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Case Report

Complete Atrioventricular Canal Defect Diagnosed at 12 Weeks Associated with Trisomy 21: A Case Presentation

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Abstract

Among the congenital malformations the most common are the congenital heart defects (CHD) that occur in about 10% of the newborns.

Congenital heart malformations diagnosis is most often made in the second trimester of pregnancy at the second trimester anomaly scan.

Early diagnosis, however, offers significant advantages regarding the opportunities of further investigations, counselling of future parents and access to management options.

Among other investigations in case of CHD diagnosis are the tests for detecting chromosomal abnormalities (CVS or cff DNA), knowing that the association of some CHD with aneuploidies that might change the management options and the counselling.

We present the case of a patient, a primigravida that came into our clinic at 11+3 days weeks of gestation for her first trimester screening and the fetus was diagnosed with a CHD-atrioventricular septal defect (AVC).

Keywords: *CHD (Congenital Heart Disease); AVC (Atrioventricular Canal); Aneuploidies; CVS (Chorionic Villous Sample)*

Introduction

CHD occur in approximately 10% of congenital malformations.

Most often the diagnosis of CHD is made in the second trimester of pregnancy at the fetal anatomy scan.

In the last years, the advances of technology in antenatal imaging have improved the detection of fetal heart anomalies and also moved the diagnosis from the 20th week of gestation, much earlier at 11 - 14 weeks, allowing the healthcare provider and future parents time for further investigations, early management and decision options and counseling, knowing the fact that depending of the severity of anomaly, sometimes, the parents decide for termination of pregnancy (TOP).

An early TOP is accompanied with less risks compared with a TOP at mid pregnancy.

The first trimester screening in Romania, like in other European countries follows the protocols from the FMF, London and ISUOG.

At 11-13+6 weeks of gestation we assess the CRL for a correct dating, the NT (nuchal translucency), NB (nasal bone), DV (ductus venosus) and a routine morphological assessment.

Due to the fact that only certified physicians are allowed to perform this screening and the availability of modern ultrasound machines it is common practice to assess at this time of gestation the fetal heart, according to the protocol of ISUOG.

The scan of the fetal heart in the first trimester includes a systematic approach and is done according the guidelines of ISUOG (International society of ultrasound in obstetrics and gynecology) [1].

These guidelines include:

4CH view:

- FHR, regular heart rhythm.
- The situs-intrathoracic.
- Cardiac axis-30-60°.
- Size = 1/3 of thoracic space.
- Presence of two distinct ventricles.
- The symmetry of right and left atria and ventricles.
- PW and CD (Axial).
- Diastolic flow from right to left.
- Absence of tricuspid regurgitation.

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- CD (color doppler).
- Direction of blood flow in aorta and pulmonary artery.
- Great arteries pointing to the left side.
- Ductus venosus, PW, CD, antegrade DV, a-wave.

Why it is so important to scan all fetal hearts in the first trimester?

From the anatomic point of view the fetal heart is completed at 8 - 10 weeks of gestation and as early as 11 to 13 - 14 weeks the fetal cardiac anomalies can be present.

The accuracy of an early heart examination is dependent of several factors:

- Gestational age (better at 13 than at 11).
- Center expertise.
- Local protocols.
- Operator experience.

- Associated factors that trigger the attention for further investigation such as NT, NB.DV.
- Mother BMI.
- Placental localization.
- Previous scars from previous surgery (e.g. C-section).
- Fetal position and mobility.

The scan may be done transabdominal and for a better evaluation may be associated a transvaginal examination.

It is important to note that there are fetal heart anomalies that can be detected at 13 - 14 weeks of GA with a similar accuracy with that at the mid trimester, with the lowest false positive and false negative as much as it can be.

There are also anomalies as the conotruncal anomalies that are challenging at the first trimester.

Among these: TOF (tetralogy of Fallot), pulmonary artery stenosis, aortic stenosis, small VSD, common arterial trunk.

In experienced hands during the first trimester scan the detection of:

- Tricuspid atresia.
- Pulmonary atresia.
- Hypoplastic left heart.
- Atrioventricular septal defect.
- Right aortic arch.
- Vascular rings.

Have been reported over 90%.

Some of congenital heart diseases are reported to be more frequent associated with chromosomal abnormalities.

The next step in the management of a patient with a newly diagnosed CHD is the search for aneuploidies.

It is well known that five types of defects, with some overlap are associated with trisomy 21 (Down syndrome):

- Ventricular septal defect-VSD.
- Atrioventricular septal defect.
- Patent ductus arteriosus.
- Tetralogy of fallot.
- Endocardial cushion defect is the predominant cardiac abnormality in T21.

The absence of transpositions and the rarity of right-and left sided obstructive lesions in trisomies indicate that there may be a genetic influence on specific embryologic mechanism.

Case Presentation and Discussion

We present the case of a primigravida, 26 years with no associated pathology, non smoker, no allergies, no exposure to environmental factors, with a BMI of 22.

No family history, no personal history of any disease.

Doesn't use drugs, tobacco, alcohol or any other medication.

Uneventful course of pregnancy until she came in our clinic for first trimester scan.

CRL was correspondent with the menstrual age, NB present, NT normal, no reverse flow in the a-wave at DV, PI of DV, normal.

The biochemical screening done in our laboratory according the protocols of FMF (fetal medicine foundation, London, UK), beta HCG and PAPP-A using the Brahms Kryptor, return normal.

At the anatomy of the fetus we detected a fetal heart anomaly with highly suspicious of atrio ventricular septal defect.

