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ORIGINAL ARTICLE

Importance of First Trimester Ultrasonography at 11-14 Weeks in Diagnosing Fetal Abnormalities vs. Scan at 20 Weeks in High-Risk Females

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ABSTRACT

Aim: To establish the role of 1st trimester ultrasound in detecting fetal abnormalities by performing a 20-week ultrasound scan as a standard.

Study Design: A cross-sectional descriptive study.

Methods: A total of 380 cases were recruited from the Obstetrics and Gynecology department of Jinnah post graduate medical center Karachi for six months duration from January, 2021 to June, 2021. The patients were assessed at 11-14 weeks of pregnancy for the diagnosis of fetal anomalies in high-risk women taking 20 weeks gestation ultrasound scan as the gold standard.

Results: Out of 380 women, mean age was 27.47 ± 2.57 years. 238 (62.6%) women underwent ultrasound at 12 + 13 weeks and 142 (37.4%) women had it at the 10-11 weeks of amenorrhea. Regarding maternal risk factors, 46 (12.1%) women had advanced maternal age> 35 years, 21 (5.5%) women with a family history of chromosomal abnormalities, 4 (2.1%) with a history of Down syndrome in previous babies, 118 (30.8)%) women with a history of congenital abnormalities, 85 (21.6%) women with a previous pregnancy complicated by spontaneous abortion, 11 (3.4%) women with a previous child with cerebral palsy, 56 (14.2%) women with previous intrauterine death, 13 (3.4%) women with a history of thalassemia, 7 (1.8%) women with a history of multiple pregnancies and 22 (5.5%) women with associated medical complications; diabetes, hypertension. The sensitivity, specificity and predictive value of ultrasound in the diagnosis of fetal defects in high-risk women compared to the ultrasound at week 20 showed 91.9% sensitivity, 100% specificity, 100% positive predictive value and negative predictive value is 96.27%, and the diagnostic accuracy is 99.2%.

Conclusion: First trimester ultrasound is effective in detecting fetal anomalies.

Keywords: First trimester ultrasound, fetal anomaly, high-risk women

INTRODUCTION

Ultrasound is considered safe in both the short and long term as it is a cost effective, non-invasive screening tool[1-2]. The detection potential of this screening modality especially in high risk women is reported to be around 85-90% **[3].** It reduces the rate of invasive testing by screening candidates without significantly reducing detection rates. A revolutionary technological achievement and the use of high-frequency ultrasound enabled detailed and accurate imaging of the structure of the fetus, such as fetal viability, dating, development, any chromosomal or structural abnormalities, and multiple pregnancy, even in later pregnancy at 13 weeks[4-5]. Most (80%) of common fetal malformations develop before 12 weeks of gestation; therefore, good fetal visualization at this stage should be able to detect these malformations. It will ensure early detection of fetal abnormalities and establishing a family history of the genetic syndrome. Congenital anomalies are observed in 5% of children[6-7]. In the United States, it is one of the leading causes of infant morbidity and mortality. High-risk women with at least one risk factor for maternal and fetal poor outcome have a higher rate of fetal malformation compared to the general population[8-9]. The rate of detection of serious prenatal abnormalities by ultrasound is 95%, of which 70% can be detected in the first trimester of pregnancy. The Den Hollander et al report on the detection of structural defects in the fetus shows an 11% risk in high-risk women with a first trimester screening sensitivity of 82% and a second trimester screening sensitivity of 100%[10-11]. Both 11-14 weeks of ultrasound are used to detect abnormalities. It shows 99.98% specificity parallel to scan at 20 weeks. Sequential screening may be of benefit to low-risk pregnancies following a reported first trimester screening result, who may benefit from the higher detection rate achieved with additional second trimester screening despite invasive procedures[12-13]. However, high-risk women may benefit from first-trimester detection of the abnormalities, helping them to make an earlier decision about undergoing invasive diagnostic tests[14] and medically indicated termination of pregnancy if necessary.

Nearly comparable screening results for 18-22 weeks and screening at 10-14 can reduce fetal morbidity and mortality related to the possibility of early termination of pregnancy, safer and more cost-effective than methods available in late pregnancy[15]. Nowadays, with a greater proportion of women with delayed child birth and shorter reproductive windows, the increased pressure for a successful outcome becomes more important for obstetricians and their patients[16]. Therefore, there is an urgent need to assess the diagnostic ability of the first trimester anatomical examination to determine its role in the current screening paradigm.

