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Medical Sciences

PRENATAL DIAGNOSIS OF FETAL CARDIAC ABNORMALITIES IN THE FIRST TRIMESTER

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ABSTRACT:
CONGENITAL HEART DEFECTS (CHDs) ARE FOUND IN UP 1% OF ALL PREGNANCIES. THE PRENATAL DIAGNOSIS OF CHDs PLAYS AN IMPORTANT ROLE IN THE MANAGEMENT OF THE DISEASE AFTER BIRTH, THIS VAST PATHOLOGY IS RESPONSIBLE FOR HIGH MORBIDITY AND MORTALITY RATES AMONG NEONATES AND INFANTS. THE FIRST TRIMESTER(FT) ANOMALY SCAN AT 11+0 to 13+6 GESTATIONAL WEEKS OFFERS TIME FOR THE FETAL CARDIOLOGIST AND FOR THE PARENTS TO OPT FOR MORE INVESTIGATIONS, SUCH AS GENETIC TESTING AND EXTENSIVE ULTRASOUND EVALUATION BEFORE THE DECIDE THE COURSE OF THE PREGNANCY. EXTENSIVE FT SCANNING PROTOCOLS ARE IMPLEMENTED IN SOME NATIONAL HEALTH SERVICES TO ALL OR IN HIGH RISK PREGNANCY.

KEY WORDS: FETAL ECHOCARDIOGRAPHY, FIRST TRIMESTER, CONGENITAL HEART DEFECT

INTRODUCTION

During the last 20 years, advancements in the obstetric sonography field had a major impact upon the approach of the first trimester (FT) scan. The emerge of state-of-the-art

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machines that can be used to evaluate and offer detailed information of fetal morphology combined with the extensive training of specialized personnel in the field of maternal fetal medicine, had an important role in transforming the FT scan. From the traditional scan to determine the viability of the pregnancy, the gestational age based on the crown-rump length (CRL), number of fetuses and chorionicity, it had evolved to a complex scan of the fetal structures and early fetal echocardiography (EFEC)[6].

The technical innovations of the past decades transformed the ultrasound in a very powerful tool, that can offer great information in the proper hands. Offering non-invasively structural and functional data about normal and complicated pregnancies, at a relatively cost-effective price, routine FT extensive scans have gained worldwide acceptance.

Congenital heart defects (CHDs) are the most common congenital malformations and are responsible for great neonatal and infancy morbidity and mortality rates[7]. The incidence CHDs is approximately 0,8 – 1% out of live births[10]. There are many forms of heart disease, from mild and asymptomatic such as small secundum atrial septal defects (ASD) or small ventricular septal defects (VSD), to complex malformations that require early postpartum surgical intervention or are incompatible with survival[9].

This pathology brings great socio-economic burden upon families and the medical system. Therefore, the early antepartum diagnosis allows the family to benefit from additional assessment in good time before the limits of legal termination are exceeded. Furthermore, earlier termination offers the mother a safer and more private decision, with less emotional implications[10]. Another important benefit of a normal extended FT scan is the earlier reassurance for the parents[11].

For the reasons mentioned above, early anomaly scan has received great interest and improvement over time[12]. Traditionally, the fetal morphologic assessment is scheduled in most healthcare systems in the second trimester between 18+0 and 23+6 gestational weeks (GW)[13]. However, with the opportunity of the FT nuchal translucency ultrasound, many specialists take their time and conduct an early structural assessment of the fetuses in the first trimester. The aim of this approach is to detect the pregnancies at risk and the cases where sever defects are highly suspected as soon as possible.

BUT IS IT WORTH IT? TO SCAN ROUTINELY EVERY FT PREGNANCY FOR CHDS?

Medical societies, such as the International Society of Ultrasound and Gynecology[1,16] recommend that the evaluation of the fetal heart conducted at 11+0 to 13+6 GW should document just the situs and the blood flow across the tricuspid valve. The documentation of the four-chambers symmetrical view is optional. Additional evaluation of the fetal heart is possible, but not routinely recommended[6]. Another question raised is if the Doppler evaluation is safe at this gestational age[14]. Some studies indicate that Doppler evaluation early in the first trimester may damage fetal structures due to high energy output. However, data is inconclusive but the general acceptance is that Doppler evaluation should be performed in the late FT and not be omitted, because it offers important details, indirectly,

[10] Hoffman JIE, Kaplan S. *The incidence of congenital heart disease*. J Am Coll Cardiol. 2002;39(12):1890-1900.

[9] Nelle M, Raio L, Pavlovic M, Carrel T, Surbek D, Meyer-Wittkopf M. *Prenatal diagnosis and treatment planning of congenital heart defects-possibilities and limits*. World J Pediatr WJP. 2009;5(1):18-22.

