormalities either incompatible with life or associated with morbidity and to evaluate the influence of various fetal anomalies in obstetric decision making.

It was a prospective study conducted at the Department of Obstetrics and Gynecology, Sri Siddhartha Medical College and Hospital Tumkur for duration of 18 months.150 antenatal mothers who attended antenatal clinic were recruited for the study. All pregnant women who came for antenatal checkup during the 11 to 22 weeks and gave written informed consent were included for the study. Multiple gestation, Gestational age(GA) <11 weeks, GA >22 weeks, Vesicular mole, Intrauterine demise on first ultrasound study itself were excluded.

Procedure: Ultrasound screening was performed by experienced radiologist transabdominally using 2 to 6MHZ curvilinear transducer and VOLUSON S8 PRO Ultrasound machine. Whenever visualization of fetal structure was suboptimal or a structural abnormality was suspected during transabdominally scan, transvaginal scan was always performed. Ultrasound screening was performed at 11-13.6 Weeks in all150 pregnant women. For those who had normal scans a follow up scan was done at 18-22 weeks for confirmation. Fetal viability was examined and crown rump length was measured.

Evaluation of fetal anatomy was done according to the following checklist.

1. Skull and brain
2. Face (Facial Profile, Nasal Bone and orbits)
3. Neck (Nuchal translucency measurement, presence of Cystic hygroma)
4. Spine (Examination of overlying skin and neural tube in longitudinal and transverse planes)
5. Heart (Four chamber view, three vessel view, heart rhythm)
6. Stomach (Its existence in left upper abdomen)
7. Abdominal wall defect
8. Kidney (existence, size, and shape)
9. Urinary bladder (existence, size, and shape)
10. Extremities (existence, size, and shape)

Along with the evaluation of the anatomy, the Nuchal translucency measurement was done according to the guidelines established by the Fetal Medicine Foundation, in fetuses with CRL between 45mm and 84mm at 11 to 13.6 weeks gestational age. The cut off value of NT measurement was taken as > 3 mm. When the NT measurement was > 3 mm it was considered to be abnormal and further confirmatory tests were combined. Women were fully counseled before their ultrasound examination and written informed consent was obtained. Based on the anomalies detected, the patients were counseled regarding termination or continuation of

pregnancy. All the patients were followed up till delivery. The number of abnormalities that were detected at 11-13 weeks and at 20 weeks were analyzed.

Statistical Analysis

Sample size was calculated using the formula n =Z^2(1-α/2)xP (1-P)/d2.

Z-Standard normal variate for 95% confidence interval=1.96, Prevalence (p)=0.003(3%), Precision(d)= 0.003(3%). On calculation n=124, Taking non-response of sample size as 10%, sample size was calculated to be 137.

Statistical Methods: Descriptive and inferential statistical analysis has been carried out in the present study. Results on continuous measurements are presented on Mean SD (Min-Max) and results on categorical measurements are presented in Number (%). Significance is assessed at 5 % level of significance. The following assumptions on data are made,

Assumptions: 1. Dependent variables should be normally distributed, 2.Samples drawn from the population should be random, Cases of the samples should be independent. Chi-square/ Fisher Exact test has been used to find the significance of study parameters on categorical scale between two or more groups, Non-parametric setting for Qualitative data analysis. Fisher exact test used when cell samples are very small.

Significant figures considered were

+ Suggestive significance (P value: 0.05<P<0.10)

* Moderately significant (P value:0.01<P 0.05)

** Strongly significant (P value: P0.01)

Statistical software: The Statistical software namely SPSS 22.0, and R environment ver.3.2.2 were used for the analysis of the data and Microsoft Word and Excel have been used to generate graphs, tables etc**.**

Results

During the one year period of 2021-2022, 165 pregnant women who attended our antenatal clinic were enrolled for evaluation for fetal structural and chromosomal abnormalities between 11 and 13.6 weeks of gestational ages. Out of the 165 antenatal mothers who had ultrasonogram, 15 patients were excluded from the study because 10 patients were lost to follow up and not seen during the second trimester, 3 of them had findings suggestive of missed abortion at the time of the 11-13 weeks scan and 2 of them had miscarriages around 16 weeks of gestation. The remaining 150 antenatal mothers underwent the second trimester scan and were analyzed based on the distribution of age, gravida, risk factors, no of anomalies detected and the outcome of their pregnancies.

