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PHENOTYPIC EXPRESSION OF THE SYNDROMIC STRUCTURAL HEART DEFECTS

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ABSTRACT

Atrioventricular canal defect is a congenital heart malformation characterized by a variable deficiency of the atrioventricular area in the developing heart. The focus of this study is to highlight the benefits of the cytogenetic examination, in the correct management of trisomy 21 syndrome associated with atrioventricular canal defect, early prenatally detected by ultrasound examination at 22 weeks of pregnancy. As a direct consequence, prenatal ultrasound detection and genetic testing are crucial for the diagnosis of rare genetic birth malformations.

KEYWORDS: trisomy 21, atrioventricular canal defect, amniocentesis ultrasound examination, karyotype, prenatal diagnosis.

INTRODUCTION

According to World Health Organization, the most common type of major congenital malformations is heart defects, neural tube defects, and Down syndrome.[1]

Atrioventricular canal defect (AVSD) is a congenital heart malformation characterized by a variable deficiency of the atrioventricular area in the developing heart. The defect occurs in the intrauterine life in the stage of development of the formations that will later form the intracardiac septa that separate the four cardiac cavities. The mitral and tricuspid valves, which separate the atria from the ventricles, are also affected.[2-5]

There are two general types of AVSD that can occur, depending on which structures are not formed correctly: complete AVSD and partial or incomplete AVSD.[6]

The causes of AVSD, among most babies, are unknown.[7,8]

The focus of this study is to highlight the benefits of the cytogenetic examination, in the correct management of extreme congenital birth defects, early prenatally detected by ultrasound examination.

MATERIALS AND METHODS

At 22 weeks of pregnancy, a 31-year-old European woman was referred to a private health clinic in Bucharest, Romania, for a routine antenatal ultrasound examination. The parents were not consanguineous and had no inherited or family-related pathologies.

The ultrasound evaluation was carried out by our medical center's Division of Maternal-Fetal Medicine and Obstetrics, in agreement with the Declaration of Helsinki - Ethical Principles and Good Clinical Practices, and with the patient's informed consent, using a General Electric Voluson E10 ultrasound device.

RESULTS

The sonographic investigation revealed a unique fetus 22.6 weeks old, in evolution, with an estimated fetal weight of 556 g (Fig. 1).

EFW (Hadlock)	*Value*	*Range*	*Age*	*Range*	*GP (Hadlock)*
AC/BPD/FL/HC	**556g**	**±95 g**	**22w6d**		**71.3%**

Figure 1: Estimated fetal weight (EFW) of the fetus.

(BPD, biparietal diameter; FL, femur length; HC, head circumference; AC, abdominal circumference).

Fetal head ultrasound biometry showed biparietal diameter: 59.7 mm, occipitofrontal diameter: 68.8 mm, and head circumferences: 202 mm, indicating a normal configuration of the fetal brain.

The sonographic scan of the thorax highlight normal size, shape, and structure with an anterior-posterior diameter of 54.2 mm and a transverse diameter of 54.1 mm.

Fetal echocardiography revealed a fetal heart with four-chamber, aspect of atrioventricular canal defect, and fetal heart rate: 150 beats/min (Fig. 2 - 5).