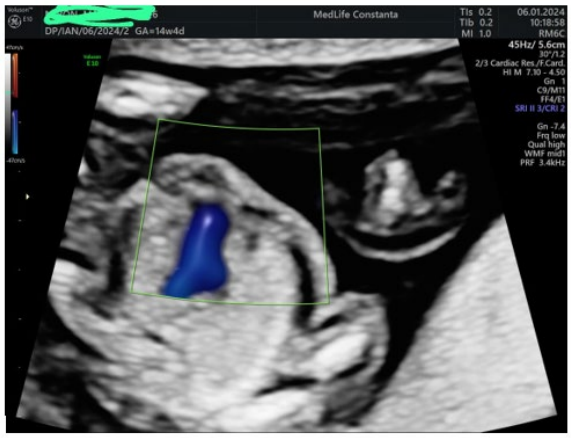
We did the ultrasound using the transabdominal approach and the transvaginal one with GE Voluson E10, and the fetal cardiac scan soft from GE electric.

Not intending to create a psychological stress for the patient we told her that we need to make another scan in almost two weeks and ask her to perform a screening test to search for cell free fetal DNA that has a greater accuracy for detecting aneuploidies associated with cardiac anomalies.

She came back in two weeks and we confirmed the cardiac anomaly atrio ventricular canal defect.

We informed the patient that we need the result of genetic testing in order for a correct counselling and decide the management.

The next week we had also the results of cell free fetal DNA that turned out positive for trisomy 21.



Figure

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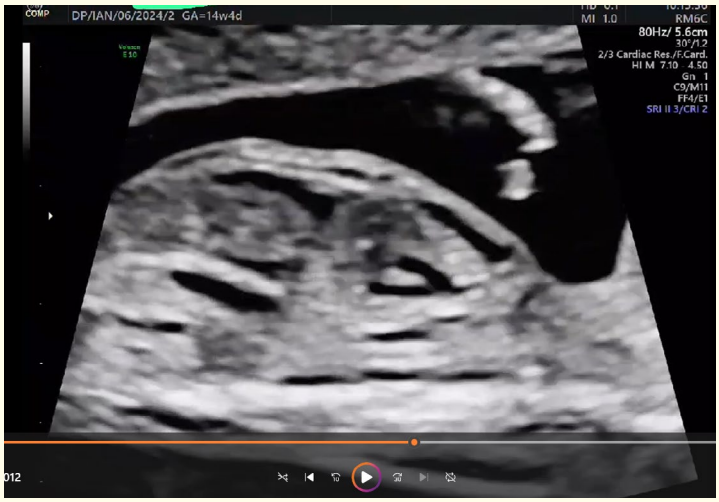
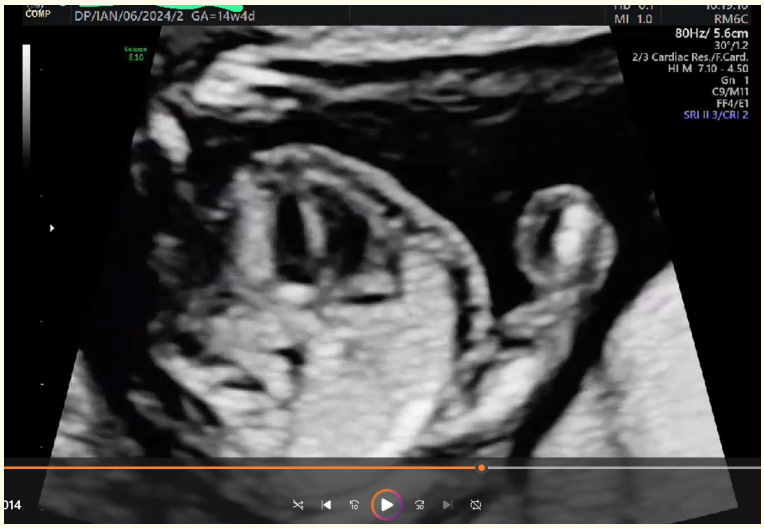
We informed the parents about the results, about the association of aneuploidy and send them to genetic counselling.

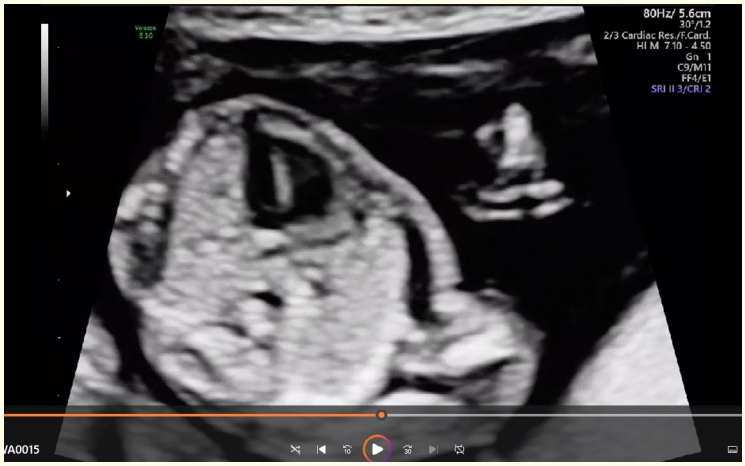
They decided for TOP but she did the termination in another clinic and they did not provide the fetus for anatomopathological exam of the heart [2-7].



Video 1



Video 2



Video 3

Conclusion

The heart examination is not so easy to perform between 11 - 14 weeks of GA and that's the reason this is a challenging examination that demands a great expertise and should be performed by trained examinators.

The scan of the heart include systematic approach and should abide to protocols of ISUOG and FMF in order to have false positive and false negative as low as the examination done at mid trimester.

These are possible nowadays with the advances of 2D and CD technologies and with adequate training.

It is important the involvement of a fetal and pediatric cardiologist as well as the geneticians in the standard practice.

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