# Diagnostic Accuracy of Early Ultrasound for Congenital Anomalies

Maham Riaz, Uzma Manzoor, Hudabia Anwar, Ayesha Khalid

## ABSTRACT

Detection of fetal anomalies on antenatal scan is very important as it may help the couple to decide whether they want continuation of pregnancy or termination and it also helps them to get the delivery in appropriate settings suitable for the resuscitation and management of the abnormal baby. Early detection even before 20 weeks if reliable may help reduce the anxiety of couple having previous history of congenital anomalies and early management with lesser morbidity. **Objective:** To find the diagnostic accuracy of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard. **Study Design:** The study adopted a cross-sectional design which consisted of validation. **Setting:** The study is conducted in "Fatima Ward, Gynaecology & Obstetrics Department, Allied Hospital, Faisalabad" in collaboration with "Allied Hospital, Faisalabad Radiology Department". **Duration of Study:** Six months after the approval of research proposal. **Period:** August 2015 to January 2016. **Methodology:** After the approval of the study by the ethical committee of the Allied Hospital and written informed consent, the cases fulfilling the inclusion criteria were registered through OPD of Allied Hospital, Faisalabad. Patients were counseled and referred to ultrasound department of this same hospital, for trans-abdominal scan. A single operator performed all the scans after obtaining a verbal consent. Ultrasound was done on the Siemens machine using 2.5 – 3.5 MHz probe. To assess the diagnostic accuracy of 11 – 14 weeks scan, for anomaly detection; the screening was done and sensitivity was evaluated; patients were followed by taking contact numbers and a detailed anomaly scan at 20 weeks was done. **Results:** In our study, out of 611 cases, 22.91%(n=140) were between 18-25 years of age while 77.09%(n=471) were aged between 26-35, with a mean$\pm$sd value of 28.33$\pm$3.71 years, the occurrence of fetal anomalies (on gold standard) was documented in 9% presenting (n=55). However, 91% respondents (n=556) had no morbidity. The Diagnostic precision of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard was calculated, it is showing that 7.36%(n=45) had true positive, while 1.80%(n=11) presented a false +ive outcome. 89.20% (n=545) had true -ive and 1.64% (n=10) had false -ive, sensitivity is calculated as 81.80%, specificity is calculated as 98.02%, positive predictive value (PPV) is calculated as 83.35%, negative predictive value (NPV) is calculated as 98.19% and overall accuracy rate is calculated as 96.56%. **Conclusion:** We finalized that the diagnostic accuracy of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard is higher and useful in our population.

**Keywords:** Fetal anomalies, high risk women, 11-14 weeks ultrasound, diagnostic accuracy

*Corresponding Author*
**Dr. Maham Riaz**
Women Medical Officer, Children Hospital, Jhang Road, Faisalabad
Contact: +92 332-8663075
Email: maham_aquarius@hotmail.com

## INTRODUCTION

In late 1970's in UK, the ultrasound was introduced as a screening tool for fetal abnormalities, in many maternity centers and became routine afterwards.[1] It is considered safe in both short and long terms being non-invasive, cost-effective screening tool. It reduces the invasive testing rate by the selection of candidates without significantly reducing the detection rates. Revolutionary technological success and use of high frequency scan has made the detailed and precise fetal structure visualization possible[2] like fetal viability, dating, development, any chromosomal or structural anomaly and multiple gestations even at earlier gestation of 12-13 weeks. The great number (80%) of common fetal malformations begin before 12 weeks gestation;

therefore a good envision of the fetus at this stage should be able to investigate these malformations.[3] It will offer earlier screening of fetal anomalies and identify a family history of genetic syndromes.

Congenital anomalies are seen in 6.5% of the babies born[4] and in the united states are a foremost cause of infant morbidity and mortality accounting for 20% of all infant deaths.[5] There is increased rate of fetal anomalies in high risk women having one or more risk factors of poor maternal and fetal outcomes as compared to the general population. Antenatal major anomalies detection rate by ultrasound is found to be 95% out of which 70% can be detected in first trimester.[6] Den Hollander et al report of detection rate of fetal structural defects shows a sensitivity of 82% for 1st trimester scan and 100% for second

trimester scan. Thereby, indicating almost comparable results.[3] The ultrasound in detecting anomalies is 99.98% specific at both 11-14 weeks and 20 weeks ultrasound.[3]

Sequential screening may be more advantageous in low risk pregnant women, who after the informed first trimester screening result, can still benefit from higher detection rate obtained with additional second trimester screening despite undergoing invasive procedures.[7] But high risk women may benefit from first trimester anomaly screening, helping them for earlier decision for having diagnostic invasive test.

