WORLD JOURNAL OF PHARMACEUTICAL RESEARCH
SJIF Impact Factor 8.084
Volume 10, Issue 6, 120-126. Research Article ISSN 2277– 7105

PRENATAL DIAGNOSIS AND FOLLOW-UP OF ISOLATED AGENESIS OF THE CORPUS CALLOSUM

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Article Received on
07 April 2021,

Revised on 28 April 2021,
Accepted on 19 May 2021

DOI: 10.20959/wjpr20216-20581

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ABSTRACT

Agenesis of the corpus callosum is a severe congenital abnormality of brain development characterized by the complete or partial absence of the corpus callosum. We are presenting here a phenotypic variety of isolated agenesis of the corpus callosum and bilateral ventriculomegaly, successfully diagnosed prenatally by ultrasound examination at 22 weeks of pregnancy, caused by a disorder of embryogenesis of a non-genetic nature, highlight the crucial role of prenatal sonography in the early detection and specialized management of severe congenital malformations.

KEYWORDS: Agenesis of the corpus callosum, ventriculomegaly, ultrasound examination, prenatal diagnosis.

INTRODUCTION

Agenesis of the corpus callosum (ACC), a rare birth defect, is an abnormality of brain development characterized by the complete or partial absence of the corpus callosum.[1, 2]

According to World Health Organization, the most common type of major congenital malformations are heart defects, neural tube defects, and Down syndrome.[3-5]

Agenesis of the corpus callosum is reported to have an incidence of about 1:4000 live births.[6]

The present study aims to present the brain malformations associated with ACC and to illustrate the benefits of ultrasound examination in the early prenatal diagnosis of severe congenital malformations.

MATERIALS AND METHODS

A 32-year-old, pregnant caucasian woman, was referred at 22 weeks of gestation in a private medical center in Bucharest, Romania, for a routine mid-trimester fetal ultrasound examination.

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

The ultrasound scan was performed with a General Electric Voluson E10 Ultrasound machine, by a maternal-fetal medicine sonographer.

RESULTS

The ultrasound examination highlights a singleton fetus 22.4 weeks old, in evolution, with an estimated fetal weight of 628 +/- 92g (Fig. 1).

EFW (Hadlock)	*Value*	*Range*	*Age*	*Range*	*GP (Hadlock)*
AC/BPD/FL/HC	**628g**	**± 92g**	**23w4d**		**91.9%**

Figure 1: Estimated fetal weight (EFW) from the measurements of abdominal circumference (AC), biparietal diameter (BPD), femur length (FL) and head circumference (HC).

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

At the level of the cephalic extremity: the biparietal diameter was 56.3 mm, the occipitofrontal diameter was 71.2 mm and head circumference was 200.7 mm (Fig. 2).