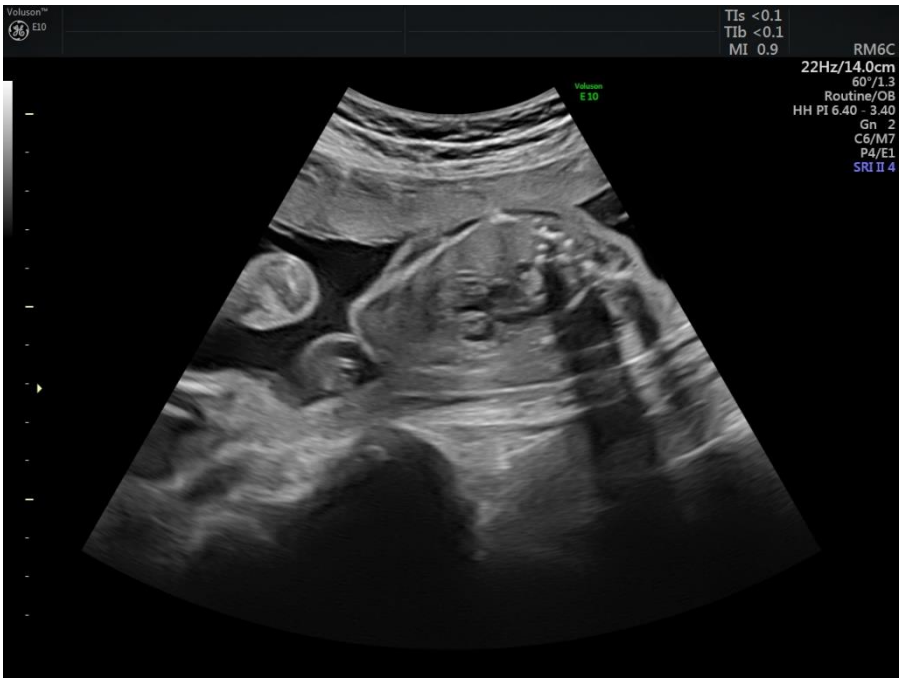

Figure 2: Fetal echocardiography showing a fetal heart with four-chamber.

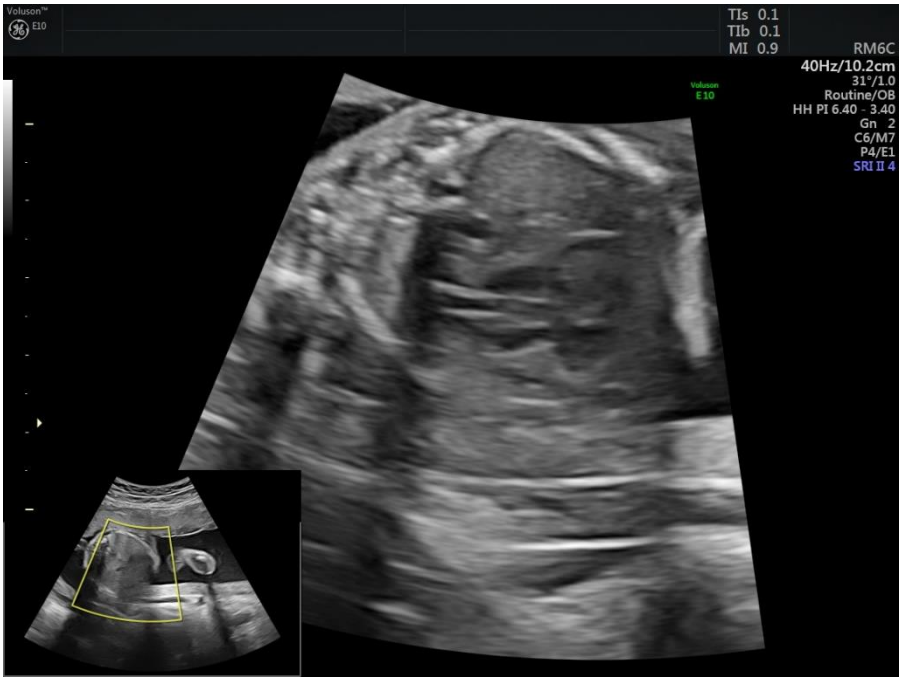

Figure 3: 2D Fetal echocardiography showing the atrioventricular canal defect.

