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THE IMPORTANCE OF FIRST TRIMESTER ULTRASOUND EXAMINATION IN THE EARLY DIAGNOSIS OF A RARE CONGENITAL MALFORMATION

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ABSTRACT
We present the clinical case of a 34-year-old, Caucasian woman, pregnant for the first time, who was referred to our clinic for a routine ultrasound screening in the first trimester of pregnancy. Ultrasound examination showed a singleton fetus with one a most severe and rare congenital malformation: fetal acrania. After a personalized genetic counselling the parents opted to terminate the pregnancy. The autopsy confirms the prenatal ultrasound diagnosis.

KEYWORDS: acrania, ultrasound examination, prenatal diagnosis, congenital malformation.

INTRODUCTION
Neural tube defects are a group of congenital malformations of the central nervous system, with a prevalence of 1/100 to 1/1000 of pregnancies and pluryfactorial causes.[1] Fetal acrania is one a most rare congenital malformation, with a frequency less than 1 per 100,000 births, incompatible with extrauterine life and characterized by the partial or complete absence of skull bones surrounding the fetal brain.[2-4] The acrania pathogenesis is unknown. Genetic counselling is very difficult because there is no evidence for a specific genetic origin, but the extreme rarity and sporadic nature suggests that their occurrence depends on genetic factors conditioned by polygenic inheritance and environmental factors.[1,5]

MATERIALS AND METHODS
A 34-year-old, caucasian woman, gravida 0, para 0, was referred to our private medical center, from Bucharest, Romania, at 11 weeks' gestation for a routine prenatal ultrasound screening. The couple was non-consanguineous and clinically healthy. The ultrasound examination was performed using General Electric Voluson E10 Ultrasound system.

RESULTS
First trimester ultrasound examination at 11 weeks of gestation revealed a singleton live fetus with crown-rump length (CRL), measurement from the top of the head to the bottom of the buttocks, 4.67 cm (Fig. 1 and Fig. 2).

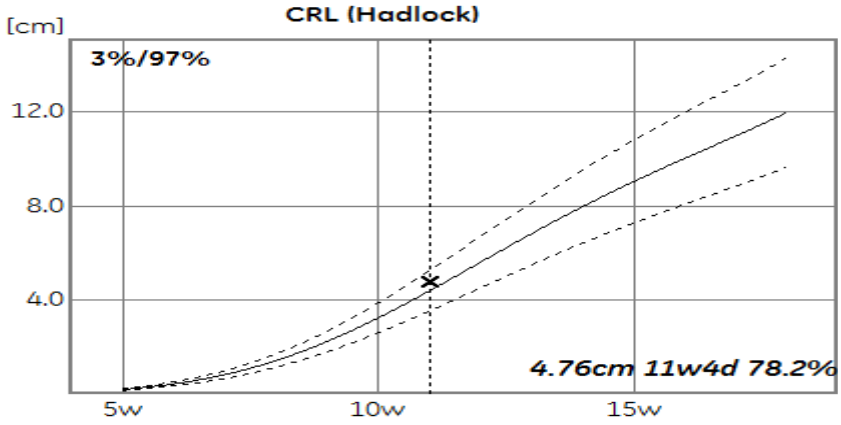

Figure 1: Graph: Fetal Crown-Rump Length (CRL), (Hadlock).

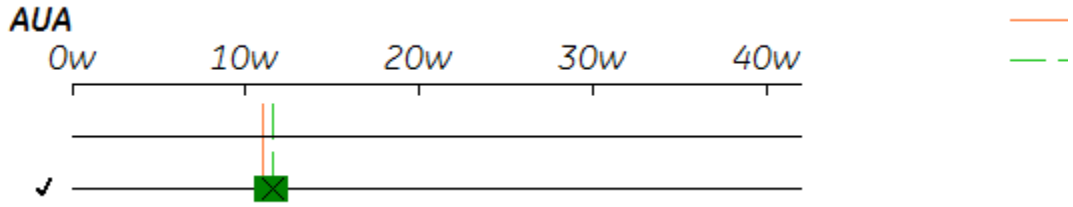

Figure 2: Bar Graph: Fetal Crown-Rump Length (CRL), (Hadlock)
Gestational age (Last Menstrual Period); GA (Actual Ultrasound Age).

At the level of the cephalic extremity we noticed the absence of a skull cap with herniated cerebral elements, which makes it is impossible to evaluate the biparietal diameter (Fig. 3 and Fig. 4).