Almost comparable results of 10-14 weeks scan for anomaly detection with 18-22 weeks scan may prove beneficial by early detection, intervention at tertiary health center, less psychological pressure due to reduced feto-maternal bonding at earlier gestation, reducing complications, timely referrals, decreased fetomaternal morbidity and mortality associated with early, safer, cost effective termination options than those available at advanced gestation.[8]

In modern age with greater proportion of women with delayed child birth and shortened reproductive window, the increased pressure for a successful outcome is of growing importance to the obstetricians and their patients. Thus there is an high priority need to assess the diagnostic ability of a first trimester anatomic review to determine its role in the present day screening paradigm.[9]

The rationale of my study is that in our country where we have less privileged society and most of women can't afford ultrasound so often and therefore lost to follow up after a booking visit and will be found absent at the time of routine anomaly scan at 22weeks, to present only at or near term when anomaly detection and therefore termination will cause more physical and psychological trauma, increased maternal morbidity and a sign of social stigma. So by stressing 1st trimester scan, we may get advantage of early detection of fetal abnormalities in high risk people of our society.

To the best of my knowledge, no local data is available regarding this important aspect, and very little literature available yet, so it needs immediate attention to be paid.

## METHODOLOGY
**Study Design:** Cross sectional study(validation)
**Setting:** Gynaecology and obstetrics dept. in collaboration with Radiology Dept., Allied Hospital, Faisalabad.
**Duration of Study:** After the approval of synopsis It took 6 months from August 2015 to January 2016.

**Sample Size:**
- Sensitivity and specificity after using Who sample size calculator

Sensitivity =82%[3], Specificity=99%[3], Prevalence= 6.5%[4], Precision for sensitivity=10%, Precision for specificity=1%, Confidence level=95%, Sample size=611

**Sample Technique:**
We adopted Non-probability sampling which is purposive.

**Sample Selection:**
**Inclusion Criteria:**
Women of all age group with gestational amenorrhea of 11 – 14 weeks with any risk factor (as mentioned in operational definition).

**Exclusion Criteria:**
Multiple pregnancy

**Data Collection:**
After the approval of the study by the ethical committee of the Allied Hospital and written informed consent, the cases fulfilling the inclusion criteria were registered through OPD of Allied Hospital, Faisalabad. Patients were counseled and referred to ultrasound department of this same hospital, for trans-abdominal scan. A single operator performed all the scans after obtaining a verbal consent. Ultrasound was done on the Siemens machine using 2.5 – 3.5 MHz probe. To assess the diagnostic accuracy of 11 – 14 weeks scan, for anomaly detection; the screening was done and sensitivity was evaluated; patients were followed by taking contact numbers and a detailed anomaly scan at 20 weeks was done. All this information was recorded in a pre-designed performa attached by myself.

**Data Analysis:**
The information was recorded in SPSS version 17 and scrutinized through it. The quantitative variables analyzed included demographic characteristics like age and was presented as Mean+/- SD. The qualitative variables; true positives were presented in the form of occurrence and percentile. Other data analysis tools are: Sensitivity, specificity, +ive predictive value, -ive predictive value and diagnostic accuracy of 11-14 weeks scan in the of fetal anomalies in high risk women was calculated by generating a 2x2 contingency table by taking 20 weeks scan as gold standard.

| 20 weeks scan | | Positive | Negative |
|---|---|---|---|
| **11-14 weeks scan** | Positive | a (TP) | b (FP) |
| | Negative | c (FN) | d (TN) |

Sensitivity = $\frac{a}{a+c}$ × 100

Specificity = $\frac{d}{b+d}$ × 100

PPV (Positive Predictive Value) = $\frac{a}{a+b}$ × 100

NPV (Negative Predictive Value) = $\frac{d}{c+d}$ × 100

Diagnostic Accuracy = $\frac{a+d}{a+b+c+d}$ × 100

## RESULTS

The total of 611 cases satisfying the inclusion or exclusion principles was entered to investigate the diagnostics accuracy of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard.