PATIENTS AND METHODS

This cross-sectional descriptive study was conducted at the Obstetrics and Gynecology department of Jinnah post graduate medical center for six months duration from January, 2021 to June, 2021. 380 cases were recruited and the patients were assessed at 11-14 weeks of pregnancy for the diagnosis of fetal anomalies in high-risk women taking 20 weeks gestation ultrasound scan as the gold standard.

All high-risk women with amenorrhea at weeks 11-14 are included. Non-consenting and unsupervised patients were excluded from the study. Patients were referred to the ultrasound department for the transabdominal examination. Experienced operator performed all scans after counselling and obtaining written consent. The ultrasound was performed on a Color Doppler Sonoscape S22 machine and Toshiba Xario 100 using a 2.5 to 3.5 MHz probe. Data were analysed by SPSS 21.0. To assess the diagnostic accuracy of the scan around 11 to 14 weeks to detect abnormalities; screening was performed and sensitivity was assessed; Then, after 20 weeks, a detailed examination was carried out for the abnormalities.

RESULTS

Out of 380, 112 women (29.5%) were aged 31-35, the minimum number of patients were in the 15-20 years of age group 23(6.1%), and the mean age was 27.47 ± 2.57 years (Table 1).

Table 1: Distribution of cases by age (n = 380)

Age (years)	No.	%
15-20	23	6.1
21-25	97	25.5
26-30	102	26.8
31-35	112	29.5
36-40	46	12.1
Total	380	100

140 (37.4%) women who underwent ultrasound at 12 - 13 weeks and 35 (9.2%) women at the 10-11 weeks of amenorrhea. (Table 2).

Table 2: Distribution of cases by gestational age (n = 380)

Gestational age (weeks)	No.	%
10 – 11	35	9.2
11^{+1} – 12	104	27.4
12^{+1} – 13	142	37.4
13^{+1} – 14	99	26

Regarding maternal risk factors, 46 (12.1%) women with advanced maternal age> 35 years, 21 (5.5%) women with a family history of chromosomal abnormalities, 4 (1.05%) women with a history of Down syndrome, 117 (30.7)%) women with a history of congenital abnormalities, 85 (22.3%) women with a previous pregnancy complicated by spontaneous abortion, 12 (3.1%) women with a previous child with cerebral palsy, 54 (14.2%) women with previous intrauterine death, 13 (3.4%) women with a history of thalassemia, 7 (1.8%) women with a history of multiple pregnancies and 21 (5.5%) women with associated medical complications; diabetes, hypertension (Table 3).

Out of 380 women fetal anomalies were detected in 37 (9.73%) women. 10 (2.6%) anencephaly, 2 (0.5%) Encephalocele, 5 (1.3%) cystic hygroma, 2 (0.5%) omphalocele with increased nuchal translucency, 3 (0.8%) of fetuses with gastroschisis, Megacystis (0.5%), polycystic kidney disease 1 (0.3%) and 1 (0.3%) cardiac anomalies. The total number of anomalies detected was 37 (9.73%). The total number of healthy fetuses was 343 (90.2%).

Table 3: Distribution of cases by maternal risk factors (n=380)

Maternal risk factors	No.	%
Advanced maternal age>35yr	46	12.1
Family history of chromosomal anomalies	21	5.5
Previous history of Down's syndrome	4	1.05
Previous history of any congenital anomaly	117	30.7
Previous pregnancy complicated by miscarriage	85	22.3
Previous child with cerebral palsy	12	3.1
Previous history with IUFD, still birth, ENND	54	14.2
Previous history of Thalassemia	13	3.4
Multiple pregnancy	7	1.8
Medical complications; diabetes, Hypertension	21	5.5

Table 4: Distribution of cases by fetal anomalies (n = 380)

Fetal anomalies	No.	%
Anencephaly	10	2.6
Encephalocele	2	0.5
Cystic hygroma	5	1.3
Omphalocele + inc. NT	2	0.5
Gastroschisis	3	0.8
Hydrops	1	0.3
Holoprosencephaly	1	0.3
Hydrocephalus	1	0.3
Anencephaly+spina bifida	5	1.3
Cystic hygroma+intra abdominal cyst	3	0.8
Megacystis	2	0.5
polycystic kidney disease	1	0.3
Cardiac anomaly	1	0.3
Anomaly not detected	343	90.2

The sensitivity, specificity and predictive value of ultrasound in the diagnosis of fetal defects in high-risk women compared to the ultrasound at week 20 showed 91.9% sensitivity, 100% specificity, 100% positive predictive value and negative predictive value is 96.27%, and the diagnostic accuracy is 99.2% (Table 5).

