Cairo University

Assessment of Fetal Heart in the First Trimester for Detection of Major Cardiac Anomalies

Thesis

Submitted for partial fulfillment of
M.D Degree in Obstetrics & Gynecology

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Faculty of Medicine,
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2014

بِسْمِ اللَّهِ الرَّحْمَنِ الرَّحِيمِ

[وَيَسْأَلُونَكَ عَنِ الرُّوحِ قُلِ الرُّوحُ مِنْ أَمْرِ رَبِّي وَمَا أُوتِيتُم مِّنَ العِلْمِ إِلاَّ قَلِيلا]

(سورة الإسراء، الآية: ٨)

Acknowledgment

Firstly and lastly thanks to Allah the most merciful for his help.

I would like to express my deepest gratitude and sincere appreciation to ***Prof. Alaa El Ebrashy***, Professor of Obstetrics & Gynecology, Director of Fetal Medicine Unit (CAIFM), Faculty of Medicine, Cairo University. I'm greatly thankful to him for his fatherly attitude, enthusiastic encouragement, his sincere advice, and supervision. He was very generous with his time and effort.

My extreme thanks and gratefulness to ***Prof. Sahar Shaker Sheta*** Professor of Pediatric Cardiology, Faculty of Medicine, Cairo University. She was very generous with her time and appreciable advice, which helped to simplify my hard task of preparing this work.

Also I would like to thank ***Prof. Mona Aboulghar***, Professor of Obstetrics and Gynecology, Faculty of Medicine, Cairo University for her keen interest, beneficial advice, constant support and scientific opinions during the conduction of this work. Also for her sincere supervision and the endless effort that she had offered throughout the period of making and revising this thesis.

I'm grateful to all my professors, staff and colleagues of the Fetal Medicine Unit (CAIFM) in the Obstetrics & Gynecology department, Cairo University for their help and cooperation throughout this work.

Many grateful thanks to my parents for their continuing love, care and strong support, to whom I'm indebted for what I am today.

Finally, I'm grateful to the patients for whom all our efforts are devoted.

Abstract

This study was carried out in Fetal Medicine Unit CAIFM in Obstetrics & Gynecology Department at Cairo University, from January 2012 till November 2013. Three hundred pregnant women at 11-13+6 weeks were included in this study. All included cases were subject to the following: explanation of the study and oral informed consent, full history taking, full first trimesteric anomaly scan with detailed heart examination and ductus venosus waveform assessment, then reassessment at 18-20 weeks followed by postnatal cardiac exam of the neonates with a neonatal 2D Echocardiography done if an anomaly is suspected sonographically or clinically.

Keywords:

FPR- ALARA-CAIFM-Anomalies

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List of Abbreviations

ALARA	As low as reasonably achievable
Ao	Aortic root
BMI	Body Mass Index
CHD	Congenital heart defects
DV	Ductus Venosus
FDA	Food and Drug Administration
FHR	Fetal heart rate
FPR	False positive rate
GA	Gestational Age
HS	Highly significant
IQR	interquartile range
IUFD	Intrauterine fetal death
IVC	Inferior vena cava
LA	Left atrium
LV	Left ventricle
MI	Mechanical Index
n	Number
NS	Non-significant
NT	Nuchal Translucency
P	Pulmonary artery
RA	Right atrium
S	Significant
S/D	Systole/Diastole
SD	Standard deviation
SVC	Superior vena cava
TA	Transabdominal
TOP	Termination of pregnancy
T1	First trimester
T2	Second trimester
TV	Transvaginal
T.V.S.	Transvaginal ultrasonography
USA	United States of America
VSD	Ventricular septum sefect
3VT	Three vessels Trachea view
2D	Two Dimensional

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Introduction

Abnormalities of the heart and great arteries are the most common congenital defects and they account for about 20% of all stillbirths and 30% of neonatal deaths due to congenital defects. **(Persico et al., 2011)**

Congenital heart disease (CHD) accounts for the majority of deaths from congenital defects in childhood, being six times more common than chromosomal abnormalities and four times more common than neural tube defects. Prenatal detection of specific cardiac anomalies such as complete transposition of the great arteries and hypoplastic left heart syndrome has been shown to improve neonatal morbidity and surgical outcome. **(Carvalho et al., 2002)**

