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EARLY NON-INVASIVE DIAGNOSIS OF FETAL ANENCEPHALY

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ABSTRACT
Anencephaly, the most severe form of cranial neural tube malformation, is a multifactorial disease which occurs as a consequence of the interaction between the genetic factors and the environmental factors. We are presenting here a particular case, of a patient aged 32 years old, Caucasian, clinically healthy, without pathological personal antecedents and without heredocolateral antecedents, pregnant for the first time, presents to A. S. Medical Center, from Bucharest, Romania, for a routine ultrasound examination. According to the first trimester ultrasonography, the following ultrasound diagnosis was established: Monofetal preganancy of 12 weeks, in evolution. Fetal anencephaly, a defect of the closure of the neural tube, early prenatally diagnosed and confirmed postnatally. The fetal ultrasound examination is a non-invasive prenatal diagnosis method, today very performant, cost-effective, and fast, through which the severe malformations can be diagnosed successfully by prenatal ultrasound screening, early, in the first trimester of pregnancy.

KEYWORDS: anencephaly, ultrasound examination, prenatal diagnosis.

INTRODUCTION
Anencephaly is a defect of the closure of the neural tube, with a prevalence from 1 to 1,000 pregnancies.[1] It is a multifactorial disease which occurs as a consequence of the interaction between the genetic factors and the environmental factors.[2] Several genes were identified which seem to be related to the occurrence of the anencephaly, among which the best studied is the MTHFR gene.[3] Other factors involved in the etiology of anencephaly are: the insufficient exogenous intake of folic acid during the pregnancy, the administration of valproic acid, the fever, especially during the first months of pregnancy, as well as decompensated insulin-dependent diabetes mellitus.[4]

In 90% of the cases, anencephaly occurs rarely, without prior family history of the disease.[5,6] A family aggregation of the cases is registered only in a low percentage, the mechanism not being known completely.[7]

MATERIALS AND METHODS
Patient aged 32 years old, Caucasian, clinically healthy, without pathological personal antecedents and without heredocolateral antecedents, pregnant for the first time, presents to A.S. Medical Center, from Bucharest, Romania, for a routine ultrasound examination.

After obtaining the consent of the patient regarding the fetal ultrasound examination, the investigation was performed with a Voluson E10 Ultrasound, state-of-art device, by an expert in maternal-fetal ultrasound examination.

RESULTS
The transabdominal ultrasound examination revealed a single fetus of 12 weeks, in evolution, with small head, underdeveloped brain and skull bones (Fig. 1).

The biometric determinations of the fetus pointed the crown-rump length (CRL): 5.20 cm, significantly reduced, and the femur length (FL): 1.13 cm. The Doppler measurements performed at the level of the umbilical artery revealed the fetal heart rate (HR) = 167 bpm (Fig. 2).

The ultrasound investigation continued with the exploration of the rest of the fetal body, which pointed out the following: thorax normally shaped, regular aspect heart, regular size abdomen, liver, spleen and kidneys with regular structure and sizes, normally shaped upper

and lower limbs, umbilical cord and placenta with regular aspect, without other congenital anomalies visible in the ultrasound examination.

According to the fetal morphology, the following ultrasound diagnosis was established: Monofetal preganancy of 12 weeks, in evolution. Fetal anencephaly (Fig. 3 and Fig. 4).

