

## Role of 1<sup>st</sup> Trimester Ultrasonography in Detecting Fetal Abnormalities versus 20 Weeks Scan in High Risk Women

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### ABSTRACT

**Aim:** To find the role of first trimester USG in detecting fetal anomalies by taking 20 weeks scan as standard.

**Methods:** This cross-sectional descriptive study was carried out at Gynaecology & Obstetrics, Lady Willingdon Hospital, Unit 3, Lahore from 4<sup>th</sup> April 2012 to 3<sup>rd</sup> Oct 2012. Six hundred and fifty cases were taken to evaluate the role of 11-14 weeks scan in the diagnosis of fetal abnormalities in high risk women by taking 20 weeks gestation as gold standard.

**Results:** Majority of the women were between 21-25 years, with the mean age of 28.40±2.35 years. Anomalies mostly were detected at 12<sup>+1</sup>-13 weeks. Prevalence of anomaly found is 11.69%. The most common anomaly detected was anencephaly. Cases by fetal anomalies includes, 21 (3.2%) fetuses with anencephaly, 5 (0.8%) encephalocele, 10 (1.5%) cystic hygroma, 5 (0.8%) omphalocele with increased nuchal translucency, 7 (1.1%) gastroschisis, 4 (0.6%) megacystitis, 3 (0.5%) multicystic kidney disease and 2 (0.3%) cardiac anomaly. Total anomalies detected were 75 (11.5%). The total number of normal fetuses is 575 (88.4%). Thirteen anomalous fetuses were not detected in the first trimester USG. Three fetuses were with multicystic kidney disease, three with anencephaly + spina bifida, one with hydrocephalus, two with cardiac anomalies, two cystic hygroma with intra-abdominal cysts and two with Megacystitis. Their detection percentage at first trimester corresponds to 0%, 57.1%, 50%, 0%, 33.3% and 50% respectively. Fetal anomaly detection through first trimester USG had a sensitivity of 82.66%, the specificity 100%, positive predictive value of first trimester scan 100%, negative predictive value of 97.78% and diagnostic accuracy of 98%.

**Conclusion:** First trimester USG is effective in detecting fetal anomaly.

**Key words:** First trimester USG, Fetal anomaly, High risk women

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### INTRODUCTION

Ultrasound 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 like fetal viability, dating, development, any chromosomal or structural anomaly and multiple gestations even at earlier gestation of 12-13 weeks.<sup>1</sup> The majority (80%) of common fetal malformations develops before 12 weeks gestation; therefore a good visualization of the fetus at this stage should be able to detect these malformations.<sup>2</sup> It will offer earlier screening of fetal anomalies and identify a family history of genetic syndromes. Congenital anomalies are seen in 5% of the babies born. In the United States are a leading cause of infant morbidity

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and mortality.<sup>3</sup> 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.<sup>4</sup> den Hollander et al<sup>5</sup> report of detection rate of fetal structural defects, show 11% risk in high risk women with first trimester scan sensitivity of 82% and second trimester scan sensitivity of 100%. The ultrasound in detecting anomalies is 99.98% specific at both 11-14 weeks and 20 weeks ultrasound.

Sequential screening may be more advantageous in low risk pregnant women, who after the informed first trimester screening result, can still take advantage of higher detection rate achieved with additional second trimester screening despite undergoing invasive procedures.<sup>6</sup> 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 fetomaternal

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.<sup>7</sup>

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 urgent need to evaluate the diagnostic ability of a first trimester anatomic survey to determine its role in the current screening paradigm<sup>8</sup>.

## PATIENTS AND METHODS

This cross-sectional descriptive study was carried out at Gynaecology & Obstetrics, Lady Willingdon Hospital, Unit 3, Lahore from 4<sup>th</sup> April 2012 to 3<sup>rd</sup> Oct 2012. Six hundred and fifty cases were taken to evaluate the role of 11-14 weeks scan in the diagnosis of fetal abnormalities in high risk women by taking 20 weeks gestation as gold standard. All high risk women with gestational amenorrhoea of 11-14 weeks were included. Non-cooperative and lost to follow-up patients were excluded from the study. Patients were counseled and referred to ultrasound department of this same Lady Willingdon 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. The data was analysed through SPSS 15. To assess the diagnostic accuracy of 11-14 weeks scan, for anomaly detection; the screening was performed and sensitivity was evaluated; a later detailed anomaly scan at 20 weeks was done.

## RESULTS

Majority of the women, 169 (26%) were between 21-25 years and minimum patients were between 15-20 years (9.7%) with the mean age of 28.40±2.35 years (Table 1). 272 (41.8%) women with USG done at 12<sup>+1</sup>-13 weeks and 70 (10.8%) women at 10-11 weeks of gestational amenorrhoea (Table 2). Regarding maternal risk factors include 132 (20.3%) women with advanced maternal age >35 years, 29 (4.5%) women with family history of chromosomal abnormalities, 13 (2%) women with previous history of Down's syndrome, 189 (29.1%) women with previous history of any congenital abnormality, 68 (10.5%) women with previous pregnancy complicated by miscarriage, 27 (4.2%) women with previous child affected with cerebral palsy, 108(16.6%) women with previous history of intrauterine death, early neonatal death or still birth, 28(4.3%) women with previous history of thalassemia, 14(2.2%) women with multiple pregnancy and 42(6.5%) women with medical

complication associated; diabetes, hypertension (Table 3). Table 4 showed the fetal anomalies, 21(3.2%) fetuses with anencephaly, 5(0.8%) encephalocele, 10(1.5%) cystic hygroma, 5(0.8%) omphalocele with increased nuchal translucency, 7(1.1%) gastroschisis, 4(0.6%) megacystitis, 3(0.5%) multicystic kidney diseases and 2 (0.3%) cardiac anomaly. Total anomalies detected were 75(11.5%). The total number of normal fetuses is 575 (88.4%). The sensitivity, specificity, and predictive value of ultrasound in diagnosis of fetal abnormalities versus 20th weeks scan in high risk women showed sensitivity 82.66%, specificity 100%, positive predictive value 100%, negative predictive value 97.78% and diagnostic accuracy 98% (Table 5).