Defects usually classified as major or critical are those that are lethal or require intervention in infancy or on long term follow up. **(Carvalho et al.,2002)**

There are several good reasons to attempt early CHD detection: differently from other organs, at this stage, the fetal heart is almost fully developed; compared with other malformations, CHD have a higher prevalence and can be associated with chromosome anomalies. **(Orlandi et al., 2014)**

Historically, detailed fetal echocardiography was performed because of a positive family history for CHD, but cardiac scanning has gradually been incorporated into routine ultrasound screening programs as routine first trimester screening of Down's syndrome is being a standard service in many countries**(Carvalho et al.,2002)**

Highly-skilled obstetricians have been performing (transvaginal) first-trimester fetal echo since the beginning of the 1990s, before cardiologists became interested in the fetal heart in early pregnancy. However, use of the transabdominal route for early scans and the ever

improving ultrasound resolution over the years has not only made it possible for cardiologists to explore the small first-trimester fetal heart but has also paved the way for sonographers, radiographers and other professionals to incorporate basic cardiac views into the routine 11 to 13 + 6 - week scan. **(Carvalho , 2010)**

The traditional method of screening for cardiac defects, which relies on family history of such defects, maternal history of diabetes mellitus and maternal exposure to teratogens, identifies only about 10% of affected fetuses. **(Chelemen et al., 2011)**

There is increasing evidence that, in certain cases of fetal cardiac and other structural anomalies, prenatal diagnosis may be helpful or even life-saving. **(Becker et al., 2006)**

If we accept that there is an obligation to detect diseases prior to delivery, we also have to accept that we inevitably diagnose conditions with a poor prognosis. The physiological and psychological impacts of a termination of pregnancy (TOP) increase with increasing gestational age, leading to ethical dilemmas in the second half of pregnancy, especially in cases of severely handicapped but viable fetuses. One main aim of prenatal diagnosis should be to provide as much relevant information as possible to the pregnant woman as early as possible. **(Becker et al., 2006)**

There is a very strong association between increased nuchal translucency and congenital cardiac malformations and various other genetic conditions; therefore, it is logical that after screening for Down's syndrome, full examination of the heart should be attempted. **(Chelemen et al., 2011)**

Recent reports have raised interest in ductus venosus Doppler variables, demonstrating the importance of the ductus venosus in first-trimester screening for fetal abnormalities. **(Teixiera et al., 2008)**

An abnormal ductus venosus blood flow velocity wave form is also associated with a high probability of underlying cardiac defects, and adverse outcomes such as intrauterine growth restriction, fetal anemia and twin–twin transfusion syndrome. **(Teixiera et al., 2008)**

Aim of the work

To assess the diagnostic efficacy of the first trimester cardiac anomaly scan at 11^{+0}-13^{+6} weeks gestation for detection of major cardiac anomalies compared to second trimester cardiac scan and post natal clinical examination with neonatal echocardiography when needed.

Nuchal Translucency

Nuchal Translucency (NT) is the normal fluid filled subcutaneous space between the back of the fetal skin and the overlying skin. An increased NT was first described as a measure greater than 95th percentile for a given crown rump length. However, reports have highlighted that adverse outcomes are much more common with an NT that exceeds a set threshold of 3.5mm, a measurement that essentially represents 99th percentile or more throughout the gestational age window for first trimester screening. **(Souka et al., 2005)**

Assessing the thickness of NT has become a well-established method in early pregnancy for detection of aneuploidy. **(Becker and Wegner, 2006)**

NT Measurement technique

Most experts recommend that NT should be measured between 11 and 13+6 weeks, corresponding to a CRL measurement of between 45 and 84 mm. This gestational age window is chosen because NT as a screening test performs optimally and fetal size allows diagnosis of major fetal abnormalities, thus providing women who are carrying an affected fetus with the option of an early termination of pregnancy. **(Salomon et al., 2013)**