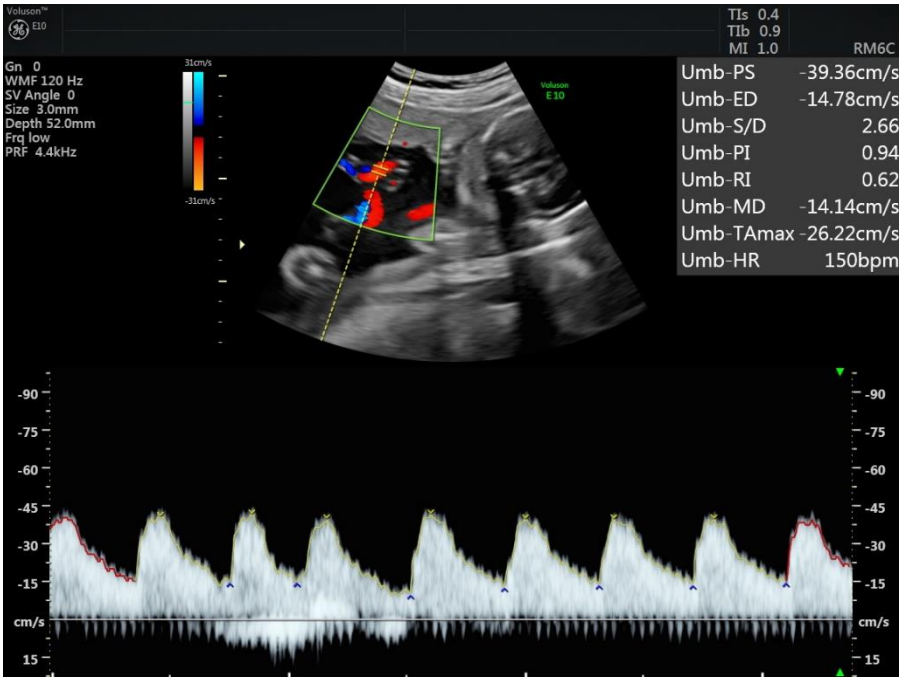

Figure 4: 2D Fetal Doppler echocardiography: Doppler measurements.

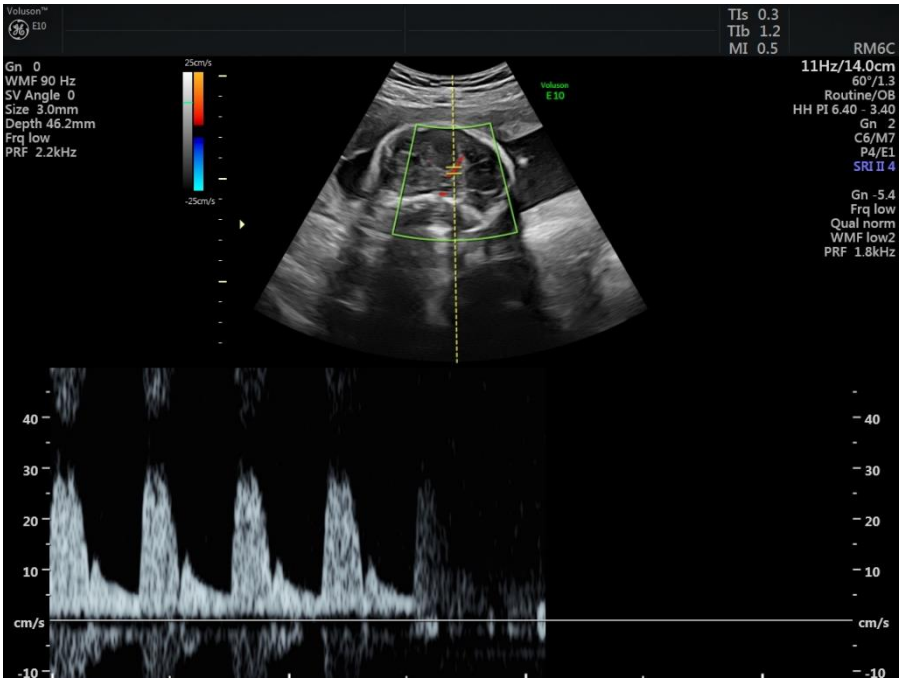

Figure 5: 2D Fetal Doppler echocardiography showing the atrioventricular canal defect.

The ultrasound examination of the abdomen and limbs showed apparently normal shape and structure. No other fetal dysmorphisms over 0.5 cm were found.

