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ORIGINAL ARTICLE

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The potential of first trimester anomaly scan and first trimester fetal echocardiography as screening procedures in a medium risk population

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OBJECTIVE(S) : To assess the diagnostic efficacy of First Trimester Anomaly Scan (FTAS) and First Trimester Fetal Echocardiography (FTFE) as screening procedures in a medium risk population

METHOD(S) : In this retrospective study, we evaluated the diagnostic efficacy of FTAS in 6423 singleton pregnancies with a crown rump length (CRL) of 38-84 mm and First Trimester Fetal Echocardiography (FTFE) in 7702 fetuses of 45-84 mm CRL.

RESULTS : The prevalence of major abnormalities was 3.7% (235/6423) with a nuchal fold of more than 2.5 mm in 47.7% (112/235) of the fetuses. The rate of detection or suspicion of major anomalies during FTAS was 67% (245/366). The prevalence of major congenital heart disease (CHD) in 7702 fetuses including multiple pregnancies was 1.4% (105/7702) with a nuchal fold of more than 2.5 mm in 29.5% of the fetuses (31/105). The rate of detection or suspicion of major CHD during FTFE was 80% (84/105).

CONCLUSION(S) : The potential of FTAS and FTFE is far beyond providing data to determine probabilities for trisomy 13, 18 and 21.

Key words : ultrasound, echocardiograph screening

Introduction

Nuchal translucency (NT) - diagnostic has become an established method. Several studies clearly indicate that at the end of first trimester, a NT may be related to chromosomal abnormalities, major cardiac defects or complex disorders. Up to now, the main goal of NT-diagnostic is to determine the individual risk of a pregnant woman for trisomy 13, 18 and 21 [1]. It is suggested that detailed assessment of fetal anatomy should be performed in pregnancies with increased risk for malformation [2]. This policy accepts that malformations in pregnancies without increased risk (younger women and fetuses without enlarged NT) are detected in late phases of pregnancy or even not at all.

The aim of our retrospective evaluation was to determine the diagnostic efficacy of First Trimester Anomaly Scan (FTAS) and First Trimester Fetal Echocardiography (FTFE) as routine measures in every patient of an unselected medium-risk population.

Methods

In 1996, we established the NT-scan in our institution to assess the probability of trisomy 13, 18 and 21. In addition to the measurement of CRL and NT, we integrated early anomaly scan which included examination of the fetal head, four limbs, urinary bladder, stomach, umbilicus and two umbilical arteries. Beginning 1997, we integrated routine fetal echocardiography into every early anomaly scan. The

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examination included the 4-chamber view in B-mode and the visualization of inflow and outflow tract in B-mode and color flow mapping. Optional additional steps were measurements of the ventricular width, the diameters of the AV-valves and of great vessels, and the ductus venosus flow. Sonographic equipments used were Acuson 128XP10, Siemens Acuson Sequoia and GE Voluson 730. Examinations were performed by four gynecologists specialized in prenatal diagnosis for at least 10 years.

For the evaluation of diagnostic efficacy of FTAS and FTFE, we performed two retrospective analyses evaluating a time period of 5 years from 1st January 1998 to December 2002.

- FTAS-group: We evaluated all scans performed in living singleton fetuses of 38 – 84 mm crown-rump-length (CRL). We performed 6845 early anomaly scans and were informed about the outcome of 6423 pregnancies (feedback rate 93.8%). Median age of the women was 35 years (Range 15-46 years).

- FTFE-group: We evaluated all cases of FTFE performed in 7702 fetuses of 45-84 mm CRL. This study included multiple pregnancies (145 sets of twins, 16 sets of triplets, and 2 sets of quadruplets). Median age of the women was 34 years (Range 15-47 years). Median of examination time was 12+4 weeks; 80% of examinations were performed between 11+3 and 13+3 weeks.

In cases of normal outcome, feedback was obtained by neonatal workup or by report of the women after delivery. In cases of anomalies, feedback was obtained either by autopsy, if available, or was based on the ultrasound diagnosis of at least two examiners.

Results

In the FTAS group, the prevalence of major anomalies was 3.7% (235/6423). The frequency of chromosomal or monogenic disorders was 2.0% (132/6423). The rate of detection or suspicion for major anomalies in the first trimester was 67.2% (158/235). Another 19.6% (46/235) of major anomalies were detected until 23+6 weeks and further 4.1% (12/235) later in pregnancy. 52.3% of fetuses with major anomalies (123/235) presented with ≤2.5mm of nuchal fold. In 19.2% (1233/6423), invasive procedures were performed (chorion villus sampling : n=876; amnioceulesis : n=353; fetal blood sampling : n=4). The uptake of invasive procedures was 9.5% (302/3194) in women younger than 35 years and 28.8% (931/3229) in women of 35 years or more. Following prenatal diagnosis, termination of pregnancy was performed in 2.7% of all cases (174/6423). 75.3% (131/174) of these terminations were done prior to 16 gestational weeks.