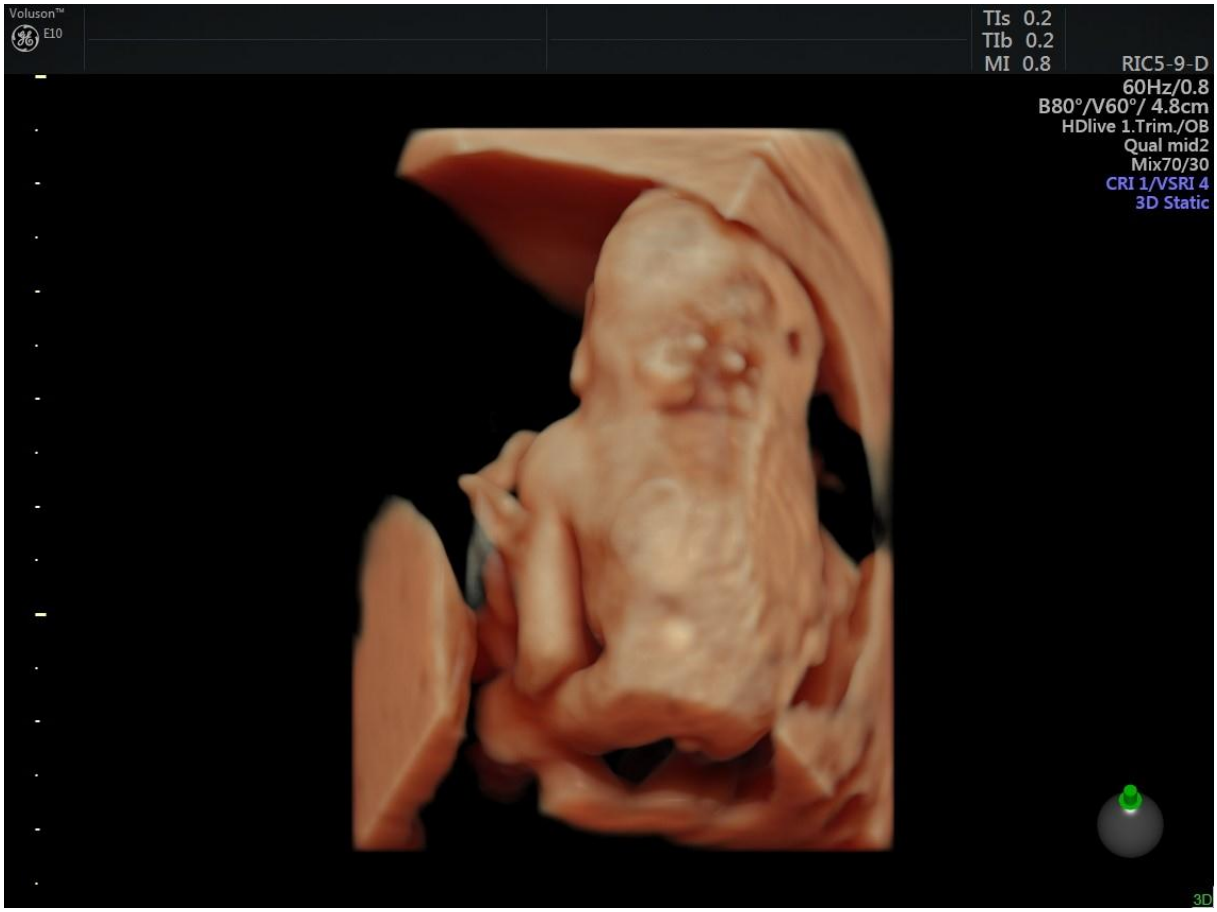

Figure 1: Fetal anencephaly. First trimester ultrasonography, 3D Static examination.

2D Measurements	AUA	Value	m1	m2	m3	Meth.	GP	GA
FL (Hadlock)	☑	1.13 cm	1.13			avg.	18.3%	13w2d
CRL (Hadlock)	☑	5.20 cm	5.20			avg.	<1%	11w6d

Doppler Measurements	Value	m1	m2	m3	m4	m5	m6	Meth.
Umbilical Art.								
HR	167 bpm	167						max

Figure 2: Biometric determinations of the fetus: femur length (FL), crown-rump length (CRL) and fetal heart rate (HR).