Amniocentesis was performed two days later, and cytogenetic analysis of the fetal chromosomes indicated a fetal karyotype: 47, XX, +21 (Figure 6).

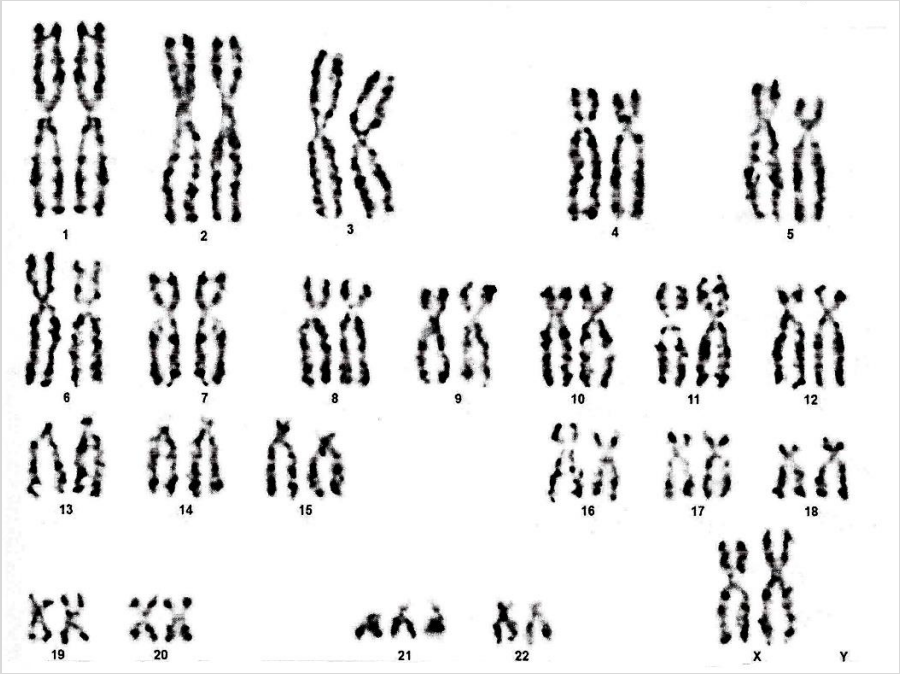

Figure 6: Fetal karyotype: 47, XX,+21.

The following diagnosis was made based on the fetal morphology scan and cytogenetic analysis of the fetal chromosomes: Mono-fetal pregnancy 22.3 weeks of gestation (chronologic) / 23.5 weeks of gestation (biometric), in evolution: Trisomy 21 (Down syndrome). Atrioventricular canal defect.

The parents were informed about the gravity of fetal abnormalities and decide to end the pregnancy with Down syndrome and atrioventricular canal defect for medical reasons. Anatomic pathology exploration of the aborted fetus certified the prenatal ultrasound diagnosis.

DISCUSSION

Trisomy 21, also known as Down's syndrome, with a frequency of 1‰ live births, is a genetic disorder in which a person has 47 chromosomes instead of 46, with a supplementary copy of chromosome 21.[9,10]

Trisomy 21 is frequently associated with various congenital malformations, including congenital intellectual disability, hypotonia, heart defects, as well as other multisystem congenital defects.[11-13]

Congenital heart defects decrease the viability of patients with Down syndrome by 72%.[14] As a direct consequence, prenatal ultrasound detection and genetic testing are crucial for the diagnosis of rare genetic birth malformations.[15,16]

CONCLUSION

Any isolated structural congenital malformation detected prenatally, must be thoroughly investigated, prenatal genetic investigations being crucial in early, correct and complete, prenatal diagnosis, as well as efficient case management.

Authors' contributions

All authors contributed equally with the first-author, in the preparing, review and editing of the article. All authors read and approved the final version of the manuscript.

