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NEW PHENOTYPIC VARIETY OF FRONTONASAL DYSPLASIA SYNDROME

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ABSTRACT
Frontonasal dysplasia represent a very rare and complex spectrum of genetic condition, with unknown prevalence, that affect both sexes equally. We are presenting here a new phenotypic variety of frontonasal dysplasia, which associates occipital meningoencephalocele with bilobed nasal pyramid, and bivascular umbilical cord, which was successfully diagnosed prenatally by ultrasound examination at 19 weeks of pregnancy and confirmed postnatally, being probably caused by a de novo spontaneous mutation or a disorder of embryogenesis of a non-genetic nature.

KEYWORDS: Meningoencephalocele, bilobed nasal pyramid, bivascular umbilical cord, ultrasound examination, prenatal diagnosis.

INTRODUCTION
Frontonasal dysplasia is a very rare complex of genetic condition, with unknown prevalence, that affect both sexes equally.[1-4]

The spectrum of frontonasal dysplasia mainly includes: median facial cleft syndrome, located at chromosome 1p13.3 (MIM 136760), cleft lip/palate with frontonasal dysostosis and postaxial polysyndactyly (MIM 201180), acrofrontofacionasal dysostosis with genitourinary anomalies (MIM 201181), frontofacionasal dysplasia (MIM 229400), and oculoauriculofrontonasal dysplasia (MIM 601452).[5-9]

MATERIALS AND METHODS
A 23-year-old, caucasian woman, without pathological personal antecedents, pregnant for the first time, reports to the Maternal-Fetal Medicine Department of a private medical center from Bucharest, Romania, in November 2020, for a routine ultrasonographic screening of the fetal malformations.

After the patient approval for the ultrasound examination, fetal sonography was performed transabdominally, with a General Electric Voluson E8 Ultrasound device, by expert ultrasonographer with competence in maternal-fetal medicine.

RESULTS
The ultrasound scanning for fetal structural abnormalities highlight a mono-fetal pregnancy, 19.0 weeks old, in evolution.

This is the first pregnancy of a young couple, clinically healthy, without hereditary or familial pathologies (Fig. 1).

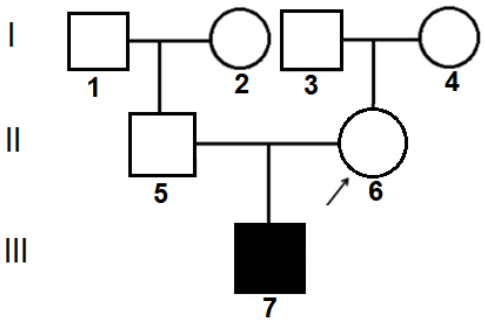

Figure 1: Genealogic tree of the studied family.

At the level of the cephalic extremity, a detailed 2D, 3D and 4D ultrasound screening of the fetus, showed two significant malformations: occipital meningoencephalocele and bilobed nasal pyramid (Fig. 2 - 5).

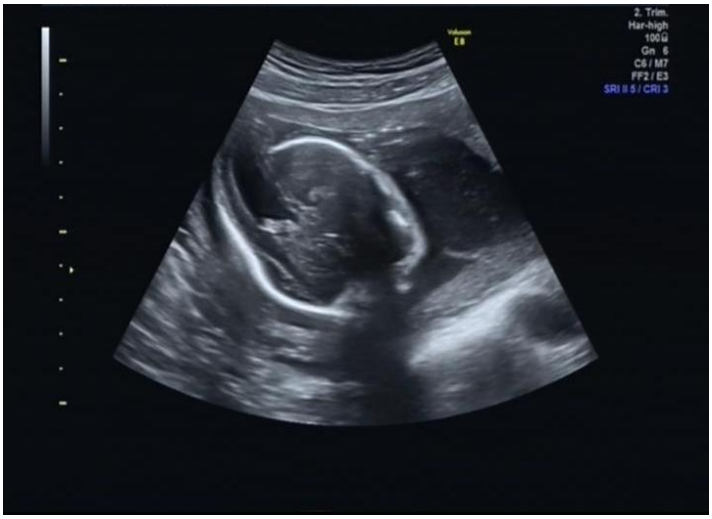

Figure 2: 2D Static ultrasound examination: Fetal head morphology at 19th week of pregnancy: Occipital meningoencephalocele.

