

A Dandy-Walker Variant Prenatally Diagnosed Using Ultrasound on One of the Fetuses of a Twin Pregnancy Obtained through In Vitro Fertilization

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Abstract

We present the case of a patient aged 42 years, who was sterile of undiagnosed nature and who resorted to IVF, as a last resort to get pregnant. Following this procedure, a twin pregnancy resulted.

Ultrasound investigation, of the "second opinion" type performed using a Voluson E8 ultrasound 4D module highlighted: twin pregnancy 34.1 weeks old in evolution; A female fetus without visible malformations on the ultrasound, estimated weight of 2,300 g; B female fetus with borderline ventriculomegalia and vermis hypoplasia with Dandy-Walker cyst appearance, plus a rare malformation association, hypotelorism. Estimated fetal weight is 2,500 g.

It is the first case described in the specialized literature of a version of Dandy-Walker syndrome prenatally diagnosed on one of the fetuses of a twin pregnancy, resulting from in vitro fertilization. The probable etiology of the malformation is genetic, hence the need for careful supervision of all high-risk pregnancies and the need of genetic counseling and consultation, with the final aim of limiting the appearance of affected newborns.

Keywords: Dandy-Walker Syndrome, Ultrasound, Twin Pregnancy, IVF, Vermis, Prenatal Diagnosis

Introduction

The Dandy-Walker syndrome is a brain malformation that occurs in the 4th week of the pregnancy [1]. This involves the total or partial absence of a portion of the cerebellum [vermis], which is associated with the important dilatation of the fourth ventricle and hydrocephalus [2].

It is a rare condition, with a frequency of 1: 25,000 to 30,000 newborns [3], with a higher incidence in females [4].

The disease may have multiple causes: genetical [5, 6], chromosomal [7], toxic or infectious. Dandy-Walker syndrome may associate other brain malformations [8], in 70% of the cases, for example agenesis of corpus callosum and malformations of other organs, in 20-30%

of cases, such as heart, kidney, facial, digital or vertebral defects [9].

The diagnosis of the Dandy-Walker syndrome may be established since intrauterine life, especially during fetal morphology examination in the second trimester of pregnancy, when there may be total or partial agenesis of the cerebellar vermis, as an isolated malformation or associated with other malformations [10].

The vermis agenesis can be viewed using ultrasound as a cystic image [11] that connects the third and fourth ventricles [12].

The Dandy - Walker Syndrome may have three versions: 1. The classical Dandy-Walker malformation (enlargement of the posterior fossa, cerebellar vermis agenesis, tall/ high tentorium, hydrocephalus);

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Case Report

2. Dandy-Walker variant [variable hypoplasia of cerebellar vermis, with or without enlargement of the posterior fossa), and
3. Megacisterna magna (enlarged cisterna magna, keeping the integrity of the cerebellar vermis and of the fourth ventricle).

The aim of this paper is to present the first case described in the specialized literature of the Dandy-Walker syndrome Version diagnosed in one of the fetuses of a dizygotic twin pregnancy, pregnancy resulting from in vitro fertilization.

Case Report

The patient E.L., aged 42 years, comes to the clinic in her 34 week pregnancy for an ultrasound specialist investigation of the "second opinion" type. From previous medical history we establish that the patient is known to have infertility, of an unspecified cause, she was subject to prolonged treatment but without getting any results. As a result, the couple decided to use IVF, as

the last opportunity to achieve a pregnancy. We have to mention that it is a Caucasian couple and non consanguine.

Following in vitro fertilization, the current twin pregnancy was obtained (**Figure 1**).

The ultrasound examination performed using a Voluson E8 ultrasound, 4D module shows the following:

The A fetus (**Figure 2**) shows lower skull, head biparietal diameter: 84.4 mm, occipito-frontal head diameter: 108.5 mm and head circumference: 304.4 mm. The cerebellum has normal structure and configuration: 44.5 mm. Cerebral hemispheres are symmetrical and have a normal compliance. Cisterna magna and Cavum septum pellucidum are also normal in size. The Sulcus lateralis cerebri is visible and the ventricular system shows normal size. The choroid plexus are homogeneous. Blood flow in the middle cerebral artery shows the following values: PSV: 63 cm/s, EDV: 7.83 cm/s, S/D: 8.05, PI: 2.23, RI: 0.88 (normal range).



Figure 1 - Twins pregnancy, 19 weeks



Figure 2 - Twin A, normal

The neck has a normal configuration, as well as the spine, without visible DTN over 0.5 cm. The thorax has normal form and structure, anterior-posterior diameter: 87 mm and transverse diameter: 82.5 mm. The heart is tetra cameral, rhythmic activity, BCF: 129 beats/min. The large vessels at the base of the heart show an apparently normal laying out.

The abdomen is normally configured with anterior-posterior diameter: 99 mm, transverse diameter: 89.2 mm and abdominal circumference: 295.7 mm. Fundic pelvis, Female. Upper and lower limbs are tri-segmental, with normal configuration with humerus: 59.3 mm and femur: 68 mm.

The umbilical cord presented is tri-vascular. Umbilical artery blood flow has the following values: PSV: 69.85 cm/s, EDV: 26.56 cm/s, S/D: 2.63, PI: 0.91, RI: 0.62 (normal range). The amniotic fluid volume is in normal quantity and presents circulation.