68% of participants belonged to 18-24 years of age, 50% of participants were primigravida and 2% of participants had previous anomalies, 3.3% had family history of anomaly and 3.3% had history of drug intake. 8% of participants had medical disorders. anomalies detected at 11-13 weeks scan were anencephaly, cystic hygroma and anomalies detected at 18-22 weeks scan were right limb amelia, hypoplastic right ventricle with pulmonary atresia, congenital diaphragmatic hernia.

Table 1: Details of USG scan

Variables	No. of Patients	%
First Trimester Scan		
·Normal	148	98.7
·Abnormal	2	1.3

Second Trimester scan		
·Normal	147	98.0
·Abnormal	3	2.0
Total	150	100.0

Table 2: details of anomalies

Total No. of Anomalies	No. of Patients	%
Normal	145	96.7
Abnormal	5	3.3
·Anencephaly	1	0.7
·Congenital Diaphragmatic Hernia	1	0.7
·Cystic Hygroma	1	0.7
·Hypoplastic Right Ventricle With Pulmonary Atresia	1	0.7
·Right Lower Limb Amelia	1	0.7
Total	150	100.0

Table 3: Association of Investigations in relation to Total number of anomalies of patients studied

Variables	Total Number of Anomalies		Total	P Value
	No	Yes		
First Trimester Scan				
·Normal	145(100%)	3(60%)	148(98.7%)	
·Abnormal	0(0%)	2(40%)	2(1.3%)	
Second Trimester Scan				
·Normal	145(100%)	2(40%)	147(98%)	<0.001**
·Abnormal	0(0%)	3(60%)	3(2%)	
Total	145(100%)	5(100%)	150(100%)	

Table 4: Association of History variables in relation to Total number of anomalies of patients studied

Variables	Total Number of Anomalies		Total	P Value
	No	Yes		
Ho Drug In Take				
· No	141(97.2%)	4(80%)	145(96.7%)	0.157
· Yes	4(2.8%)	1(20%)	5(3.3%)	

HO_Medical Disorders				
Normal	136(93.8%)	2(40%)	138(92%)	0.003**
Abnormal	9(6.2%)	3(60%)	12(8%)	
· DM	2(1.4%)	1(20%)	3(2%)	
· EPI	2(1.4%)	1(20%)	3(2%)	
· GDM	1(0.7%)	1(20%)	2(1.3%)	
· HTN	2(1.4%)	0(0%)	2(1.3%)	
· HD	1(0.7%)	0(0%)	1(0.7%)	
· Hypothyroid	1(0.7%)	0(0%)	1(0.7%)	
Total	145(100%)	5(100%)	150(100%)	

Table 5: Association of Pregnancy outcome in relation to Total number of anomalies of patients studied

Variables	Total Number of Anomalies		Total	P Value
	No	Yes		
Pregnancy Outcome				
·Alive	145(100%)	1(20%)	146(97.3%)	<0.001**
·Termination of pregnancy	0(0%)	4(80%)	4(2.7%)	

Discussion

This was a prospective study conducted in the department of obstetrics and gynecology in a tertiary care center to assess the outcomes in early anomaly scans at 11-13 weeks of gestation. The mean age of the participants was 24.19(3.57). The anomalies were found in 80% of primigravidas and 20% of multigravida. 40% of mothers with anomalous infants had consanguineous marriage. Among the 5 anomalies detected there was no recurrent anomaly. There was no previous history of anomaly in the anomalous group. One among five mothers with anomalous infants had a history of drug intake of antiepileptic one among 5 anomalies had family history of anomalies. 8% of participants had medical history and this was statistically significant. Among mothers with anomalous babies, one had gestational diabetes, one had type 2 diabetes mellitus and the other had epilepsy. Four out of five anomalous babies were terminated medically and one was delivered by cesarean section and it was followed up with a pediatric surgeon for congenital diaphragmatic hernia.

