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THE INCIDENCE OF FETAL CARDIAC ANOMALY IN ASSOCIATION WITH PRESENCE AND ABSENCE OF ANEUPLOIDY MARKERS IN FIRST TRIMESTER – TIME TO EDGE FORWARD

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Abstract

Background: Among the three trimesters of pregnancy, first trimester ultrasound plays an important role to accurately estimate gestational age, to know the viability of the fetus and so on. More recently, we have enclosed the term aneuploidy markers in first trimester. These are soft markers which are not abnormal, but indirectly gives a clue to few non-chromosomal anomalies. **Aim:** the Main aim of this research is to check the significance of association between aneuploidy markers and fetal cardiac anomaly in first trimester**. Methodology:** The sample size of the study is 700 patients.Antenatal patients with gestational age from 11th week of gestation onwards till the 14th week of gestation presented to VMKVMCH, Salem had been analysed in this study for detection of fetal cardiac anomaly with presence and absence of first trimester aneuploidy markers. **Results:** This study examines the link between fetal heart defects and presence or absence of aneuploidy markers in the first trimester. Among 700 patients, A female 24-year-old showed naturally occurring corrected the major arteries' transposition, with notable anomalies including modified right ventricle, disproportionate four chamber view, two vessel tracheal view and aberrant right subclavian artery were also observed. **Conclusion:** These findings emphasize the importance of early detection and management of fetal cardiac anomalies, especially when associated with aneuploidy markers.

Keywords: Fetal Cardiac Anomaly, First Trimester, Aneuploidy Markers, Fetal Cardiac Circulation.

INTRODUCTION

Our mother earth is the only planet which embrace varying organisms from low level prokaryotes to eukaryotes. The theory of Evolution has contributed significantly to the evolution of human beings. It took millions of years for this evolution to happen. Now, we humans use sonological machines to visualise body structures. This invention has brought mother and fetus in utero closer, before the baby is brought to the world.

The circulation system of heart orchestrates the symphony of life. Very rarely, this balance gets disrupted during fetal development1. Since the heart is a critical organ, for a human life, it is imperative to include fetal cardiac examination as part of a comprehensive fetal evaluation in first trimester2.

In the age of non-invasive prenatal diagnostics, the value of sonographic evaluation between 11 and 14 weeks of gestational age is becoming increasingly clear, going beyond screening for chromosomal abnormalities. In fact, one could argue that

prenatal ultrasound has returned to its former glory with the assessment about Nuchal translucency and its global significance in the practice of obstetrics. Due to its ability to detect severe anomalies, such as heart abnormalities, earlier in the gestational period, this test has radically transformed prenatal treatment. This facilitates the provision of additional reproductive options and earlier and better planning for pregnancy and delivery.

The viability of cardiac first trimester examination in a risky situation groups, such as those who have a mother's diabetes, a family history of cardiac problems, or exposure to teratogens, has been shown in several studies conducted over the last 20 years. However, only a tiny percentage of heart abnormalities will be detected if the criteria for cardiac first trimester examination are limited due to these elevated risks groups. About 50% of significant cardiac problems may be indirectly identified by extending the scope of the first trimester cardiac assessment to include women who have aberrant soft indicators like increased translucency of nuchal matter, an inversion of the wave in the tricuspid regurgitation and the venosus ductus 7–10. As of right now, only the four chamber perspective, location, and heart rate are recommended for fetal heart examination in first trimester ultrasonography recommendations.

The time has come to reconsider first trimester cardiac evaluation of emerging technologies and the capacity to identify significant structural and chromosomal problems at a younger gestational age. We should also think about incorporating standard images of the second trimester heart, encompassing the three vessel view, the chamber 4 view and the views outflow tracts of the left & right into our first trimester assessment.

The most prevalent congenital abnormality, birth defect in the heart abnormalities (CHDs), affect less than 1% of live births. Given the severity and mortality of many of these complicated disorders, the real incidence during the prenatal period is up to five times greater. Most congenital cardiac abnormalities happen in a group at low risk when there are no known danger indicators.

