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THE POWER OF NON-INVASIVE PRENATAL DIAGNOSIS IN THE EARLY DETECTION OF SEVERE STRUCTURAL BIRTH DEFECTS

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ABSTRACT

Severe congenital anomalies have a significant impact on neonatal, infant, and childhood mortality and morbidity. We are presenting here a novel phenotypic variety of isolated, sporadic, non-hereditary birth defect, which associates partial agenesis of the corpus callosum, mandibular hypoplasia, cystic hygroma of the neck, transposition of the great vessels and bivascular umbilical cord, and which was successfully diagnosed prenatally by ultrasound examination at 19 weeks of pregnancy, being probably caused by a de novo spontaneous mutation or a disorder of embryogenesis of a non-genetic nature.

KEYWORDS: birth defect, mandibular hypoplasia, cystic hygroma, agenesis of the corpus callosum, ultrasound examination, prenatal diagnosis.

INTRODUCTION

Severe congenital anomalies have a significant impact on neonatal, infant, and childhood mortality and morbidity.[1-3] According to World Health Organization, the most common type of major congenital malformations are heart defects, neural tube defects, and Down syndrome.[4-7]

The spectrum and prevalence of severe birth defects may vary over time or with geographical area, reflecting the interaction between genetic and non-genetic factors.[8-10]

The aim of the present study is to present the benefits of ultrasound examination and the importance of early prenatal diagnosis of severe congenital malformations.

MATERIALS AND METHODS

A 39-year-old, caucasian woman, pregnant for the first time, was referred at 17.3 weeks of gestation in a private medical center in Bucharest, Romania, for a routine prenatal ultrasound examination of the fetal malformations.

The couple was non-consanguineous and clinically healthy, without hereditary or familial pathologies.

The ultrasound examination was performed after the patient was informed about the examination with her informed consent for the ultrasonographic investigation.

The fetal sonography was performed with a General Electric Voluson E8 Ultrasound system, by an expert ultrasonographer with competence in maternal-fetal medicine.

RESULTS

The ultrasound examination highlights a unique fetus 17.3 weeks old, in evolution, with an estimated fetal weight of 200 g.

A detailed ultrasound evaluation of the fetus showed numerous and significant malformations.

At the level of the cephalic extremity: the biparietal diameter was 40.8 mm, the occipitofrontal diameter was 48.9 mm and head circumference was 140.7 mm.

Fetal neurosonography and biometry of the corpus callosum by three-dimensional ultrasound examination showed partial agenesis of the corpus callosum and second trimester ultrasound scan of fetal viscerocranium highlight mandibular hypoplasia (Fig. 1 and Fig. 2). Also, a cystic hygroma of the neck (8.2 mm) was detected (Fig. 3 - Fig.5).

The thorax showed apparently normal shape and structure, anterior-posterior diameter: 31 mm and transverse diameter: 38 mm.

Fetal echocardiography pointed tetracameral heart with transposition of the great vessels and reversed A-wave in fetal ductus venosus.