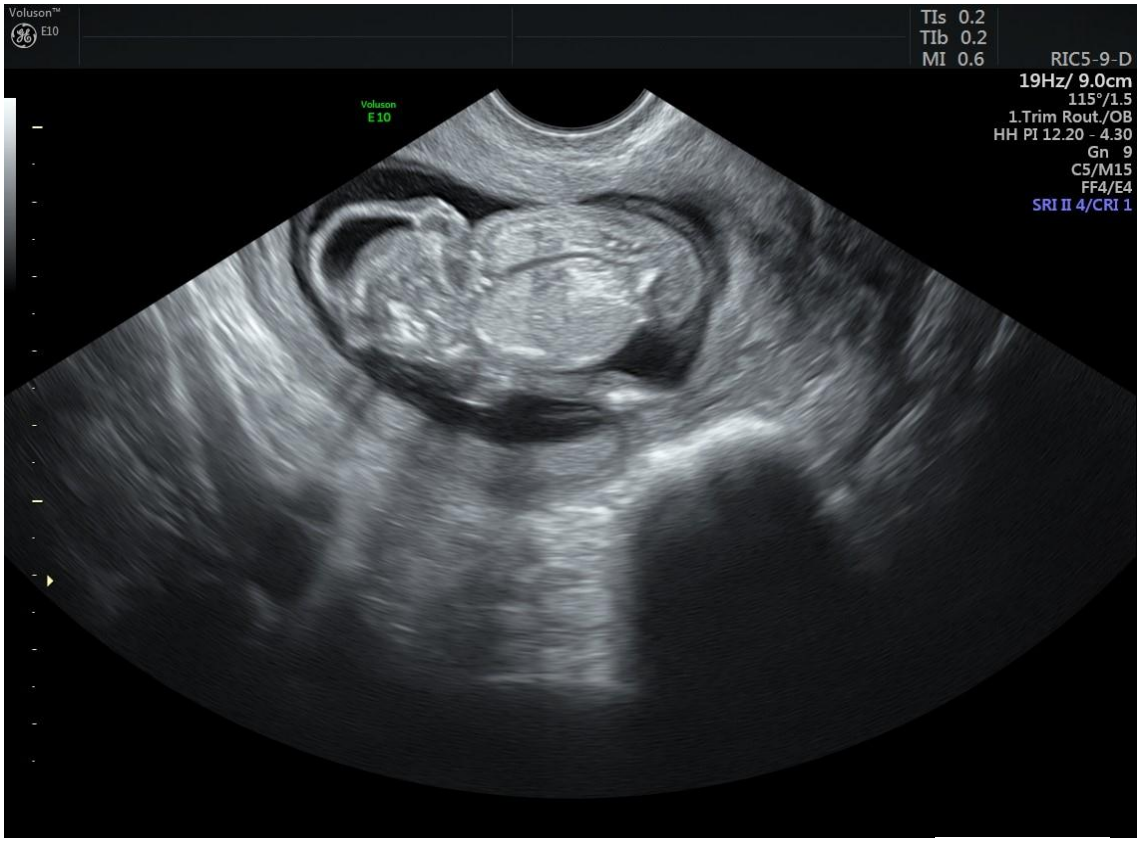

Figure 3: Fetal anencephaly: First trimester ultrasonography, 2D examination, sagittal section.