Utilizing sonographic aneuploidy markers to their fullest potential is a new trend, particularly during the first trimester of pregnancy. The ability to identify chromosomal abnormalities has increased over the last several decades due to technological and medical developments. Since many fetal anatomical malformations manifest before the end of the first trimester, there are many possibilities to identify chromosomal abnormalities early in pregnancy. The transvaginal ultrasonography is the most preferred and pleasant treatment; however, a transabdominal probe may also be used for an Ultrasound analysis during the first three months. The introduction of early screening procedures might provide a genuine potential to reduce the proportion of neonates born with chromosomal aberrations as soon as feasible. The most prevalent chromosomal aneuploidy, such as trisomy 21, may have a recognizable look during the first trimester. As opposed to this, an early diagnosis allows for a pregnancy to end with fewer difficulties and more time for arranging future follow-up and interventions11–15. Women who are identified with fetal ultrasound signs of aneuploidy during the first trimester should be provided the option of undergoing noninvasive testing or prenatal cytogenetic analysis.

Thus incidence of fetal cardiac anomalies in association with presence and absence of first trimester aneuploidy markers are crucial in understanding the relationship between cardiac anomaly and aneuploidy markers.

AIM & OBJECTIVE

To Study The Incidence Of Fetal Cardiac Anomaly In Association With Presence And Absence OF First Trimester Aneuploidy Markers Presented To The Radiology Department For First Trimester Obstetric Ultrasound.

MATERIALS AND METHODS

The VMKV Medical College & Hospitals, Salem served as the study's location, involving all pregnant women who presented to the Radiology Department for first-trimester obstetric ultrasound over a three-year period. It was a prospective study with a sample size of 700 women who were 11 to 14 weeks along during their pregnancy and who underwent obstetric ultrasound examination at the Radio-Diagnosis Department.

Inclusion criteria for the study comprised pregnant patients with single pregnancies who knew the correct dates of their last menstrual period (LMP) and whose gestational age was confirmed by dating scan (<12 weeks). Exclusion criteria included twin gestations, fetuses with poor visualization of anatomical landmarks necessary for assessment, gestational age above 14 weeks, and patients unwilling to provide consent or undergo follow-up.

The investigation was carried out at Vinayaka Missions Kirupananda Variyar Hospital and Medical College, Salem, in the Department of Radiodiagnosis. This study will be done in 700 pregnant women between 11^{th} week of gestation and 14^{th} week of gestation referred from the Obstetrics and Gynaecology OPD. Ultrasound examination was performed after the acquisition of signed F form and written informed permission using 2D ultrasound to image soft aneuploidy markers like Tricuspid regurgitation, Absence of ductus venosus or reversal of A wave, Absent nasal bone, Echogenic intracardiac focus, enlarged Nuchal translucency, and aberrant right subclavian artery. Suprapubic manipulative jerks gives better position. In situs solitus, axial view of upper abdomen shows stomach with cardiac axis towards left. In situs inversus totalis, stomach and apex lies on right side and it may be associated with other anomalies. Viewed in four chambers, situs, size, axis and the location of the heart is evaluated. Left axis deviation strongly correlates with cono-truncal anomalies. Cephalad to the chamber of four view is the vessel tracheal three view from the vessel ductal and aortic arch join to form V or tick sign which should point to the left of the spine. The direction of flow should be same in both the vessels.

Anomalies based on chamber disproportion and abnormal three vessel view is being diagnosed in first trimester.Antenatal patients with gestational age from 11^{th} week of gestation onwards till the 14^{th} week of gestation for detection of fetal cardiac anomaly with presence and absence of first trimester aneuploidy markers. Detailed clinical history and examination of the mother, along with comprehensive sonography of the fetus, are essential for thorough data collection in prenatal care. For the statistical analysis, SPSS software version 19 was used.

RESULTS

The results obtained after the scrutiny of the patients with Fetal Cardiac Anomaly in Association with Presence and Absence OF Aneuploidy Markers in First Trimester.

During the first trimester scan of a 24-year-old female, several significant abnormalities were identified in the fetus:

1. Great artery transposition that was naturally corrected: This condition involves the inversion of the ventricles, with the Rt(right) ventricle positioned posteriorly on left and the Lt(left)ventricle situated anteriorly on the right side. This abnormal positioning alters the normal the heart's blood flow.
2. Anomalous arterial in the lungs and aorta positioning: The pulmonary circulation, which typically originates in the right ventricle, instead start from the left-side and aorta originate from the left ventricle which is situated on the right side.
3. Abnormal three-vessel tracheal view: The tracheal view, typically characterized by the presence of three vessels (Pulmonary artery, Aorta and Superior vena cava) in the fetal thorax, appears abnormal, indicating potential structural anomalies or deviations from normal anatomy.
4. Ductus venosus connects the umbilical vein to the inferior vena cava shows reversal in Doppler waveform.
5. The right subclavian artery is abnormally present: An additional finding is Anomalous right subclavian artery, which represents an anomalous origin or course of this artery arising from the aortic arch.