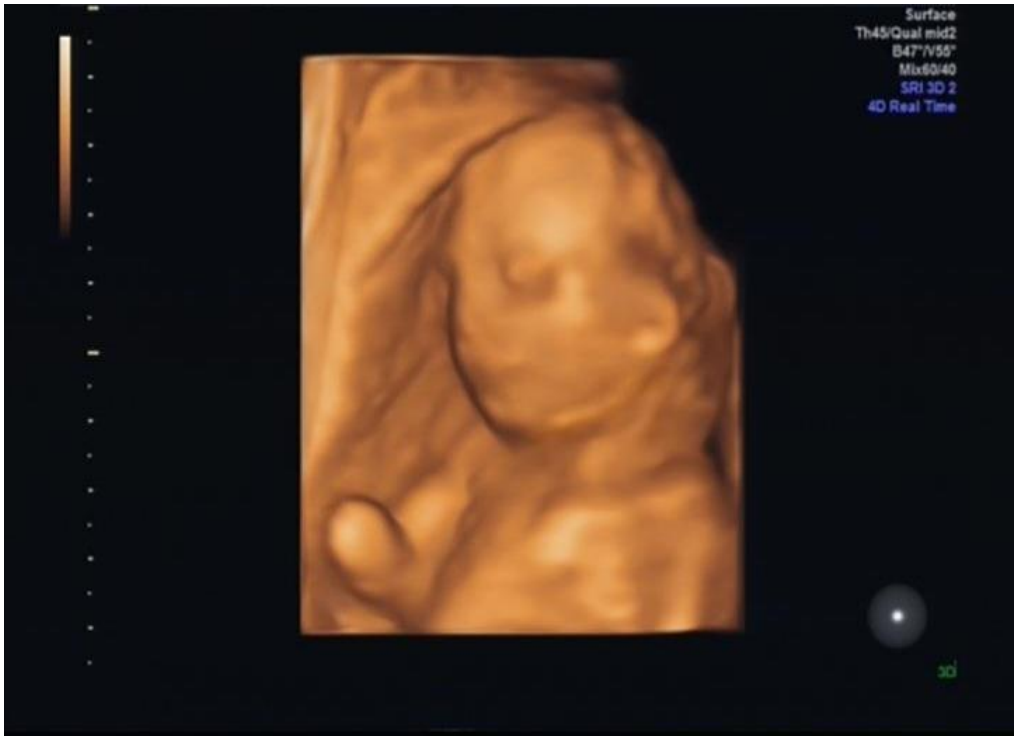

Figure 1 - 4D Real Time ultrasound examination: Mandibular hypoplasia.

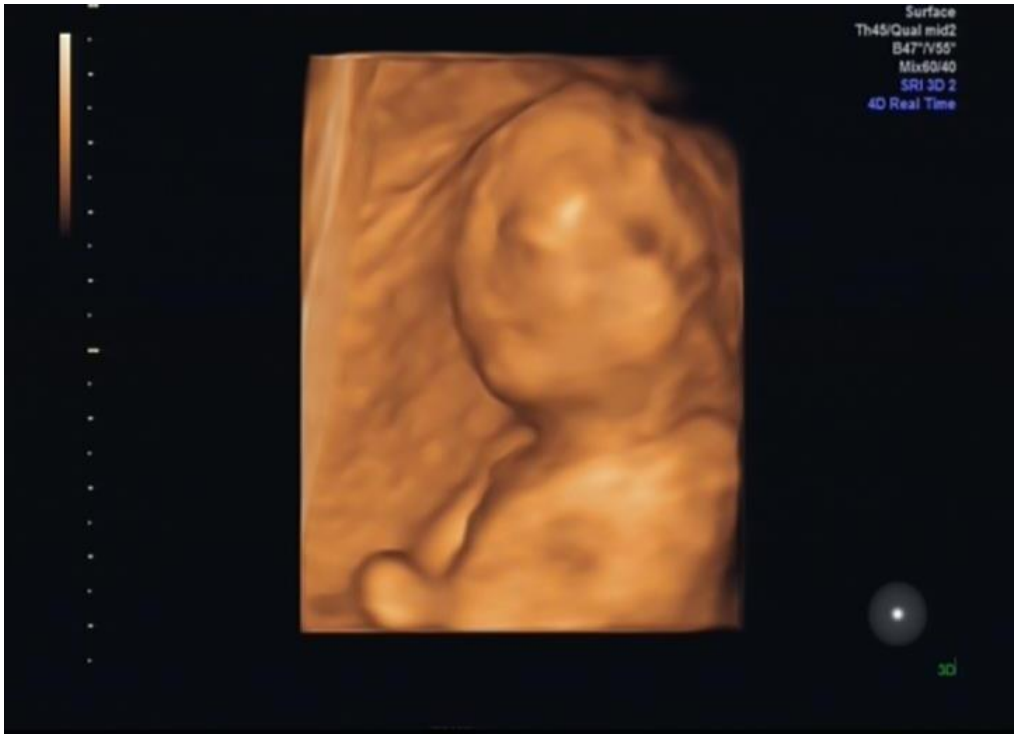

Figure 2 - 4D Real Time ultrasound examination: Mandibular hypoplasia.