[10] Johnson B, Simpson LL. *Screening for congenital heart disease: a move toward earlier echocardiography.* Am J Perinatol. 2007;24(8):449-456.

about the heart structure and function which cannot be identified using only the gray-scale function[11].

Another important aspect in the FT is the introduction of the non-invasive prenatal testing (NIPT) for aneuploidy screening as routine in some medical systems[15]. Therefore, the scan for nuchal translucency (NT) might not be appropriate in all cases. Hence the question if the FT anatomy scan and the EFEC are justified and cost-effective[16]. It is generally accepted that an increased nuchal translucency is a modest predictor for CHDs. However, when combining nuchal translucency measurement with doppler interrogation of the tricuspid and ductus venosus flow, we can identify pregnancies which present a higher risk of CHDs[17]. When a high nuchal translucency alone was determined, just one third of the cases confirmed with Trisomy 21 were associated with a CHD (usually an ASD)[18]. On the other hand, just 15% of the fetuses that suffer from CHDs have an increased NT in the absence of Down Syndrome[19]. Some authors consider that Trisomy 21 represents sufficient grounds to choose to terminate the pregnancy in the first trimester and that the rate of false positive results at the EFEC scan are too high to justify the need of this early anatomy scan[16]. However, our protocols do not support this approach and an early scan, translated in an earlier diagnosis is preferred.

THE EARLY FETAL ANOMALY SCAN AND EARLY FETAL ECHOCARDIOGRAPHY

Gradually, the first scan, which is typically scheduled from 11+0 to 13+6 GW, was transformed into a morphology scan. More and more parameters are checked with the occasion of this medical visit for an earlier diagnosis of CHDs (and other extracardiac abnormalities) and for parents' earlier reassurance. This scan can and should include a basic checklist that covers the most important major fetal abnormalities (MA) that can be debilitating or even lethal[12]. However, the success of the FT anomaly scan depends on the level of detail of the morphological protocol. If in a certain center the CHDs diagnosed in the FT are just coincidental or the detailed protocols are applied just to high risk pregnancies (such in the case of an increased NT), the FT scan will have low and unrealistic detection rates of the major CHDs.

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[11] Khalil A, Nicolaides KH. *Fetal heart defects: Potential and pitfalls of first-trimester detection*. Semin Fetal Neonatal Med. 2013;18(5):251-260.

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[14] Fernández SG, Ramirez JA, Chouza MTO, Alonso BR-V, Coto ÁPL. *Early fetal ultrasound screening for major congenital heart defects without Doppler*. Eur J Obstet Gynecol Reprod Biol. 2019; 233:93-97.

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[16] Gardiner HM. *First-trimester fetal echocardiography: routine practice or research tool?* Ultrasound Obstet Gynecol. 2013;42(6):611-612.

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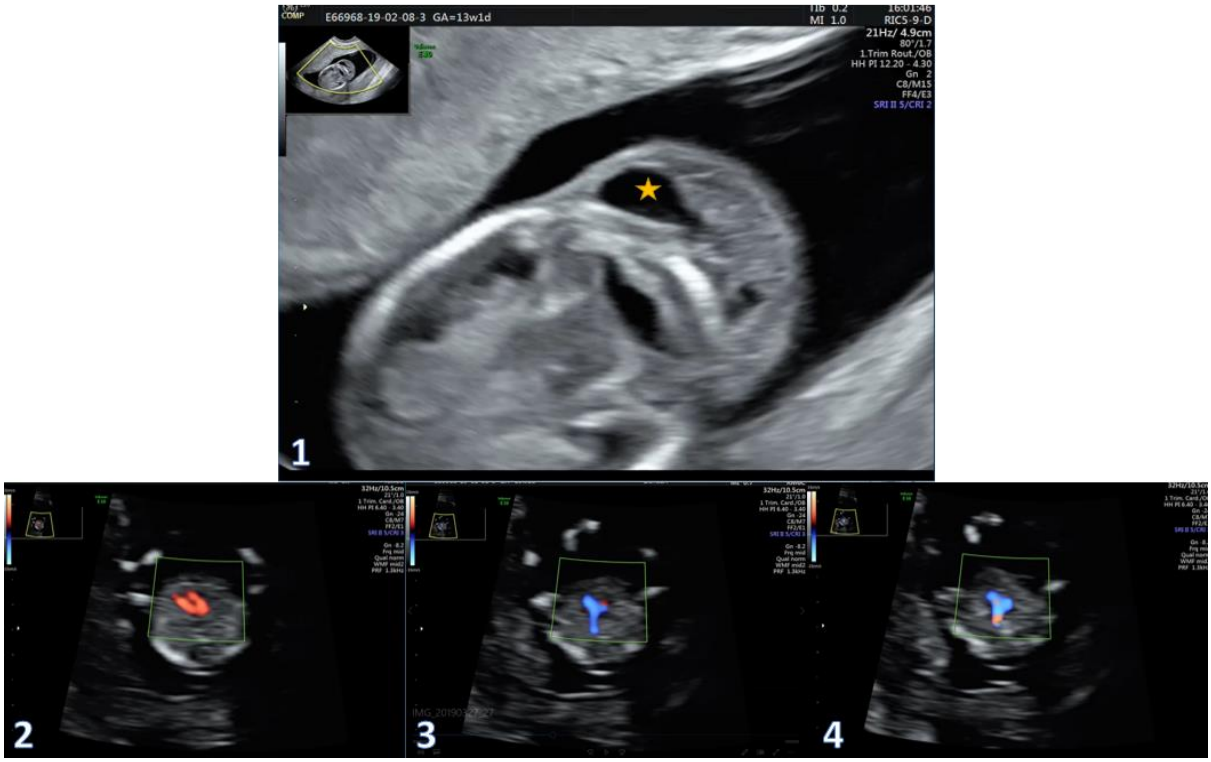