Patients were distributed according to gender showing that 22.91%(n=140) were between 18-25 years of age while 77.09%(n=471) were aged between 26-35 yrs, with a mean±sd was calculated as 28.33±3.71 years. (Table No. 1)

### Table 1: Age of patients (n=611)

| Age (in yrs) | Patients | % |
|---|---|---|
| 18-25 | 140 | 22.91 |
| 26-35 | 471 | 77.09 |
| **Total** | **611** | **100** |
| **mean±sd** | **28.33±3.71** | |

Incidence of fetal anomalies (on gold standard) was recorded in 9%(n=55) while 91%(n=556) had no findings of the morbidity. (Table No. 2)

### Table 2: Frequency of fetal anomalies (On gold standard) (n=611)

| Fetal anomalies | Patients | Percentage |
|---|---|---|
| Yes | 55 | 9 |
| No | 556 | 91 |
| **Total** | **611** | **100** |

### Table 3: Diagnostic accuracy of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard (n=611)

| 11-14 Weeks Ultrasound | 20 Weeks Ultrasound | | Total |
|---|---|---|---|
| | **Positive** | **Negative** | |
| Positive | True positive(a) 45 (7.36%) | False positive (b) 11 (1.80%) | a + b 56(9.16%) |
| Negative | False negative(c) 10 (1.64%) | True negative (d) 545 (89.20%) | c + d 555 (90.84%) |
| **Total** | **a + c 55 (9%)** | **b + d 556(91%)** | **611 (100%)** |

Sensitivity = a / (a + c) x 100 =81.81%
Specificity = d / (d + b) x 100 = 98.02%
+ive predictive value = a / (a + b) x 100 =80.35%
-ive predictive value = d / (d + c) x 100 =98.19%
Accuracy rate = a + d / (a + d + b + c) x 100= 96.56%

The Diagnostic precision of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard was calculated, it is showing that 7.36% (n=45) had true positive, while 1.80%(n=11) presented a false +ive outcome. 89.20% (n=545) had true -ive and 1.64% (n=10) had false -ive, sensitivity is calculated as 81.80%, specificity is calculated as 98.02%, positive predictive value (PPV) is calculated as 83.35%, negative predictive value (NPV) is calculated as 98.19% and overall accuracy rate is calculated as 96.56%.

## DISCUSSION

Detection of fetal anomalies on antenatal scan is very important as it may help the couple to decide whether they want continuation of pregnancy or termination and it also helps them to get the delivery in appropriate settings suitable for the resuscitation and management of the abnormal baby. Early detection even before 20 weeks if reliable may help reduce the anxiety of couple having previous history of congenital anomalies and early management with lesser morbidity.

The principle of this study was that in our country where we have less privileged society and most of women can't afford ultrasound so often and therefore lost to follow up after a booking visit and may not present at the time of routine anomaly scan at 22weeks, to present only at or near term when anomaly detection and therefore termination may cause more physical and psychological trauma,

increased maternal morbidity and a sign of social stigma. So by stressing 1st trimester scan, we may get advantage of initial detection of fetal abnormalities in high risk people of our society. To the best of our knowledge, no local data is available regarding this important aspect, and very little literature available yet, so it needs immediate attention to be paid.

In our work, out of 611 cases, 22.91%(n=140) were between 18-25 years of age while 77.09%(n=471) were between 26-35 years of age, mean$\pm$sd was calculated as 28.33$\pm$3.71 years, frequency of fetal anomalies (on gold standard) was recorded in 9%(n=55) while 91% (n=556) had no findings of the morbidity. Diagnostic precision of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard was calculated, it is showing that 7.36%(n=45) had true positive, while 1.80%(n=11) presented a false +ive outcome. 89.20% (n=545) had true -ive and 1.64% (n=10) had false -ive, sensitivity is calculated as 81.80%, specificity is calculated as 98.02%, positive predictive value (PPV) is calculated as 83.35%, negative predictive value (NPV) is calculated as 98.19% and overall accuracy rate is calculated as 96.56%.

The conclusion of our study is in agreement with Den Hollander et al report of detection rate of fetal structural defects show a sensitivity of 82% for 1st trimester scan and 100% for second trimester scan. Thereby, indicating almost comparable results.[3] The ultrasound in detecting anomalies is 99.98% specific at both 11-14 weeks and 20 weeks ultrasound.[3]

Prior studies reported the sensitivity 78.7%, specificity 99.94%, positive predictive value 97.27%, negative predictive value 99.38% and diagnostic accuracy 97%. The results are comparable with the international literatures.[10-12]

Another study evaluated the accuracy of first-trimester ultrasound analysis in identifying considerable congenital heart disease (CHD) using a systematic evaluation of the literature and recorded that Ten studies (involving 1243 patients) were appropriate for inclusion. Of these, four used transabdominal ultrasonography, four used transvaginal and two used combo of both. Combined sensitivity and specificity were 85% (95% CI, 78–90%) and 99% (95% CI, 98–100%), respectively. They concluded that ultrasound examination of the fetus in the first trimester is convenient for accurately detecting major CHD. It may be offered to women at high risk of having children with CHD.[13-16]

However, the findings of our study are in agreement with other studies and we are of the view that 1st trimester scan has advantage of early detection of fetal abnormalities in high risk people of our society and every high risk pregnant women should be screened.

