Original Paper

The Contribution and the Importance of Modern Ultrasound Techniques in the Diagnosis of Major Structural Abnormalities in the First Trimester – Case Reports

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ABSTRACT We describe a series of cases where modern ultrasound (US) techniques diagnosed major structural abnormalities of the fetus in the first trimester (FT), unapparent when using the basic protocol of US investigation. In some cases, major structural abnormalities can be revealed in the FT scan solely to specialized personnel. Perhaps early screening should be confined in specialized centers, because congenital abnormalities detailed diagnostic has a huge impact in counseling the couple and also in prenatal advice of future pregnancies.

KEY WORDS *Ultrasound, Structural Abnormalities, First Trimester*

Introduction

US diagnosis represents a huge impact on obstetrical practice and perinatal medicine because it offers important information regarding the morphological and physiological development of the fetus (1). Many studies have shown that major structural abnormalities can be diagnosed very early in pregnancy, during the FT genetic ultrasound (2-19). Similar to the second trimester evaluation, the detection rate is reported with high variations, between 12% (4) to 84% (13), in our opinion depending on the extension of the ultrasound examination protocol.

We report cases of first trimester pregnancies that were accurately studied following an extended US study protocol of fetal anatomy. The cases presented severe abnormalities that were detected and confirmed because of the use of detailed US and pathologic evaluations.

Method

The early fetal evaluation aimed to assess the genetic and morphologic parameters suitable for evaluation at the respective gestational age using a detailed sonographic protocol.

The examinations were performed by obstetricians with special interest in fetal medicine, and accredited for assessment at 11- to 14-week scan according to Fetal Medicine Foundation (FMF) criteria.

Transabdominal and transvaginal examinations were performed using Voluson 730 Pro and Expert, GE Medical Systems, Kretztechnik, ZIPF, Austria US machine, equipped with 5–7 MHz and 5–10 MHz curvilinear transducers. Transvaginal route was added only when necessary, in case of unfavorable fetal position, unfriendly maternal conditions or for better visualization of fetal abnormalities.

Results – Case Report Series

We report and present a series of cases detected during 2011 at the prenatal diagnostic unit of University of Medicine and Pharmacy Craiova, showing severe structural abnormalities diagnosed due to extended morphological investigation.

Atrio-ventricular septal defect cases

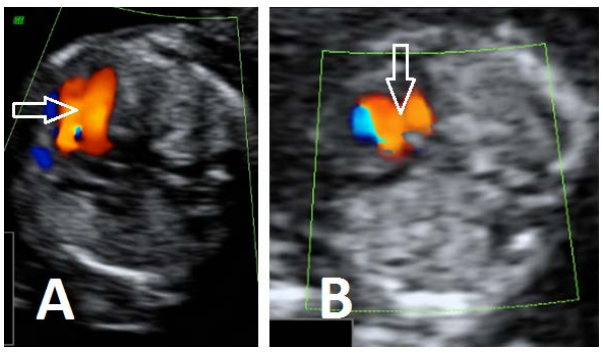
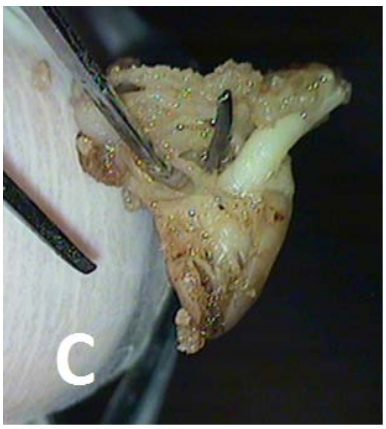


Figure 1. Atrio-ventricular septal defects (A, B). Color Doppler mode shows communication bethween the right and left side of the fetal hearth (open arrows).