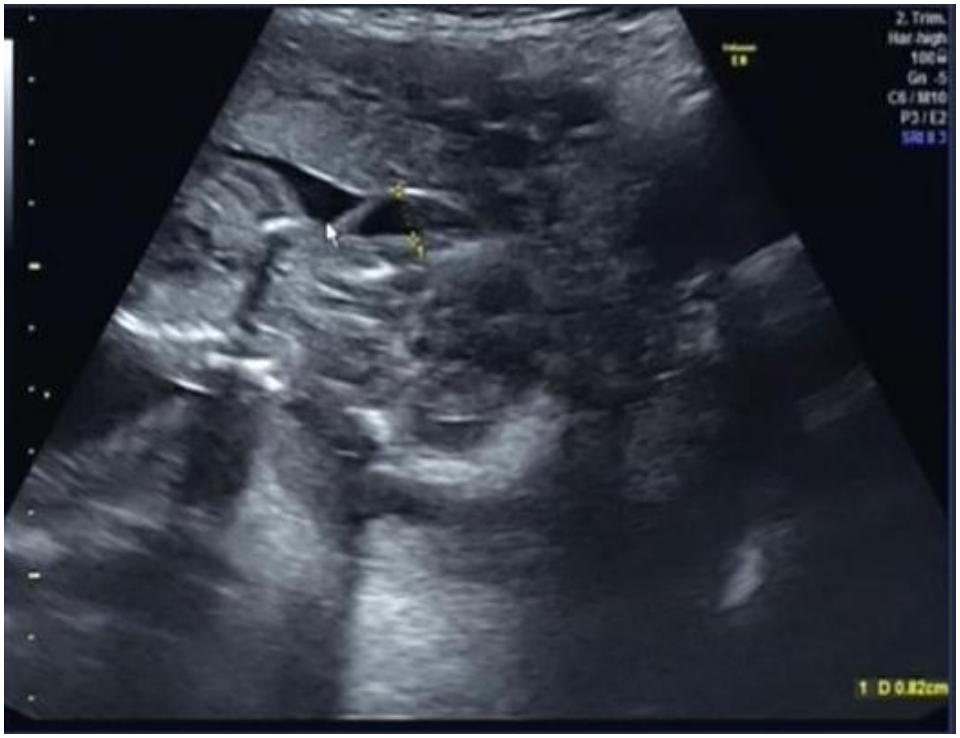

Figure 3 - 2D Ultrasound examination: Cystic hygroma of the neck.