*Figure 1: First trimester (13weeks+1 day) anomaly scan in a primi gesta, primi para pregnant, 30 years old woman reveals: cystic hygroma with apparent normal Doppler cardiac sweep in first trimester. (1) Cervical cyst marked with *; (2) Four-chamber view; (3) Three vessel view; (4) Ductus arteriosus.*

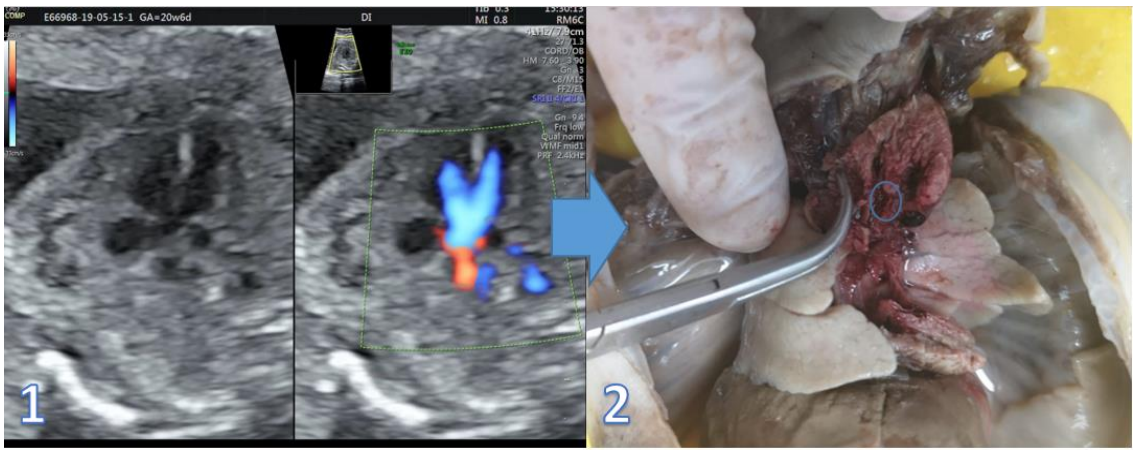

Figure 2: Ultrasound examination in the second trimester of the same fetus. Membranous VSD was suspicioned (1) and confirmed at the autopsy (2) (the family choose medical termination of the pregnancy as the fetus associated ventriculomegaly, unilateral renal agenesis, persistent cystic hygroma, single umbilical artery, small stomach and suspicion of anal atresia).

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However, when an extensive protocol is applied systematic with the occasion of every FT scan by an expert sonographer, it can detect 80% of the MA; a performance similar with the second trimester anomaly scan[20]. One systematic review[21] reported a sensitivity of 85% (95% CI, 78-90%) and a specificity of 99% (95% CI, 98-100%) in the detection of CHDs at the FT scan. The sensitivity when scanning normal hearts was 85% in the same study.

The ISUOG guidelines recommend for the FT scan just the documentation of the heart activity and the normal four-chamber view of the heart. However, in expert hands and with a modern machine, heart situs, aortic outflow tract, pulmonary outflow tract, crossing of the pulmonary, confluence of the arterial arches, the three vessel and trachea view and the ductus venosus flow can be assessed in the late FT. At 13 GW the rate of cardiac evaluation reaches 92%[22]. Therefore, at the end of the FT the visualization of the fetal heart anatomy is more than possible and brings important advantages concerning the management of the pregnancy and family counseling[12].