C: confirmation of the anomaly at the pathologic evaluationMitral atresia with VSD (Hypoplastic right hearth syndrome)

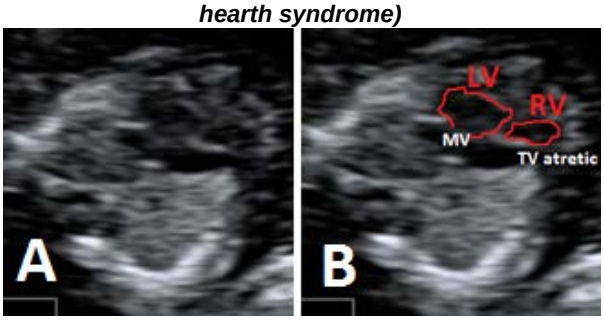
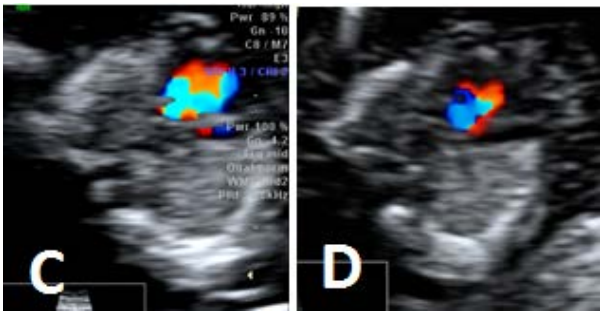
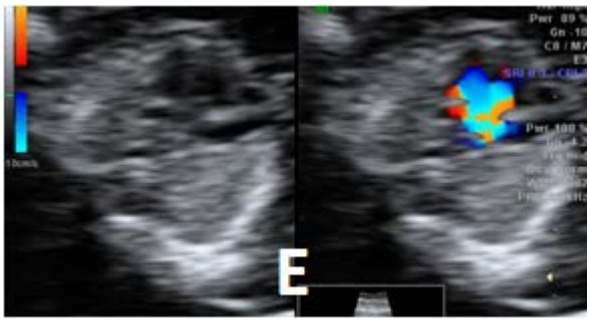


Figure 2. A, B: Abnormal four-chamber view with a diminutive right ventricle, septal defect and atretic, thikened tricuspid valve. LV, left ventricle; RV, right ventricle; MV, mitral valve; TV, tricuspid valve.



C, D: Color Doppler at the four-chamber view during diastole. C: Absence of flow across the atretic, and thikened tricuspid valve. D: The right ventricle receives blood from the left ventricle across the ventricula septal defect in late diastole and systole.



E: Dual mode (B mode and color Doppler) at the four-chamber view during systole. Presence of mitral valve rgurgitation.Hypoplastic left heart syndrome

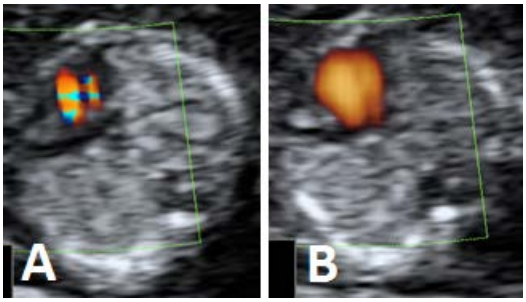
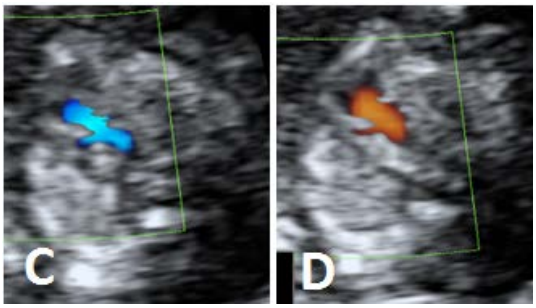


Figure 3. A, B: A narrow width of the left ventricle in comparison to that of the right ventricle in color (A) and power Doppler (B) at the level of the four-chamber view of the heart.



C, D: Narrowed left outflow tract with forward flow is noted in the color (C) and power Doppler (D) at the level of the three vessel-trachea view.