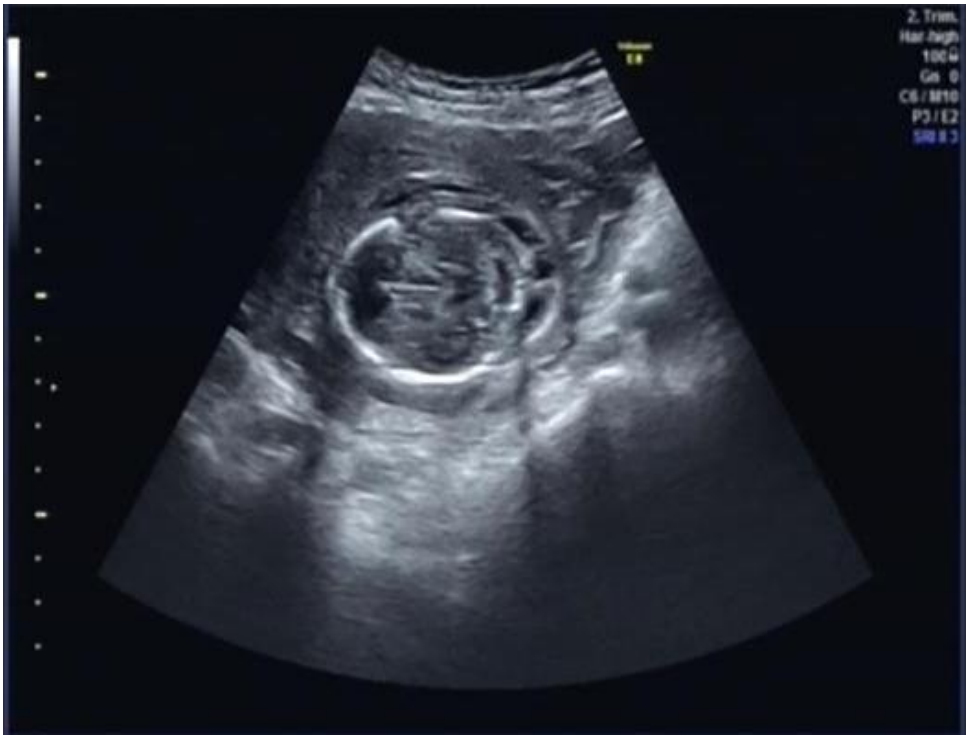

Figure 4 - 2D Ultrasound examination: Cystic hygroma of the neck - transverse view.

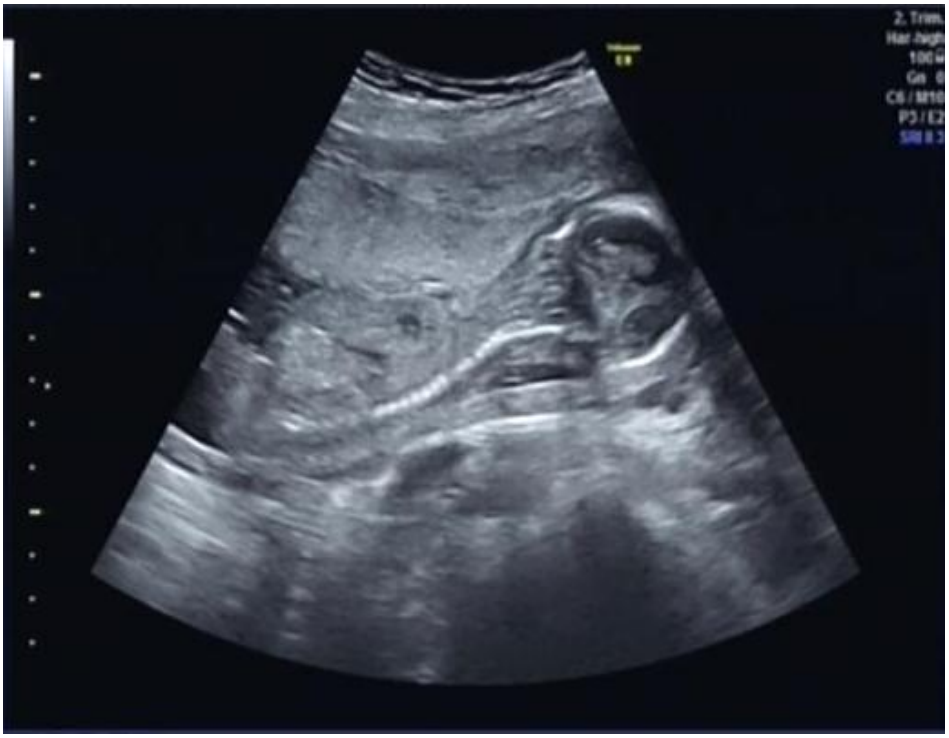

Figure 5 - 2D Ultrasound examination: Cystic hygroma of the neck - mid-sagittal view.

Fetal abdominal ultrasound illustrated ileal hyperechogenicity and bivascular umbilical cord (Fig. 6). No other fetal dysmorphisms over 0.5 cm were found.