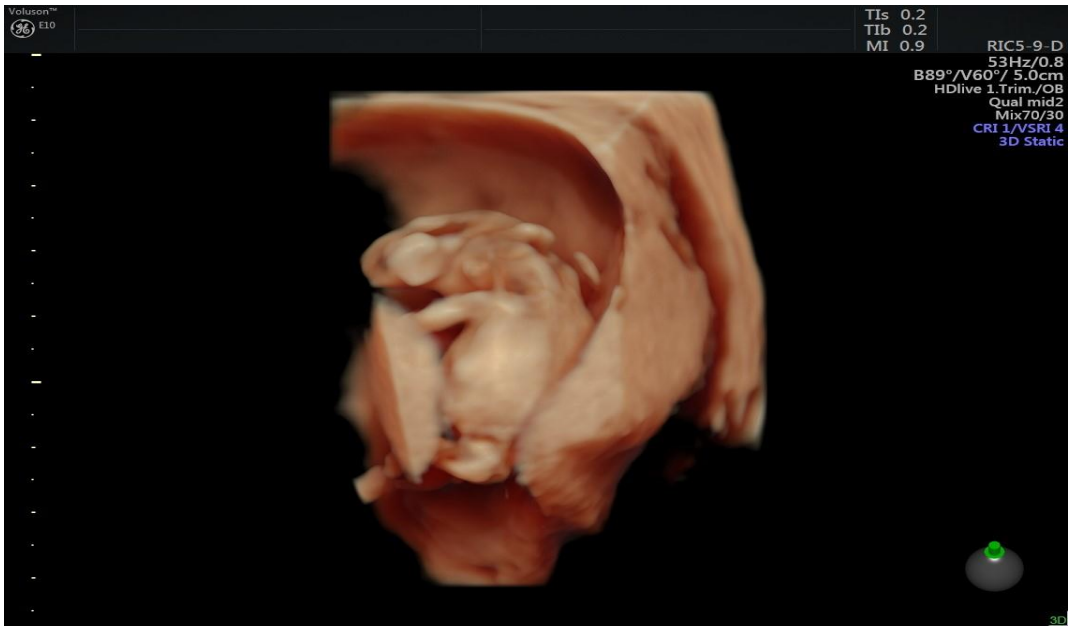

Figure 3 – Fetal Acrania.
3D Static Ultrasound examination.

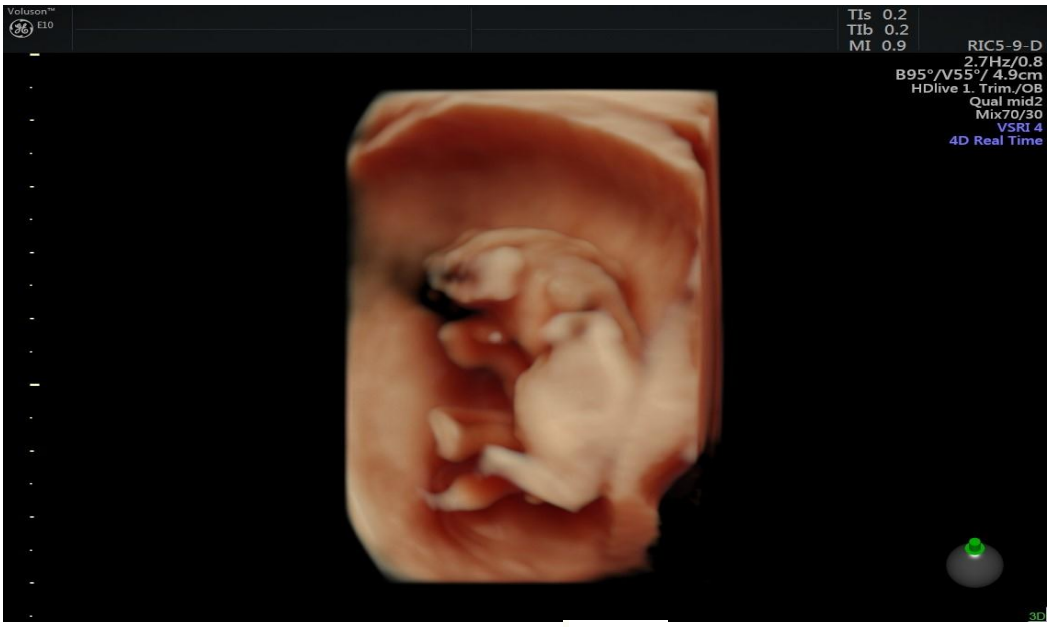

Figure 4 – Fetal Acrania.
4D Real Time Ultrasound examination.

The nuchal translucency of the fetus was 2.5 mm and the fetal cardiac-circulatory activity present with an average fetal heart rate 187 beats per minute with normal rhythm. The fetal spine was apparently normal and the movements of the fetal body were normal and well coordinated. No other fetal anomaly was seen. After the prenatal first trimester ultrasound examination the following diagnosis was established: Pregnancy 11 weeks in evolution. Acrania. After a personalized genetic counselling the parents opted to terminate the pregnancy. The fetus was therapeutically aborted. The fetal autopsy was confirmed the prenatal ultrasound diagnosis.

DISCUSSION

Fetal acrania is an uncommon developmental congenital anomaly with a reserved prognosis and still unclear etiology, occurs during the beginning of the fourth week of development.[6] The fetal acrania is the result of a complex process involving genetics and environmental factors. Differential diagnosis of fetal acrania should be made with anencephaly, encephalocele, osteogenesis imperfecta type II, hypophosphatasia and amniotic band syndrome.[7,8] First trimester ultrasound examination has the potential to identify embryo-fetal defects in pregnant women who are at increased risk of congenital anomalies.[9] Prenatal diagnosis of fetal acrania can be established by ultrasound scan in the first trimester of pregnancy, which indicates the absence of the calvarium, and a large mass of disorganized brain tissue.[8,10,11] Prenatal diagnosis of fetal acrania is important to management the affected pregnancy as early as possible, to prevent the complication and avoid the negative psychological impact on patients.

CONCLUSION

The first trimester ultrasound examination was decisive in the prenatal diagnosis and management of fetal acrania, early at 11 weeks of gestation.

Authors' contributions

All authors contributed equally to preparing, review and editing of the article.
All authors read and approved the final version of the manuscript.

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