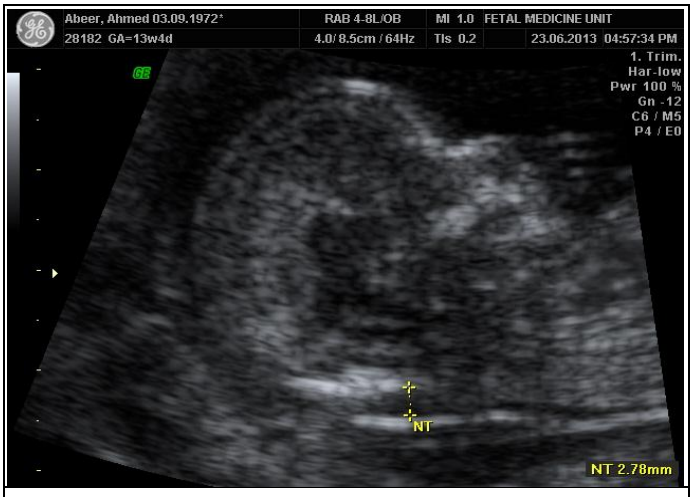

Figure (1): Nuchal Translucency = 2.78 mm

How to measure NT? (ISUOG Practice Guidelines, 2013)

NT can be measured by a transabdominal or transvaginal route. The fetus should be in the neutral position, a sagittal section should be obtained and the image should be magnified in order to include only the fetal head and upper thorax. Furthermore, the amniotic membrane should be identified separately from the fetus. The median view of the fetal face is defined by the presence of the echogenic tip of the nose and rectangular shape of the palate anteriorly, the translucent diencephalon in the center and the nuchal membrane posteriorly. **(Salomon et al., 2013)**

If the section is not exact median, the tip of the nose will not be visualized and the orthogonal osseous extension at the frontal end of the maxilla will appear. The ultrasound machine should allow measurement precision of 0.1 mm. Calipers should be placed correctly (on-on) to measure NT as the maximum distance between the nuchal membrane and the edge of the soft tissue overlying the cervical spine. If more than one measurement meeting all the criteria is obtained, the maximum one should be recorded and used for risk assessment. **(Salomon et al., 2013)**

NT use for screening anomalies

NT detection by ultrasound has emerged as a powerful prenatal screening strategy to diagnose a myriad of syndromes, but per se cannot be applied as a stand-alone benchmark in detecting structural and karyotyping related anomalies.**(Shaista, 2013)**

Screening for chromosomal abnormalities by NT identifies fetuses at increased risk of major anomalies, including cardiac defects, and encourages the early diagnosis of major anomalies such as anencephaly, holoprosencephaly, exomphalos and megacystis, even in the presence of normal NT. **(Huggon et al., 2002)**

NT use for screening cardiac anomalies

A major improvement in screening for cardiac defects came with the realization that many affected fetuses have increased NT thickness at 11–13 weeks' gestation.**(Teodora et al., 2011)**

The higher the NT thickness, the higher the prevalence of major heart abnormalities.If a heart defect is diagnosed in a fetus with increased NT, the risk of an unfavorable outcome is increased.**(Carvalho, 2004)**

A meta-analysis of studies examining the screening performance of NT thickness for the detection of cardiac defects in euploid fetuses reported that the detection rate was 23% for an NT cut-off of the 99th centile. **(Persico et al., 2011)**

The prevalence of congenital heart disease in areferral population (1/20) is higher than for families with one previously affected child (2% - 3%) (Higher prevalence for higher NT thickness).**(Carvalho, 2001)**

Diagnosis of a heart defect at the time of first trimester scan provides additional information that can reduce uncertainty in parents whose fetus has an increased NT thickness, since in experienced hands the false-positive rate is low. **(Haak et al., 2002b)**

The time between the NT measurement and the result of the chorionic villus sampling is known to be experienced as a difficult waiting period by the parents. Diagnosis of a heart defect in a chromosomally normal fetus gives parents more time to gain information about treatment and prognosis of the malformation. The early diagnosis also gives the opportunity, if it is the wish of the parents, to terminate the pregnancy at an earlier stage.**(Haak et al., 2002b)**

The emerging effects and possible pathogenic mechanisms of enlarged NT include fetal heart failure secondary to a cardiac defect, anemia, infection, inappropriate expression of atrial natriuretic peptide;