These findings collectively indicate complex congenital cardiac and vascular abnormalities in the fetus, necessitating further evaluation and management to assess the potential impact on fetal development and postnatal health.

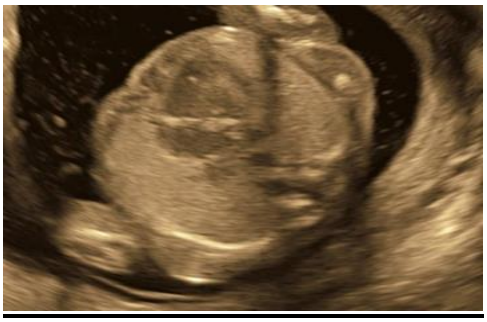

Figure 1: Disproportionate 4 chamber view.

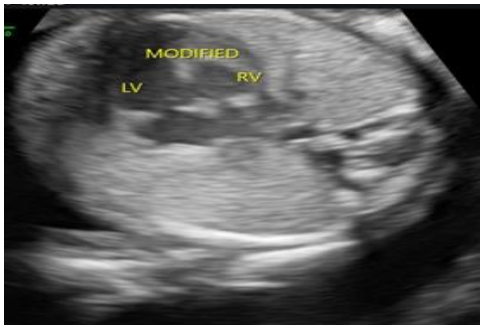

Figure 2: Modified right ventricle.

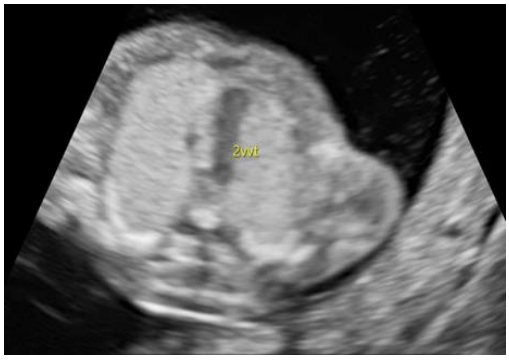

Figure 3: Two vessel tracheal view is seen instead of three vessel tracheal view.

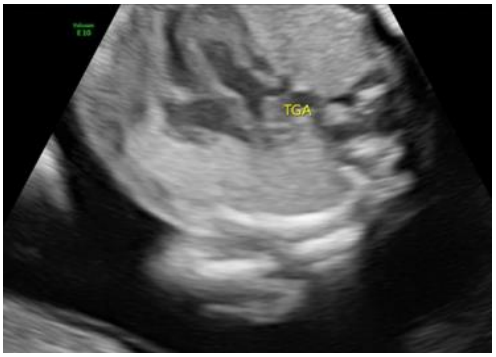

Figure 4: Transposition of great arteries.

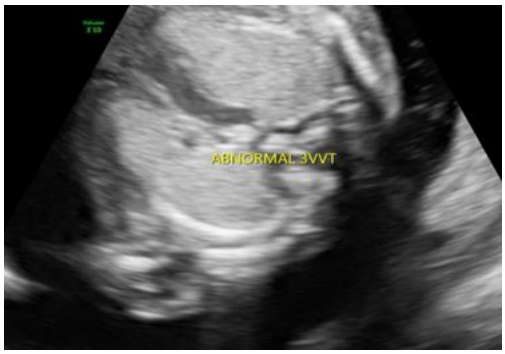

Figure 5: Abnormal three vessel tracheal view.

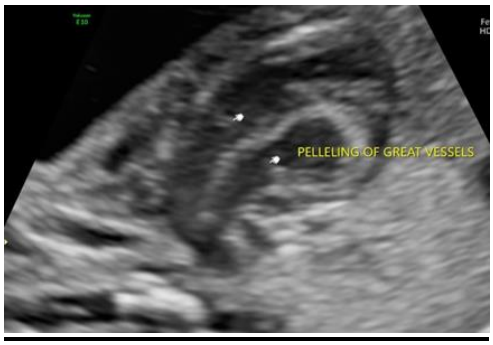

Figure 6: Paralleling of great vessels.