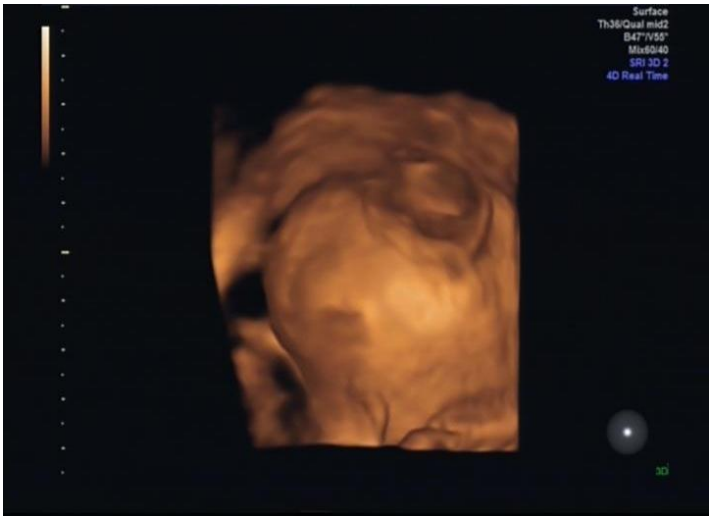

Figure 3: 4D Real Time ultrasound examination: Fetal head morphology at 19th week of pregnancy: Occipital meningoencephalocele.

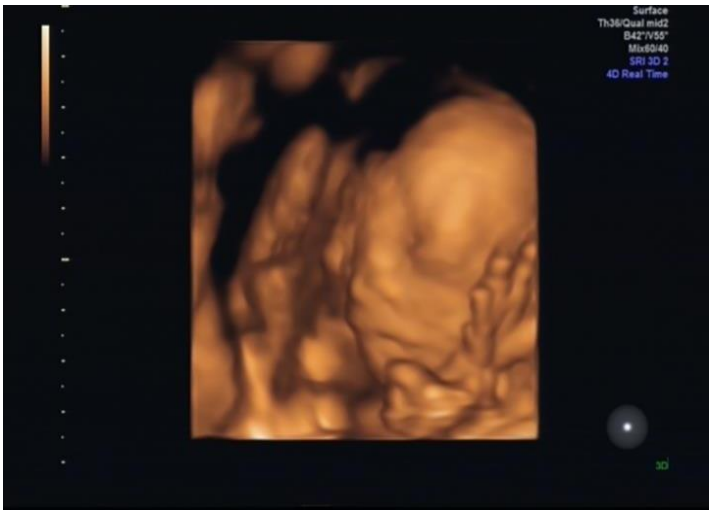

Figure 4: 4D Real Time ultrasound examination: Fetal craniofacial morphology at 19th week of pregnancy: Bilobed nasal pyramid.

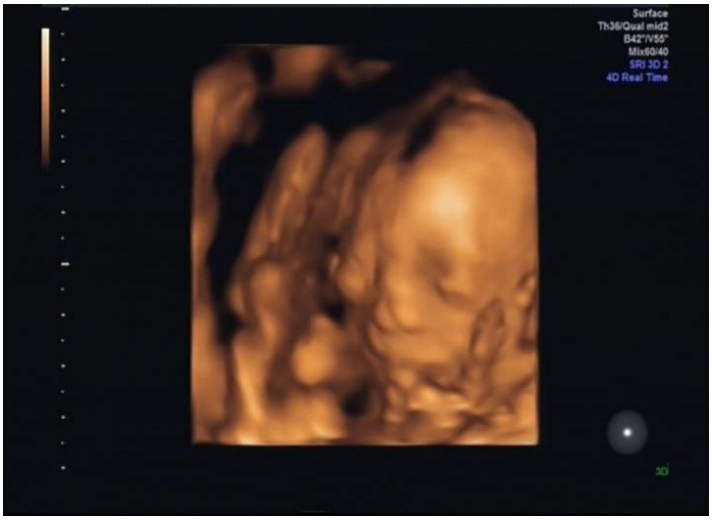

Figure 5: 4D Real Time ultrasound examination: Fetal craniofacial morphology at 19th week of pregnancy: Bilobed nasal pyramid, detailed image.