REFERENCES

1. Albu CC, Vasilache A, Tanase M, Stanciu IA, Albu DF, Albu SD. The power of non-invasive prenatal diagnosis in the early detection of severe structural birth defects. World J Pharm Res, 2021; 10(5): 39-46. DOI: 10.20959/wjpr20215-20315.
2. Briggs LE, Kakarla J, Wessels A. The pathogenesis of atrial and atrioventricular septal defects with special emphasis on the role of the dorsal mesenchymal protrusion. Differentiation, 2012; 84(1): 117-30. DOI: 10.1016/j.diff.2012.05.006.
3. Craig B. Atrioventricular septal defect: from fetus to adult. Heart, 2006; 92(12): 1879-85. DOI:10.1136/hrt.2006.093344.
4. Pugnaloni F, Digilio MC, Putotto C, De Luca E, Marino B, Versacci P. Genetics of atrioventricular canal defects. Ital J Pediatr, 2020; 46(1): 61. DOI:10.1186/s13052-020-00825-4.
5. Lana-Elola E, Watson-Scales S, Slender A, Gibbins D, Martineau A, Douglas C, Mohun T, Fisher EM, Tybulewicz VL. Genetic dissection of Down syndrome-associated congenital heart defects using a new mouse mapping panel. Elife, 2016; 5: e11614. DOI:10.7554/eLife.11614. Erratum in: Elife, 2020; 9.
6. Rigby M. Atrioventricular Septal Defect: What Is in a Name? J Cardiovasc Dev Dis, 2021; 8(2): 19. DOI:10.3390/jcdd8020019.
7. Layangool T, Sangtawesin C, Kirawittaya T, Prompan W, Prachasilchai P, Pechdamrongsakul A. Survival analysis of Down syndrome with congenital heart disease: a 5-years registry at QSNICH. J Med Assoc Thai, 2014; 97(6): S108-114.

8. Blue GM, Kirk EP, Sholler GF, Harvey RP, Winlaw DS. Congenital heart disease: current knowledge about causes and inheritance. Med J Aust, 2012; 197(3): 155-9. DOI:10.5694/mja12.10811.
9. Albu CC, Albu D, Albu S, Patrascu A, Musat A, Goganau AM. Early prenatal diagnosis of an extremely rare association of Down syndrome and transposition of the great vessels. Rev. Chim.[internet], 2019; 70(7): 2574-2578. DOI:10.37358/RC.19.7.7383.
10. Weijerman ME, de Winter JP. Clinical practice. The care of children with Down syndrome. Eur J Pediatr, 2010; 169(12): 1445-52. DOI:10.1007/s00431-010-1253-0.
11. . Elmagrpy Z, Rayani A, Shah A, Habas E, Aburawi EH. Down syndrome and congenital heart disease: why the regional difference as observed in the Libyan experience? Cardiovasc J Afr, 2011; 22(6): 306-9. DOI:10.5830/CVJA-2010-072.
12. Ko JM. Genetic Syndromes associated with Congenital Heart Disease. Korean Circ J, 2015; 45(5): 357-61. doi:10.4070/kcj.2015.45.5.357.
13. Colvin KL, Yeager ME. What people with Down Syndrome can teach us about cardiopulmonary disease. Eur Respir Rev, 2017; 26(143): 160098. DOI:10.1183/16000617.0098-2016.
14. de Rubens Figueroa J, del Pozzo Magaña B, Pablos Hach JL, Calderón Jiménez C, Castrejón Urbina R. Malformaciones cardíacas en los niños con síndrome de Down [Heart malformations in children with Down syndrome]. Rev Esp Cardiol, 2003; 56(9): 894-9. Spanish. DOI:10.1016/s0300-8932(03)76978-4.
15. Carlson LM, Vora NL. Prenatal Diagnosis: Screening and Diagnostic Tools. Obstet Gynecol Clin North Am, 2017; 44(2): 245-256. DOI:10.1016/j.ogc.2017.02.004.
16. Albu CC, Albu DF, Albu SD. The Potential of Prenatal Diagnosis in the Early Detection of Congenital Malformations. Int J Med Res Rev, 2021; 9(1): 54-57. DOI:10.17511/ijmrr.2021.i01.08.