Transposition of the great arteries

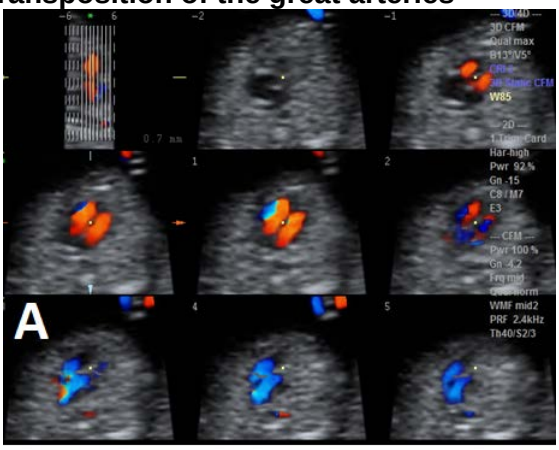
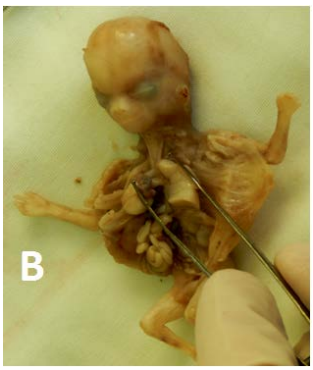


Figure 4. A: 4D (STIC) evaluation with color Doppler, showing the parallel emergence of the great vessels at the base of the heart.



B: Pathologic specimen showing the left and right outflows emergence

Hyoplastic right heart syndrome – mitral atresia with intact ventricular septum

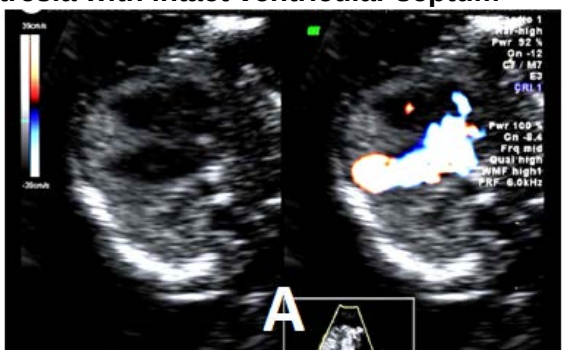
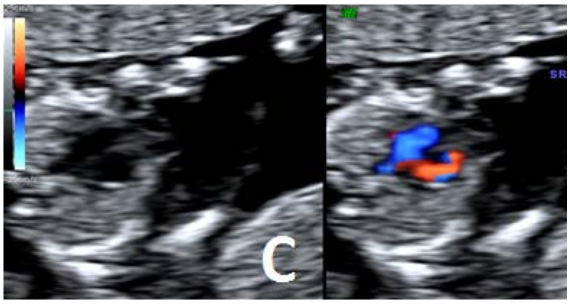
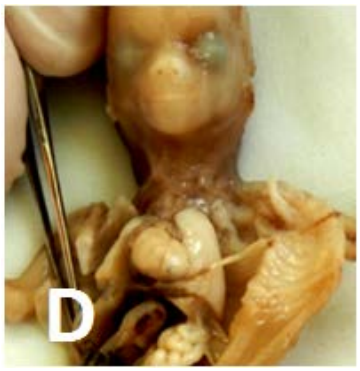


Figure 5. A: Apparent normal four chamber view of the fetal heart and absent filling of the right ventricle during diastole through the tricuspid valve.

B: discretely dilated right atrium.



C: Transversal three vessel and trachea view with color Doppler applied. Reversed flow in the right outflow tract.



D. Enlarged right atrium at the authopsy exam, confirming the imagistic suspicion formulated in B.

Agenesis of ductus venosus

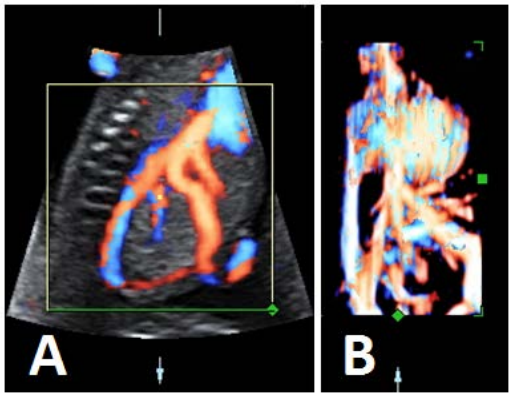
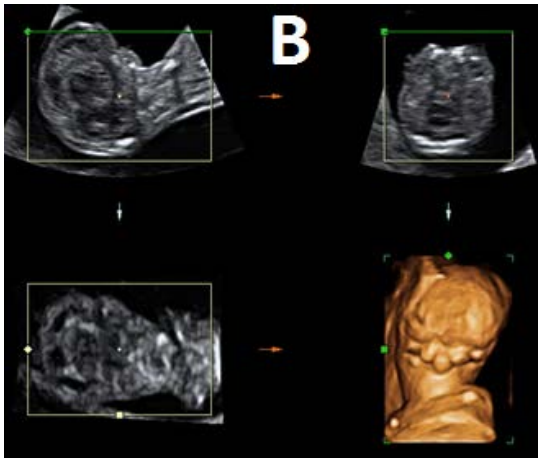