The ultrasound examination of the thorax pointed, thorax normally shaped and regular aspect heart, with fetal heart rate: 155 beats per minute with normal rhythm. The spine has a normal configuration, without visible abnormalities over 0.5 cm. Also, ultrasound examination of the fetus showed abdomen with normal conformation, as well as liver, spleen and kidneys with regular structure and sizes.

The ultrasound scanning continued with the examination of the extremities, amniotic liquid, umbilical cord, and placenta which highlight: tri-segmental upper and lower limbs, normally shaped, amniotic liquid and placenta with regular aspect, and bivascular umbilical cord, without other congenital anomalies visible in the ultrasound examination (Fig. 6).

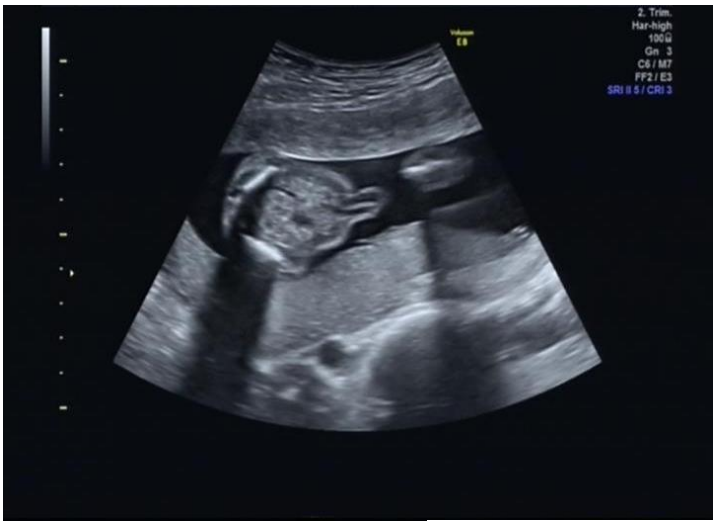

Figure 6: 2D ultrasound examination: Bivascular umbilical cord.

In consonance with the fetal morphology ultrasound, the following diagnosis was established: Mono-fetal pregnancy 19.0 weeks, in evolution. Occipital meningoencephalocele Bilobed nasal pyramid. 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

Following the corroboration of all the prenatal fetal investigations data was established the diagnosis of occipital meningoencephalocele, bilobed nasal pyramid, and bivascular umbilical cord, a rare phenotypic variety of frontonasal dysplasia, isolated, sporadic, non-hereditary case, prenatally ultrasound diagnosed, at 19 weeks of pregnancy and postnatally confirmed, probably determined by a de novo spontaneous mutation or a disorder of embryogenesis of a non-genetic nature.

Meningoencephalocele is a very rare congenital malformation, characterized by the herniation of cerebral tissue, with unknown etiology, usually diagnosed at birth.[10-14] Frequently is associated with other congenital abnormalities, such as corpus callosal malformations, hydrocephalus, and cerebral dysgenesis.[15-19]

Nasal duplication, bifid nose or bilobed nasal pyramid is an extremely rare congenital deformity with many subtype, representing a mild degree of frontonasal dysplasia, which results from aberrant embryological development.[20-12]

CONCLUSION

In conclusion, the presented case is an atypical case, an extremely rare phenotypic variety of frontonasal dysplasia, which associates occipital meningoencephalocele with bilobed nasal pyramid, and bivascular umbilical cord, a new spontaneous phenotypic association, which was successfully diagnosed prenatally by ultrasound examination at 19 weeks of pregnancy and confirmed postnatally, being probably caused by a de novo spontaneous mutation or a disorder of embryogenesis of a non-genetic nature.

Authors' contributions

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

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