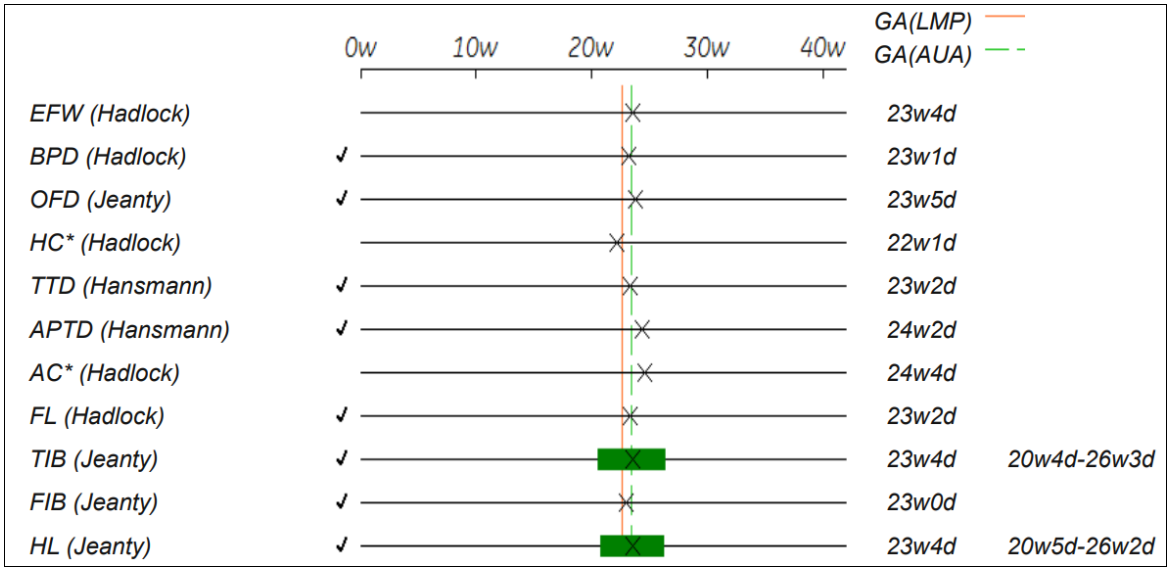

Figure 2: Fetal biometry: occipital frontal diameter (OFD), transverse trunk diameter (TTD), anteriorposterior trunk diameter (APTD), abdominal circumference (AC), femur length (FL), tibia length (TIB), fibula length (FIB) and humerus length (HL).

Fetal brain ultrasonography showed colpocephaly (posterior horn 9.87 mm), "dangling" of choroid plexus, absent septum pellucidum, and partial agenesis of the corpus callosum. (Fig. 3-5).

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

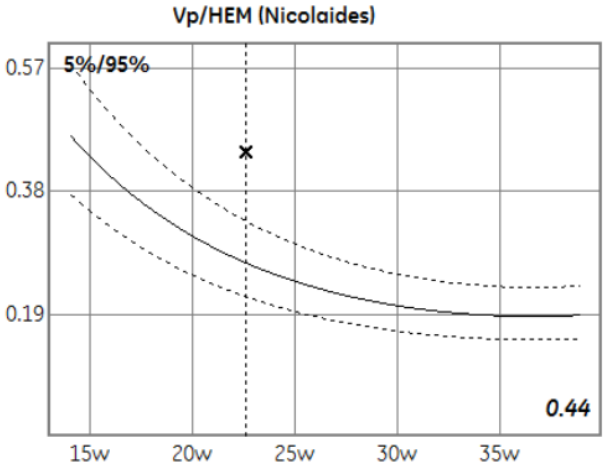

Figure 1: The ratio between the posterior horn of the lateral ventricle (Vp) and hemisphere (HEM): Colpocephaly.

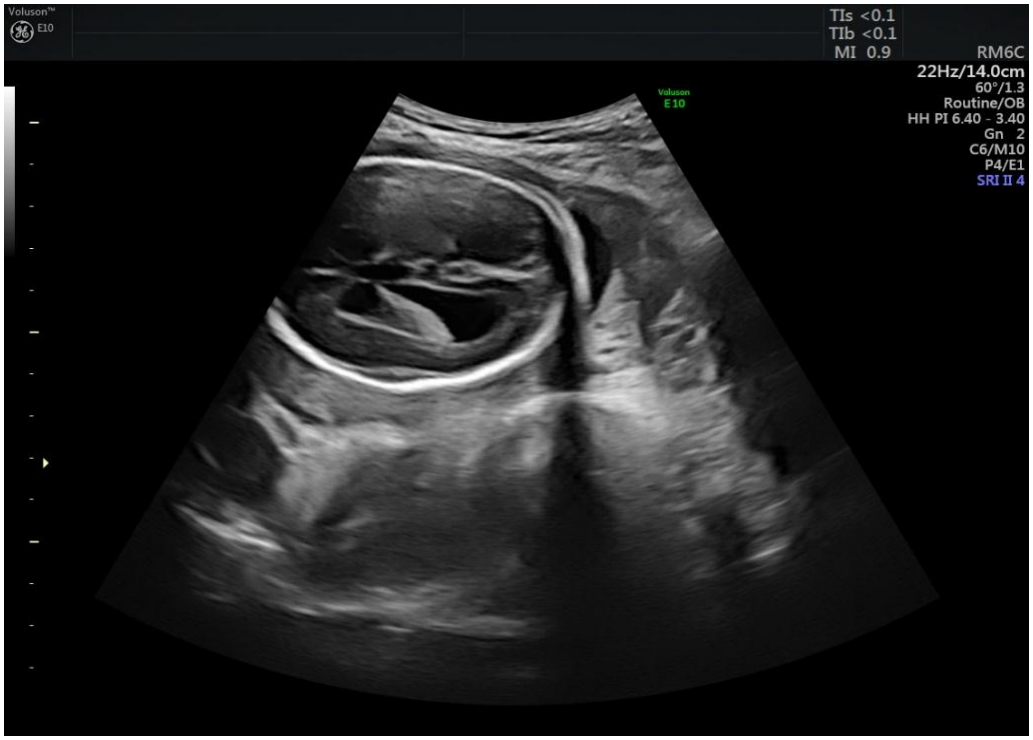

Figure 2: 2D Ultrasound examination: Agenesis of corpus callosum. Tear drop sign.