Table 1: Distribution of cases by age (n = 650)

Age (years)	No.	%
15-20	63	9.7
21-25	169	26
26-30	116	17.8
31-35	154	23.7
36-40	148	22.8

Table 2: Distribution of cases by gestational age (n = 650)

Gestational age (weeks)	No.	%
10 – 11	70	10.8
11 <sup>+1</sup> – 12	179	27.5
12 <sup>+1</sup> – 13	272	41.8
13 <sup>+1</sup> – 14	129	19.8

Table 3: Distribution of cases by maternal risk factors (n=650)

Maternal risk factors	No.	%
Advanced maternal age>35yr	132	20.3
Family history of chromosomal anomalies	29	4.5
Previous history of Down's syndrome	13	2
Previous history of any congenital anomaly	189	29.1
Previous pregnancy complicated by miscarriage	68	10.5
Previous child with cerebral palsy	27	4.2
Previous history with IUFD, still birth, ENND	108	16.6
Previous history of Thalassemia	28	4.3
Multiple pregnancy	14	2.2
Medical complications; diabetes, Hypertension	42	6.5

Table 4: Distribution of cases by fetal anomalies (n = 650)

Fetal anomalies	No.	%
Anencephaly	21	3.2
Encephalocele	5	0.8
Cystic hygroma	10	3.2
Omphalocele + inc. NT	5	0.8
Gastroschisis	7	1.1
Hydrops	4	0.6
Holoprosencephaly	2	0.3
Hydrocephalus	2	0.3
Anencephaly+spina bifida	7	1.1
Cystic hygroma+intra abdominal cyst	4	0.6
Megacystis	4	0.6
MCKD	3	0.5
Cardiac anomaly	2	0.3
Anomaly not detected	574	88.3

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

Anomaly at 1 <sup>st</sup> trimester USG	Anomaly at 2 <sup>nd</sup> trimester USG		Total
	Present	Absent	
Present	62	0	62
Absent	13	575	588
Total	75	575	650

Sensitivity = 82.66%                      Specificity = 100%  
 Positive predictive value = 100%        Negative predictive value = 97.78%  
 Diagnostic Accuracy = 98%

**DISCUSSION**

Pakistan is the fifth most populous country in the world now. With high prevalence of anomalies in the high risk women, there is greater number of the women with the anomalous fetuses and poor fetal outcome, in terms of the morbidity and the mortality. This creates anxiety of the parents and burden on the health system. In order to reduce the perinatal morbidity and the mortality, it needs timely diagnosis and prompt management plan to be followed.

Age of the women during pregnancy varies greatly in different regions of the world. Present study observed the age range of 15-40 years, with the mean age of 28.40±2.35 years. In a study conducted at Agha Khan University Hospital, Karachi, mean maternal age was 30.98 years (18-42 years).<sup>9</sup> In the present study, the gestational age at which maximum anomaly scans were done was 12<sup>1+</sup>-13 weeks of gestation (41.8%). However, the mean gestational age at Agha Khan University study was 12.4 weeks.<sup>9</sup> The results are comparable with the national literature.

In my study, the distribution of cases according to the maternal risk factors include 132 (20.3%) women with advanced maternal age >35 years, 29(4.5%) women with family history of chromosomal abnormalities, 13 (2%) women previous of Down's syndrome, 189 (29.1%) women with previous history of any congenital abnormality, 68 (10.5%) women with previous pregnancy complicated by miscarriage, 27 (4.2%) women with previous child affected with cerebral palsy, 108 (16.6%) women with previous history of intrauterine death, early neonatal death or still birth, 28 (4.3%) women with previous history of thalassemia, 14 (2.2%) women with multiple pregnancy and 42 (6.5%) women with medical complication associated; diabetes, hypertension. And in the study conducted at Karachi, Agha Khan University, all these maternal factors were taken into account.

As far as the fetal anomalies were concerned, the most commonly detected anomaly among all was cranial defects; anencephaly (prevalence 3.2%). All

of it was detected in the first trimester scan. Hydrocephaly, holoprosencephaly and cardiac anomaly were detected as least prevalent (0.3%). The anomalies that were not detected as 100% in the first trimester scan include multicystic kidney disease, anencephaly with spina bifida, hydrocephalus, cystic hygroma+ intra-abdominal cyst, megacystitis and cardiac anomaly. Thereby, indicating less sensitivity of first trimester scan for detecting these anomalies.

In the present study, the true positive first trimester scans were 62 in number, true negative 575, false negative 13 and false positive as nil. The sensitivity, specificity, and predictive value of ultrasound in diagnosis of fetal abnormalities versus 20th weeks scan in high risk women showed sensitivity 82.66%, specificity 100%, positive predictive value 100%, negative predictive value 97.78% and diagnostic accuracy 98%. While Lee et al<sup>10</sup> and Whitworth et al<sup>11</sup> 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.

**CONCLUSION**

In the high risk women, the first trimester USG has an effective role in detecting fetal anomalies, however, the second trimester fetal anatomic survey USG (18–20 weeks) should not be abandoned.

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