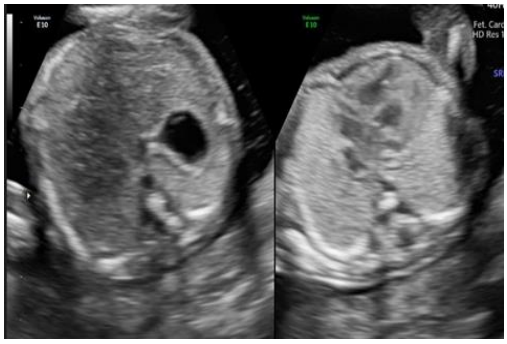

Figure 7: Situs solitus.

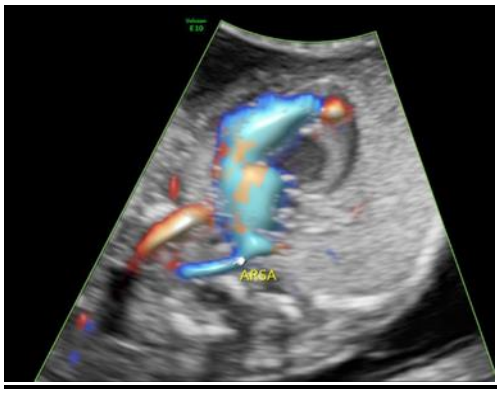

Figure 8: Aberrant right subclavian artery

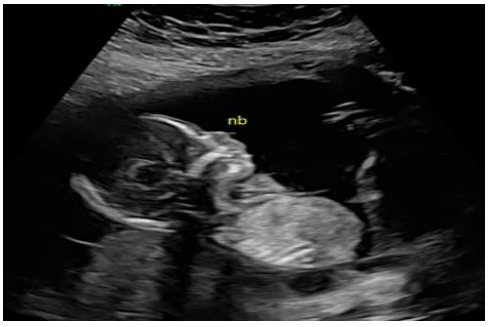

Figure 9: Nasal bone present

The presence of fetal cardiac anomaly, specifically the major arteries' transposition, suggests potential aneuploidy markers, while no other abnormalities were detected.

DISCUSSION

This study, encompassing 700 patients, identified a single congenital cardiac anomaly. An uncommon congenital heart defect known as complete by forcing the aorta to emerge from the morphological Rt ventricle and Lt ventricle gives the pulmonary artery, transposition of the great arteries (cTGA) results in ventriculoarterial discordance with atrioventricular concordance. This condition leads to parallel blood flow and a cyanotic congenital heart defect. Congenital heart defects (cTGA) represent 5-7% of all birth defects, with a frequency of 0.3 occurrences per 1000 live births. Depending on the partnership between the pulmonary and aortic valve, cTGA can present as dextro-loop TGA or levo-loop TGA, the latter also known as congenitally corrected TGA, where the aorta is positioned to the left and in front of the pulmonary artery, a rare abnormality with atrioventricular and ventriculoarterial discordance

(Warnes, 2006)[16]. The etiopathogenesis of cTGA involves the failure of normal regression of the subaortic conus and growth of the subpulmonic conus, coupled with the failure of the aortopulmonary septum to assume a spiral form (Kutty et al., 2018)[17].

Ultrasound features of cTGA include the absence of normal "crossing over" inside the three-vessel perspective of the major arteries, as they arise in parallel. In d-TGA, the three-vessel tracheal view displays a single large vessel (the transverse aortic arch) with a right-sided SVC (superior vena cava), accompanied by a right convex shape (reverse boomerang sign) or a straight orientation (I sign) of the aorta (Hornung & Calder, 2010)[18]. The differential diagnosis for cTGA includes both major arteries mostly emerge from the Rt ventricle with a double outlet (DORV), often with a ventricular septal defect (VSD) (Lapierre et al., 2010)[19]. In conclusion, cTGA is a complex congenital heart anomaly that requires careful diagnosis and management due to its significant impact on cardiac function and patient outcomes. Early detection and appropriate interventions are crucial in improving the prognosis for individuals with cTGA.

CONCLUSION

Transvaginal ultrasound aids in detecting cardiac anomaly in first trimester better than transabdominal ultrasound. The incidence of congenital anomaly in first trimester is 0.14 percent and is more common with presence of soft aneuploidy markers. Thus, in the presence of aneuploidy markers congenital heart anomaly should be very keenly searched for.

Conflict of Interests

Authors declare that they have no conflict of interests.

Ethical Issues

This research study received approval from the Institute Ethics Committee of Vinayaka Mission's Kirupananda Variyar Medical College & Hospital, Salem.

Availability of Data

The dataset of this study is available upon request from the corresponding author.

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