In the FTFE group, the prevalence of major cardiac anomalies was 1.4% (105/7702). Thirty-three of these had isolated heart disease, and in 72 the heart disease was associated with other anomalies. Eighty percent (84/105) of major congenital heart diseases (CHDs) were suspected or detected during the first trimester anomaly scan. Table 1 demonstrates type and detection time of major CHDs. The prevalence of chromosomal abnormalities in the group with karyotyped fetuses with major CHD was 71.2% (52/73) (Table 2). 29.5% of major CHD (31/105) presented with a NT of ≤2.5 mm. The detection rate for major CHD was 52% (16/31) in cases of ≤2.5 mm NT and 92% (68/74) in the cases of >2.5 mm NT.

Table 1. Type and detection time of major congenital heart disease in 105 of 7702 fetuses of the First Trimester Fetal Echocardiography group seen initially at 11+0 to 13 + 6 gestational weeks.

Type of congenital heart disease	Time of detection/suspicion				Total
	11+0 to 13 + 6 weeks Number	14 + 0 to 23 + 6 weeks Number	24+0 to birth weeks	After birth Number	
Atrio-ventricular septal defect	29	1		1	31
Hypoplastic left heart syndrome	5				5
Aortic stenosis / atresia +/- ventricular septal defect	15	3	1		19
Pulmonary stenosis / atresia +/- ventricular septal defect	6	2	1		9
Tricuspid atresia					
Ectopia cordis	5				5
Single ventricle	5	1			6
d-transposition of great arteries		4		1	5
Others	19	5		1	25
All CHDs	84	16	2	3	105
	(80%)	(15%)	(2%)	(3%)	(100%)

Table 2. Results of prenatal karyotyping of 84 fetuses with congenital heart disease detected at first trimester fetal echol cardiography.

Cytogenetic result	Number
Trisomy 21	21
Trisomy 18	16
Trisomy 13	7
Turner syndrome	8
Normal	21
Not examined	11
Total	84

Discussion

Traditionally, in prenatal medicine, detailed anomaly scan is performed in the second trimester. With growing experience of sonographers and improving equipment, detailed depiction of fetal structures including the fetal heart became possible [3,4]. Since more than a decade, there is growing evidence [5] that some of the major anomalies including major cardiac defects may already be detected at the end of the first trimester [6]. Meanwhile, there is growing acceptance that at least in high risk pregnancies, detailed FTAS and FTFE should be performed [2,7,8].

Table 3. Prevalence and detection rate of major anomalies in low-risk study groups.

Author	Gestational age (weeks)	Number	Prevalence	Detection rate
Hernadi and Torocsik [10]	11	3991	64 (1.6%)	35 (55%)
Economides and Breaithwaite [11]	12+0 to 13+6	1632	17 (1.0%)	11 (65%)
D'Ottavio et al [12]	14	4078	88 (2.5%)	54 (61%)
Carvalho et al [8]	11 to 14	2853	130 (4.6%)	29 (22%)
Taipale et al [9]	13 to 14	4855	33 (0.7%)	6 (18%)

Up to now, few studies are published focusing on the efficacy of routine application of these methods in larger study groups that are not at elevated risk (Table 3). Our retrospective single-center evaluation shows that the prevalence of major anomalies and major cardiac defects in the first trimester is remarkably high. This observation is not unexpected since fetuses with anomalies have a high probability of intrauterine demise. Our study emphasizes that a large number of major anomalies can be detected or suspected at the end of the first trimester [9], especially in a time period of 12+4 to 13+5 gestational weeks [10]. Thus, restricting the detailed sonography only to fetuses at high risk (e.g. >2.5 mm NT) leads to loss of essential information. It also shows that at the moment there are some anomalies that will be detected at the 21 week scan which may be missed at the FTAS, for example d-transposition of great arteries. Our study confirms the observation that an additional second trimester scan detects some of the major anomalies and cardiac malformations that are missed during first trimester scan [2].

Keeping the above mentioned data in mind, we follow the philosophy to do a predominantly *genetic* FTAS including FTFE which may detect a major part of severe anomalies with the potential consequence of termination of pregnancy. A second supportive scan performed at about 21 weeks of gestation has the potential of saving life (detection of placenta previa, pathological uterine blood flow, ductus-dependent cardiac anomalies) or give autonomy enhancing valuable information to the women (for example cleft lip). It remains open to studies of other groups whether these results can be confirmed. However the success of FTAS and FTFE should depend strongly upon the qualification of the examiners [9,15]. Further research is needed to get more statistically reliable data. Our study results clearly indicate that FTAS and FTFE are powerful means to reduce the number of late terminations of pregnancies.

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