Figure 6. A: 4D STIC reconstruction of fetal circulation (thorax and abdomen). Absence of a normal conformation of ductus venosus with caval drainage of the umbilical flow. B: Normal development of hepato-portal circulation.

Tetramicromelia and severe facial defect

Figure 7. Tetramicromelia and facial defect in 3D reconstruction and pathologic specimen. A: longitudinal view.



B: coronal view.

Polydactily

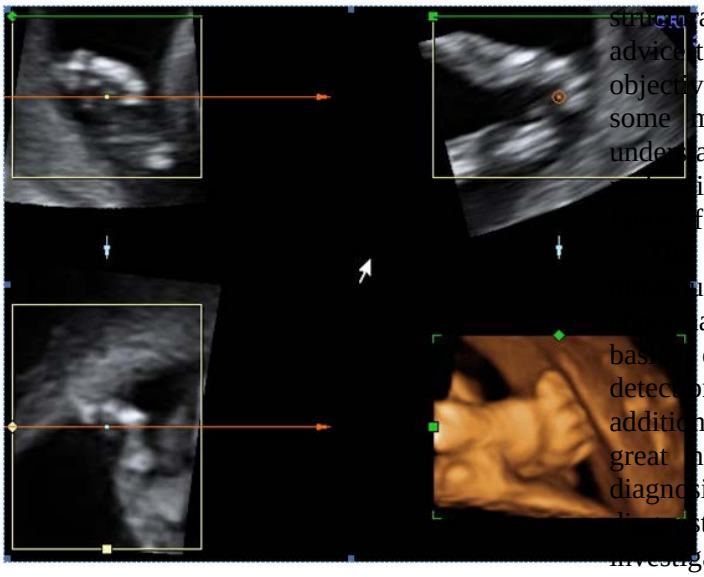


Figure 8. Polydactily in 3D evaluation

Disscusion

The respect for freedom of choice is the main principle of medical ethics. Providing a high quality screening service increases the couple autonomy regarding the prenatal decision.

A large group of major congenital malformations are detectable using ultrasonography (US) in the first trimester (FT). It would appear that some severe structural anomalies are not as hard to detect as formerly believed, and can be diagnosed at 12 to14 gestational weeks (GW) if practitioners are cognizant of their early sonographic appearance and structural or functional associated abnormalities.

The detailed study of early pregnancies with severe or complex structural abnormalities is important for early counseling and long term counseling for two main reasons. An efficient morphological ultrasound can detect fetal abnormalities early in the pregnancy so that the termination of pregnancy is safer with diminished economical and emotional costs both for the medical system and also for the couple. Second, the detailed exploration of the affected fetuses and the diagnostic of all malformations can have an important role in the prenatal advice of future pregnancies.

Two-dimensional gray-scale US has been shown to be an effective tool for imaging the normally developing FT fetus, and may have more to offer than does within the traditional checklist. Color Doppler is a valuable tool for early heart investigation of anatomy and functional parameters. 3D/4DUS, power Doppler and high definition flow provides confidence in the suspected diagnostic especially in complex structural defects and assists in offering cogent advice to patients. Using these techniques we can objectively present to the parents the features of some malformations which allow for a better understanding of the pathologic condition and make it easy for the couple to decide or not in favor of termination.

first trimester detailed morphological ultrasound can detect genetic and structural abnormalities undetectable when using the usual basic examination protocol. The increased detection of different abnormalities implies additional time to the standard examination, a great number of specialists in early prenatal diagnosis and a high technology equipment of the diagnostic centers necessary for a detailed investigation protocol of scanning in the first trimester.

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