Table 5: Diagnostic accuracy of the first trimester scan in fetal anomaly detection

Anomaly at 1 trimester USG	Anomaly at 2 trimester USG		Total
	Present	Absent	
Present	34	0	34
Absent	3	343	346
Total	37	343	380

DISCUSSION

Pakistan is currently the fifth most populous country in the world. With the high incidence of abnormalities, high-risk women have more women with abnormal fetuses and poor fetal outcomes for morbidity and mortality[15]. This causes anxiety for parents and burdens the health care system. To reduce maternal morbidity and mortality, follow a timely diagnosis and management plan. The age of women during pregnancy varies greatly in different parts of the world. An age range from 15 to 40 years was observed in this study,

with a mean age of 27.47 ± 2.57 years. Shariq S, et. Al reported the mean age of the mother was 30.98 years (18-42 years) in a study conducted in Liaquat National Hospital Karachi on prenatal diagnosis of fetal anomalies.[16] The gestational age at which screening for maximum abnormalities in this study was conducted at 12 -13 weeks (37.4%). However, the median gestational age in the Aga Khan University study was 12.4 weeks. The results can be compared with the domestic literature[17-18]. In our study, Regarding maternal risk factors, 46 (12.1%) women with advanced maternal age> 35 years, 21 (5.5%) women with a family history of chromosomal abnormalities, 8 (2.1%) women with a history of Down syndrome, 117 (30.8)%) women with a history of congenital abnormalities, 82 (21.6%) women with a previous pregnancy complicated by spontaneous abortion, 13 (3.4%) women with a previous child with cerebral palsy, 54 (14.2%) women with previous intrauterine death, 13 (3.4%) women with a history of thalassemia, 7 (1.8%) women with a history of multiple pregnancies and 21 (5.5%) women with associated medical complications; diabetes, hypertension. In a study conducted at Aga Khan University in Karachi, all these factors of motherhood were taken into account. As for fetal anomalies, the most common anomaly of all anomalies was skull defects; anencephaly (frequency 2.6%). Everything was detected on ultrasound in the first trimester[19-20]. Abnormalities that are not 100% detectable in the first trimester of pregnancy include polycystic kidney disease, anencephaly with spina bifida, hydrocephalus, cystic + abdominal cyst, megacystitis, and heart abnormalities. Therefore, it points to a lower sensitivity of first trimester screening in detecting these abnormalities. In this study, the sensitivity, specificity and predictive value of ultrasound in the diagnosis of fetal defects in high-risk women compared to the ultrasound at week 20 showed 91.9% sensitivity, 100% specificity, 100% positive predictive value and negative predictive value is 96.27%, and the diagnostic accuracy is 99.2%. Lee et al and Whitworth et al. There was 78.7% sensitivity, 99.94% specificity, 97.27% positive predictive value, 99.38% negative predictive value and 97% diagnostic accuracy[21-22]. The results can be compared with the international literature[23].

CONCLUSION

First trimester ultrasound plays an active role in detecting fetal defects in high-risk women; however, the second trimester ultrasound scan (18 to 20 weeks) should not be omitted.