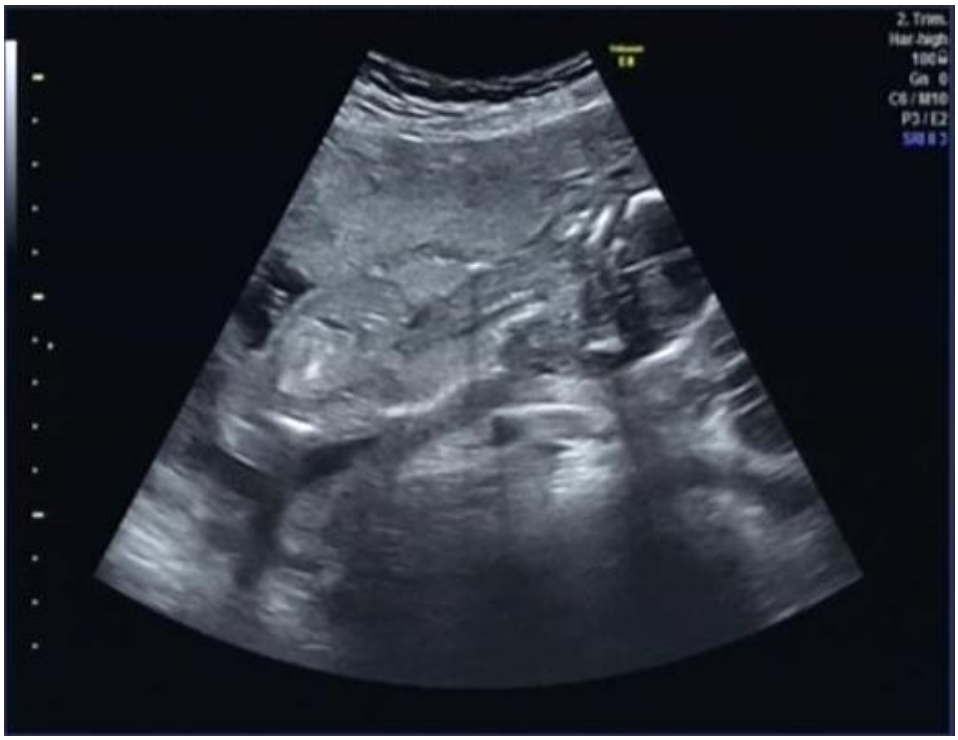

Figure 6 - 2D Ultrasound examination: Ileal hyperechogenicity.

In conformity with the fetal morphology scan the following diagnosis was established: Mono-fetal pregnancy 17.3 weeks' of gestation, in evolution with multiple congenital anomalies: Partial agenesis of the corpus callosum. Mandibular hypoplasia. Cystic hygroma of the neck. Transposition of the great vessels. Ileal hyperechogenicity. Bivascular umbilical cord.

The parents were informed about the severity of fetal malformations and decide to ending the pregnancy for medical reasons. Anatomic pathology examination of the aborted fetus confirmed the prenatal ultrasound diagnosis.

DISCUSSION

Fetal structural birth defects are found in up to 3% of all pregnancies, and approximately one-half of all major structural malformations can be detected early by sonography in the first trimester of pregnancy.[11-12]

In Europe, ultrasound examination for fetal abnormalities is increasingly becoming part of routine prenatal care, generally, the majority of ultrasound screening for fetal structural abnormalities being recommended at 19-21 weeks of gestational age.[13-14]

We consider that the present case represents a new phenotypic complex of birth defect, isolated, sporadic, non-hereditary, successfully detected prenatally by ultrasound examination at 17.3 weeks of pregnancy, being probably caused by a de novo mutation or a disorder of embryogenesis of a non-genetic nature.

Genetic counseling after prenatal diagnosis of complex structural birth defects is very difficult because of the bad prognosis, but the early prenatal diagnosis of extremely severe birth defects is crucial for the prevention of newborns with congenital disabilities.[15-17]

CONCLUSION

A novel phenotypic variety of birth defect, which associates partial agenesis of the corpus callosum, mandibular hypoplasia, cystic hygroma of the neck, transposition of the great vessels, and bivascular umbilical cord, highlight the power of non-invasive ultrasound prenatal diagnosis in the early detection and management of severe structural birth defects.

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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