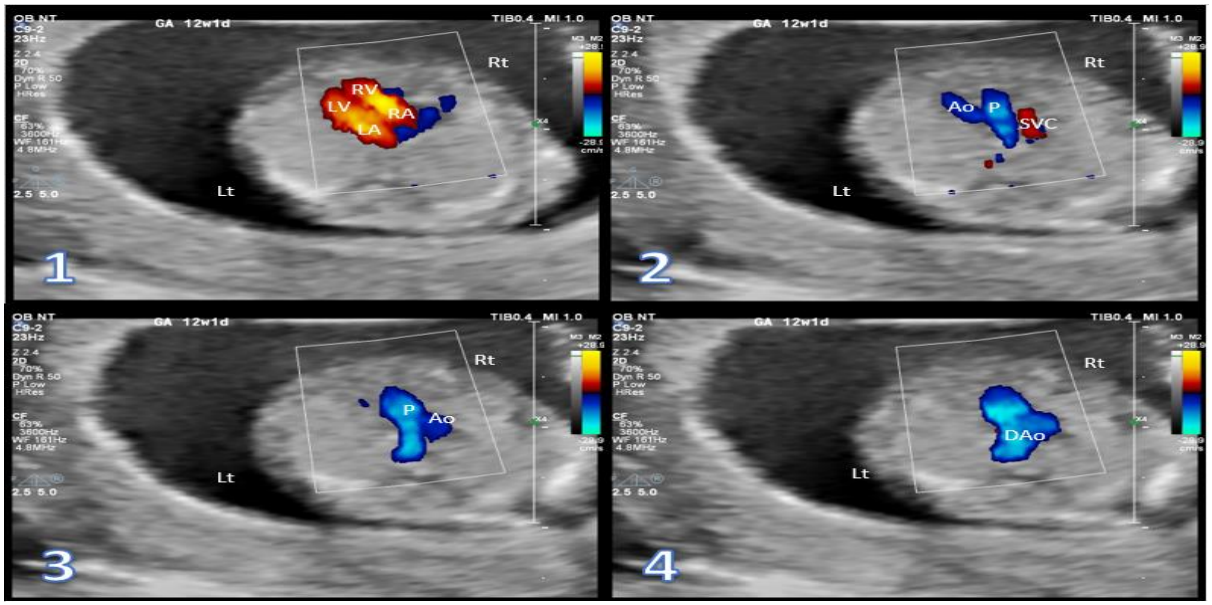

Figure 3: First trimester (12 weeks + 1day gestational age) cardiac sweep in a normal fetus. (1) Four-chamber view Doppler mode: parallel flows from both atria to both ventricles; (2) 3-vessel view; (3) Crossing of the great vessels. (4) Ductus arteriosus. Lt, left; Rt, right; LV, left ventricle; RV, right ventricle; LA, left atrium; RA, right atrium; Ao, aorta; P, Pulmonary trunk; DAo, ductus arteriosus; SVC, superior vena cava.

Unfortunately, not all CHDs can be diagnosed in the first or second trimester. A normal cardiac anatomy scan in the FT does not exclude a CHD. Some pathologies develop later in the pregnancy (e.g. tetralogy of Fallot, pulmonary stenosis, coarctation of the aorta[12]), or even after birth (e.g. cardiomyopathies[23]). Also, some smaller defects can be missed (e.g. small VSD) or mislead the sonographer with an apparent normal aspect (e.g. the right aortic arch with right ductus arteriosus)[24]. Other defects can evolve from the FT scan and be visible in the mid-pregnancy screening (e.g. pulmonary and aortic stenosis)[11]. Thus, the parents must be counselled correctly to understand that an apparently normal FT scan does not translate in a perfectly healthy fetus.

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CONCLUSIONS

The FT screening for CHDs is recommended when an extensive scanning protocol is implemented. The main advantage of this approach is the time the practitioner is saving for further investigations and additional examinations, rather than waiting for the first scan after 18 GW. These 4 – 10 weeks saved offer the families greater reassurance and also the option of a safer and more private termination of the pregnancy in case of severe CHDs or termination at request before 14 GW. An earlier medical termination of the pregnancy has less emotional and phycological impact upon the parents.

This early scan is an operator dependent tool. Even if it can be extremely valuable in expert hands, novice scanners can bring many false positive or false negative results that may inadvertently affect the course of the pregnancy. Taking into consideration that the expertise of the sonographer is the most important factor involved in the first trimester morphology scan, the scarcity of properly trained specialist becomes a problem, especially in developing countries. Another disadvantage is the high price of the machines that can offer these early images.

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All authors had the same contribution.

All authors report no potential of interest.

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