It was found that family H/O anomalies and positive history of drug intake (anticonvulsants and other teratogenic drugs) had a very high risk for presence of anomalies. Hence first trimester ultrasound is useful in early diagnosis of structural anomalies in high risk populations. The incidence of major fetal anomalies in this study was about 3.3% and it falls within the range reported in the literature. The RADIUS study, a large randomized trial of more than 15,000 women, reported a

relative detection rate for fetal anomalies of 2.7 (95% confidence interval: 1.3–5.8) in tertiary compared to non-tertiary settings.[5]

In this study, detailed examination of fetal structures at 11-13.6 wks and 18-22 weeks of pregnancy revealed 40% of major structural abnormalities in low risk pregnant women. This result is similar to the results reported from other studies. In the study by C. Rydberg et al. from Sweden ,had a detection rate of fetal abnormalities of 40%, when detected before 22 completed weeks of gestation, and an additional 4%, when detected between 22 weeks and birth.[6]

Carvalho et al., (2002), reported about 38% of structural anomalies detectable in the first trimester.[7]Souka et al., (2006) and Dane et al (2007) had reported high detection rates of about 50% and 70% respectively. This high detection rate of structural abnormalities at 11-13.6 wks of gestation, reflects the fact that experienced obstetrician or radiologist has a major role and good training is mandatory with high resolution ultrasound machine .[8]

The introduction of routine first trimester scanning will have important implications for second trimester scan. Most of the chromosomal abnormality are detected in the first trimester scan (11 – 13 weeks) using nuchal translucency thickness and hence it is used for screening for Down's syndrome and other abnormalities.[9] Once it is found to be screen positive, the confirmatory test such as chorionic villus sampling, and amniocentesis are done. The chorionic villus sampling done at 11 to 13 weeks of gestation helps in earlier diagnosis of aneuploidy and early amniocentesis is no longer optimal at 11 – 13 weeks because of its higher association for fetal loss, fetal clubfoot, and procedure failure. Previously the cell culture and karyotype results took 2 to 3 weeks. But now the FISH technique helps in providing the karyotype results within 48 hrs. Hence early detection of fetal malformation allows early termination of malformed fetuses. Once an abnormality is diagnosed, parents will choose for elective termination. Prenatal diagnosis not only allows termination but also has profound implication on antenatal and intrapartum management such as in utero treatment of diaphragmatic hernia and other fetal therapies.[10]

Main limitation of early fetal anomaly scan is that the majority of abnormalities are difficult to identify, due to ongoing fetal development and delayed onset of some anomalies. Our study concluded that a higher percentage of abnormalities could be detected by early scan. But role of mid trimester scan is undisputed as some abnormalities could not be detected by early screening owing to late development of some fetal organs which is in concurrence with study conducted by Oztekin et al.T.[11]

Conclusion

1. The detection of the major anomalies at this early gestation (11-13 wks) offers to the parents the option of an earlier, safer and psychologically less traumatic termination of the pregnancy.
2. The ultrasound examination at 11-14 wks to screen for fetal abnormalities is effective and can be an adjunct to the routine 18-22 weeks anomaly scan.
3. Hence first trimester ultrasound (11-14wks) must be made mandatory, not only for the detection of major fetal anomalies, but also for diagnosing multiple pregnancy and abnormal pregnancy like missed abortion, molar pregnancy and ectopic pregnancy. Towards the end of the first trimester, the scan additionally offers an opportunity to identify gross fetal irregularities and, in health systems that offer

first trimester aneuploidy screening, the measuring of the nuchal translucency thickness (NT) is also available.

4. In general, screening for fetal structural and chromosomal abnormalities is a crucial part of antenatal care, the main purpose of a fetal ultrasound scan is to provide precise information that will simplify the delivery of enhanced antenatal care with the best possible outcomes for both the mother and fetus

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