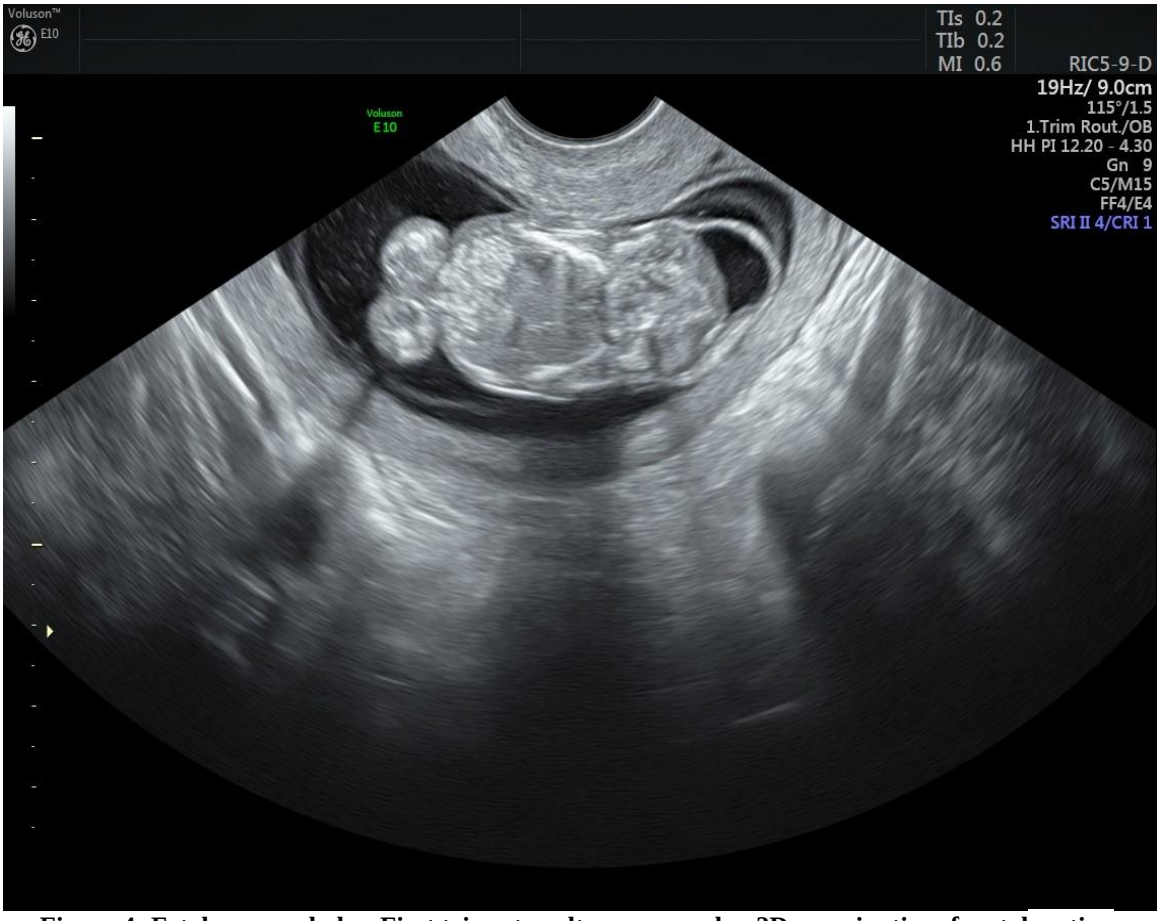

Figure 4: Fetal anencephaly - First trimester ultrasonography, 2D examination, frontal section.

Considering the severity of the fetal malformation, the patient was genetically counseled. She was described the fetal malformation and presented the prognosis of the disease, as described in the specialty literature. Following the genetic consultation and the genetic counseling, the patient, understanding the gravity of the fetal malformation, chose to terminate the pregnancy, for medical reasons, and subsequently she shall perform additional genetic investigations to prevent the occurrence of a new abnormal pregnancy. The fetal anatomic-pathological examination confirmed the prenatal diagnosis.

DISCUSSION

The anencephaly diagnosis can be set prenatally, through laboratory tests, represented by the determination of the alpha-fetoprotein in the maternal serum or in the amniotic liquid, sampled by amniocentesis.[8-10] The increased values of the alpha-fetoprotein are associated with an increased risk of fetal anencephaly.[11]

The fetal ultrasound examination is a non-invasive prenatal diagnosis method, through which the fetal anencephaly can be diagnosed successfully in the first trimester of pregnancy.[12-15]

The differential diagnosis of anencephaly can be done with the acrania, the microcephaly or the encephalocele.[16, 17] In the case of the parents who have had a child born with anencephaly, the risk for the second baby to be a newborn with anencephaly ranges between 4% and 5%. If there are two children born with anencephaly in antecedents, the risk for the third baby to be affected increases up to 10%-13%.[18]

Prophylactically, the patients who presented pregnancies with anencephaly in antecedents require the administration of folic acid, both during the evolution of the pregnancy and 1-2 months before conception.[19]

CONCLUSION

The fetal ultrasound examination is a non-invasive prenatal diagnosis method, today very performant, cost-effective, and fast, through which the fetal anencephaly can be diagnosed successfully by prenatal ultrasound screening, early, in the first trimester of pregnancy.

Prenatal screening through the first trimester ultrasound examination has a crucial role in early prenatal diagnosis of severe congenital malformations, incompatible with postnatal life.

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.

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