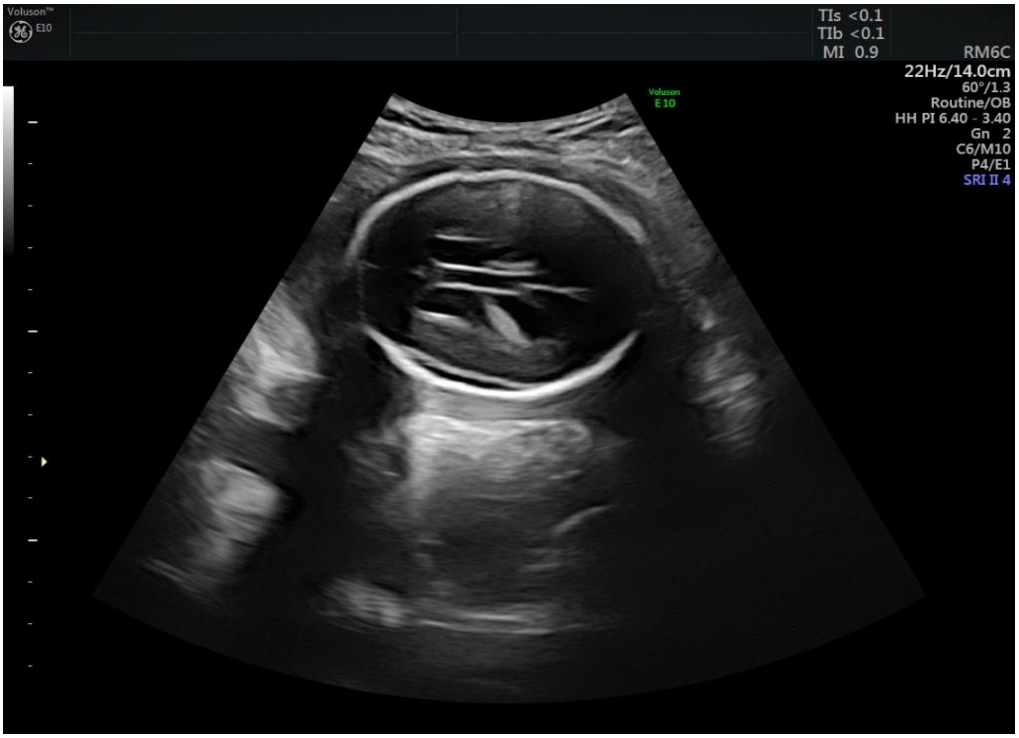

Figure 3: 2D Ultrasound examination: Agenesis of corpus callosum.

In conformity with the fetal morphology scan the following diagnosis was established: Mono-fetal pregnancy 22.4 weeks' of gestation, in evolution with malformed fetus: bilateral ventriculomegaly and agenesis of the corpus callosum.

The parents were informed about the severity of fetal malformations and decide to end the pregnancy for fetal health reasons. Autopsy findings confirmed the ultrasound diagnosis.

DISCUSSION

Agenesis of the corpus callosum is hereditary in isolation or as part of a syndrome, but is often assumed to be sporadic and sometimes associated with chromosomal anomalies.[7, 9] It occurs about 9th–16th weeks of gestation and can be detected prenatally by sonography.[7, 10,11]

The present case report represents an isolated, sporadic, non-hereditary case of agenesis of the corpus callosum, successfully detected prenatally by ultrasound examination at 22 weeks of gestation, being probably caused by a de novo mutation or a disorder of embryogenesis of a non-genetic nature.

Genetic counseling is very difficult because of the bad prognosis but the early identification of the disorder is crucial for the prevention of newborns with serious congenital anomalies.[5,12-15]

CONCLUSION

The phenotypic variety of serious congenital anomalies, which associates agenesis of the corpus callosum, and bilateral ventriculomegaly, highlight the crucial role of prenatal sonography in the early detection and specialized management of severe congenital malformations.

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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