REFERENCES

1. Yagel S, Achiron R, Ron M, Revel A, Antehy E. Transvaginal ultrasonography at early pregnancy cannot be used alone for targeted organ ultrasonographic examination in a high-risk population. American journal of obstetrics and gynecology. 1995 Mar 1;172(3):971-5.
2. Neilson JP. Ultrasound for fetal assessment in early pregnancy. Cochrane Database of Systematic Reviews. 1998(4).
3. Todros T, Capuzzo E, Gaglioti P. Prenatal diagnosis of congenital anomalies. Images in paediatric cardiology. 2001 Apr;3(2):3
4. Whitlow BJ, Chatzipapas IK, Lazanakis ML, Kadir RA, Economides DL. The value of sonography in early pregnancy for the detection of fetal abnormalities in an unselected population. BJOG: An International Journal of Obstetrics & Gynaecology. 1999 Sep;106(9):929-36.
5. Comstock CH, Love Jr JJ, Bronsteen RA, Lee W, Vettraino IM, Huang RR, Lorenz RP. Sonographic detection of placenta accreta in the second and third trimesters of pregnancy. American journal of obstetrics and gynecology. 2004 Apr 1;190(4):1135-40.
6. Comstock CH, Love Jr JJ, Bronsteen RA, Lee W, Vettraino IM, Huang RR, Lorenz RP. Sonographic detection of placenta accreta in the second and third trimesters of pregnancy. American journal of obstetrics and gynecology. 2004 Apr 1;190(4):1135-40.
7. Economides DL, Braithwaite JM. First trimester ultrasonographic diagnosis of fetal structural abnormalities in a low risk population. BJOG: An International Journal of Obstetrics & Gynaecology. 1998 Jan;105(1):53-7.
8. Chitty LS, Hunt GH, Moore J, Lobb MO. Effectiveness of routine ultrasonography in detecting fetal structural abnormalities in a low risk population. British Medical Journal. 1991 Nov 9;303(6811):1165-9.
9. Bromley B, Lieberman E, Benacerraf BR. The incorporation of maternal age into the sonographic scoring index for the detection at 14–20 weeks of fetuses with Down's syndrome. Ultrasound in Obstetrics and Gynecology: The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology. 1997 Nov;10(5):321-4.
10. Souka AP, Krampl E, Bakalis S, Heath V, Nicolaides KH. Outcome of pregnancy in chromosomally normal fetuses with increased nuchal translucency in the first trimester. Ultrasound in Obstetrics and Gynecology: The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology. 2001 Jul;18(1):9-17.
11. Chitty LS, Pandya PP. Ultrasound screening for fetal abnormalities in the first trimester. Prenatal Diagnosis: Published in Affiliation With the International Society for Prenatal Diagnosis. 1997 Dec;17(13):1269-81.
12. QAISRANI HG, HUSSAIN MA, ALI M. Role of 1 st Trimester Ultrasonography in Detecting Fetal Abnormalities versus 20 Weeks Scan in High Risk Women.
13. Eleftheriades M, Tsapakis E, Sotiriadis A, Manolakos E, Hassiakos D, Botsis D. Detection of congenital heart defects throughout pregnancy; impact of first trimester ultrasound screening for cardiac abnormalities. The Journal of Maternal-Fetal & Neonatal Medicine. 2012 Dec 1;25(12):2546-50.
14. Hendler I, Blackwell SC, Bujold E, Treadwell MC, Wolfe HM, Sokol RJ, Sorokin Y. The impact of maternal obesity on midtrimester sonographic visualization of fetal cardiac and craniospinal structures. International journal of obesity. 2004 Dec;28(12):1607-11.
15. Sonek J. First trimester ultrasonography in screening and detection of fetal anomalies. InAmerican Journal of Medical Genetics Part C: Seminars in Medical Genetics 2007 Feb 15 (Vol. 145, No. 1, pp. 45-61). Hoboken: Wiley Subscription Services, Inc., A Wiley Company.
16. uz Zaman M. Abstracts of 33rd RSP Conference (27-29th October 2017) Karachi, Pakistan. PJR. 2017 Oct 20;27(4).
17. Hernádi L, Töröcsik M. Screening for fetal anomalies in the 12th week of pregnancy by transvaginal sonography in an unselected population. Prenatal Diagnosis: Published in Affiliation With the International Society for Prenatal Diagnosis. 1997 Aug;17(8):753-9.
18. Syngelaki A, Hammami A, Bower S, Zidere V, Akolekar R, Nicolaides KH. Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks' gestation. Ultrasound in Obstetrics & Gynecology. 2019 Oct;54(4):468-76.
19. Papp Z, Tóh-Pál E, Papp CS, Tóz Z, Szabó M, Veress L, Torok O. Impact of prenatal mid-trimester screening on the prevalence of fetal structural anomalies: a prospective epidemiological study. Ultrasound in Obstetrics and Gynecology: The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology. 1995 Nov 1;6(5):320-6.
20. Nicolaides KH, Azar G, Byrne D, Mansur C, Marks K. Fetal nuchal translucency: ultrasound screening for chromosomal defects in first trimester of pregnancy. British medical journal. 1992 Apr 4;304(6831):867-9.
21. Souka AP, Nicolaides KH. Diagnosis of fetal abnormalities at the 10–14-week scan. Ultrasound in Obstetrics and Gynecology: The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology. 1997 Dec;10(6):429-42.
22. Grande M, Arigita M, Borobio V, Jimenez JM, Fernandez S, Borrell A. First-trimester detection of structural abnormalities and the role of aneuploidy markers. Ultrasound in obstetrics & gynecology. 2012 Feb;39(2):157-63.
23. Timor-Tritsch IE, Fuchs KM, Monteagudo A, D'alton ME. Performing a fetal anatomy scan at the time of first-trimester screening. Obstetrics & Gynecology. 2009 Feb 1;113(2):402-7.