The placenta is located in the right corner, presents homogeneous structure, degree of maturation II and thickness: 39 mm at the insertion of the umbilical cord.

Case Report

Fetus B (**Figure 3**) presents the lower skull, brain biparietal diameter: 90.8 mm, occipito-frontal brain diameter: 113.8 mm and head circumference: 321 mm. Cerebellum, 48 mm, with abnormal structure and configuration. Vermis hypoplasia: 23 mm (Dandy-Walker cyst aspect) (**Figure 4**). Cerebral hemispheres are symmetrical and have normal configuration. TCED: 33 mm. Cavum septum pellucidum and Sulcus lateralis cerebri - visible. The ventricular system presents posterior corner: 12.9 mm (0.39 ICp), homogeneous choroid plexus. Blood flow in the middle cerebral artery: PSV: 64.36 cm/s, EDV: 8.85 cm/s, S/D 7.27, PI: 2.3, RI: 0.86 (normal range).



Figure 3 - Twin B, affected



Figure 4 - Dandy-Walker cyst aspect, Vermis hypoplasia

Anterior fossa: external interorbital distance: 52.5 mm (hypotelorism), internal: 21 mm. The neck has a normal configuration, as well as the spine without visible DTN over 0.5 cm. The thorax presents normal shape and structure, with anterior-posterior diameter: 76.4 mm and transverse diameter: 85 mm. The heart is tetra cameral, rhythmic activity, BCF: 143 beats/min.

The large vessels at the base of the heart show apparently normal configuration. Aperture visible. The abdomen has a normal configuration with anterior-posterior diameter: 92.4 mm, transverse diameter: 100.6 mm and abdominal circumference: 303.3 mm. Fundic pelvis background female.

Upper and lower limbs are tri-segmental, of normal configuration, with humerus: 58.4 mm and femur: 65.6 mm.

The umbilical cord is tri-vascular. Umbilical artery blood flow presents the following values: PSV: 55.08 cm/s, EDV: 17.75 cm/s, S/D: 3.1, PI: 1.07, RI: 0.68 (normal range).

The velocimetry of uterine arteries records the following data: the right uterine artery blood flow: L/R: 2, PI: 0.76, RI: 0.50 (normal range), left uterine artery blood flow: S/D: 1.96, PI: 0.74, RI: 0.49 (normal range).

Amniotic fluid volume is in normal quantity and presents circulation. The placenta located on the bottom and rear wall to the left, presents homogeneous structure, maturation degree I-II and thickness: 33 mm at the insertion of the umbilical cord.

On the basis of ultrasound final diagnosis includes twin pregnancy of 34.1 weeks in evolution. 2nd fetus diagnosed with borderline ventriculomegalia, hypoplastic vermis (Dandy-Walker cyst aspect), hypotelorism. Estimated weight: A fetus: 2,300 g; B fetus: 2,500 g.

Discussion

Since the ultrasound examination performed at 34 weeks of pregnancy indicates: ventricular system with posterior corner: 12.9 mm (0.39 ICp) and hypoplastic vermis: 23 mm (Dandy-Walker cyst aspect), the result is that the B fetus presents the Dandy Walker version/ variant (cerebellar vermis variable hypoplasia, with or without enlargement of the posterior fossa) and not the classical Dandy-Walker malformation [13] (enlargement of the posterior fossa, cerebellar vermis agenesis, high tentorium, hydrocephalus).

Regarding cause of the malformation, given the maternal physiological personal history (age over 40 years) and the maternal pathological personal history (sterility) its genetic etiology can be incriminated for two reasons: on

Case Report

the one hand, the sterility can be the the genetical cause, and on the other hand, older age of the mother may favor the occurrence of non-disjunction of chromosome and therefore numerical chromosomal abnormalities in fetuses [14].

In terms of elucidating the etiology, we must mention that no cytogenetic tests were performed either on the couple before IVF or to the two fetuses in the necessary prenatal diagnosis, considering the risk pregnancy [15].

The chorionic villus biopsy performed 10 to 12 weeks of pregnancy or at least, the amniocentesis performed later, in pregnancy weeks 14-16 would have been indicated [7]. By performing fetal karyotypes it would have been able to determine if the fetus B shows a chromosomal abnormality [9,17].

The Prognosis

The prognosis [9,18] in cases of Dandy-Walker syndrome is not good especially if the existence of a chromosomal abnormality is suspected.

The peculiarities of the case

1. Twin pregnancy, obtained after in vitro fertilization, two female fetuses, one normal (fetus A) and the other affected (the fetus B).
2. Fetus B, malformed, presents the Dandy-Walker syndrome Variant and not the classical Dandy-Walker malformation.
3. The Dandy-Walker variant, presented by B fetus is associated with hypotelorism, a very rare association.
4. Prenatal diagnosis was established later, at 34 weeks of pregnancy.

Conclusions

From the above-mentioned information, the following conclusions are drawn:

1. The necessity of accurate, complete and multidisciplinary investigation of all couples before performing the in vitro fertilization procedure.
2. The need for careful supervision of all high-risk pregnancies, in specialized centers.
3. The need for prenatal genetic consultation and genetic counseling aimed ultimately at limiting the incidence of malformed newborns.

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Case Report

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