## CONCLUSION

We concluded that the diagnostic accuracy of 11-14 weeks ultrasound in fetal anomalies detection among high risk women taking 20 weeks scan as gold standard is higher and useful in our population.
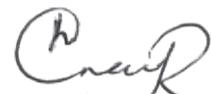
## REFERENCES

1. Patahak S, Lees C. Ultrasound structural fetal anomaly screening. an update. Arch Dis Child Fetal Neonatal Ed. 2009;94:384-90.
2. Ebrashy A, El-Kateb A, Momtoz M, El-Sheikhah A, Ibrahim M. 13-14 fetal anatomy scan a 5-year perspective study. Ultrasound Obstet Gynaecol 2010;35:292-6.
3. Nanhorngue' K, Ville Y. First-trimester screening for fetal abnormalities. In. Philip JS, Carolene AC, Carl PW, Stephen CR, Gonik B. High risk pregnancy. Management options. 4th ed. China. Elseveir Saunders 2011;97-119.
4. Levi S. Ultrasonographic screening for fetal malformations:www.obygyn.net/articles/ultrasonographic-screening-fetal-malformations.
5. Methew TJ, MacDorman MF. Infant mortality statistics from the 2008 period linked birth/infant death data set. National Vital Statistics Rep. 2012;60:1-27.
6. Abu-Rustum RS, Daou L, Abu-Rustum SE. Role of first trimester sonography in the diagnosis of aneuploidy and structural fetal anomalies. J Ultrasound Med. 2010;29.1445-52.
7. Yeo L, Anthony M, Vintzileos. Second –trimester screening for fetal abnormalities. In. Philip JS, Carolene AC, Carl PW, Stephen CR, Gonik B. High risk pregnancy. Management options. 4th ed. China. Elseveir Saunders, 2011;121-42.
8. Gagnon A, Wilson RD, Allen VM, Audibert F, Blight C, Brock JA. Evaluation of prenatally diagnosed structural congenital anomalies. J Obstet Gynaecol 2009;31:875-89.
9. Timor-tritsch IE, Fuchs KM, Monteagudo A, D' alton ME. Performing a fetal anatomy scan at the time of first-trimester screening. Obstet Gynecol 2009;113(2).402-7.
10. Harding ME. Maternity. In. Harding ME. The Way of All Women. New York, Harper and Row, 1975;160-70.
11. Deutsch H. Motherhood. In Deutsch. The Psychology of Women, New York, Grune and Stratton, 1945;2:144.
12. Merkatz R. Psychosocial support for women with pregnancy complications. In. Cherry S, Merkatz I (eds). Complications of Pregnancy. Medical, Surgical, Gynecologic, Psychosocial, and Perinatal, Baltimore, Williams and Wilkins. 1992;251.
13. Stoller R. Primary femininity. J Amer Psychoanal Assn 1976;24(5):59-78.

14. Gilligan C. Woman's place in man's life cycle. In Gilligan. In a Different Voice, Cambridge, Harvard University Press, 1982, pp 23.
15. Theut SK, Pedersen FA, Zaslow MJ, Rabinovich BA. Pregnancy subsequent to perinatal loss. Parental anxiety and depression. J. Am Acad Child Adolesc Psychiatry. 1989;27(3):289-92.
16. Friedman E. The graphic analysis of labor. Am J Obstet Gynecol 1954;68:1568.

## AUTHORSHIP AND CONTRIBUTION DECLARATION

| AUTHORS | Contribution to The Paper | Signatures |
|---|---|---|
| **Dr. Maham Riaz** <br> Women Medical Officer, Children Hospital, Jhang Road, Faisalabad | Revised the manuscript, Corresponding author | |
| **Dr. Uzma Manzoor** <br> Women Medical Officer, Gynae Department, Allied Hospital, Faisalabad | Idea generation and Literature review | |
| **Dr. Hudabia Anwar** <br> Women Medical Officer, Children Hospital, Jhang Road, Faisalabad | Worked on conclusion and Introduction | |
| **Dr. Ayesha Khalid** <br> Senior Registrar, Gynae Department, Allied Hospital, Faisalabad | Technical correction & Proof reading | |