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Case Report

Moderate associated fetal ventriculomegaly: prenatal diagnosis

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ABSTRACT

Ventriculomegaly (VM) is a descriptive term, indicating the enlargement of the ventricles of the brain. We present the case of a 32-year-old primiparous women, at 18 weeks of pregnancy, who was referred in our clinic for a routine prenatal ultrasound examination. The ultrasound scan highlighted a single malformed fetus with several major abnormalities of the fetal head: hypoplastic nasal bone, agenesis of corpus callosum, choroid plexus hyperplasia, solid hypoechoic mass on the external wall of the left ventricle, no visible cavum septum pellucidum, third ventricle visible, moderately enlarged and VM with evolution to bilateral hydrocephalus. The parents were informed about the major severity of anomalies and decide to terminate the pregnancy. Prenatal ultrasound examination was decisive in the early prenatal diagnosis and optimized management of the malformed fetus with VM.

Keywords: Fetal ventriculomegaly, Ultrasound investigation, Prenatal diagnosis, Agenesis of corpus callosum

INTRODUCTION

Ventriculomegaly (VM) is a descriptive term, indicating the enlargement of the ventricles of the brain caused by the presence of an excess of cerebrospinal fluid, in the lateral ventricles of the developing brain.[1] VM is a severe fetal brain anomaly identified with a prevalence of 0.3 to 1.5 per 1000 live births, more frequent in males, with a male-to-female sex ratio of 1.7.[2-4]

VM recognizes multifactorial causes that result in neurologic, motor, and/or cognitive impairment.[5,6] Common causes of VM can include cerebral and non-cerebral structural abnormalities, congenital infection (Toxoplasma, cytomegalovirus and rubella) and chromosomal anomalies (mainly trisomies 21, 18 or 13), but sometimes the cause may not be evident.[7,8] In severe cases of fetal VM, the overall risk of neurological abnormality is over 75%, even when excluding fetal infection and chromosomal abnormalities.[9]

Fetal VM can be isolated or associated to other anomalies.[10,11] In the presence of associated abnormalities, the prognosis is poor with a high incidence of childhood morbidity.[12-14] The severity of VM can be further classified as: mild/borderline fetal VM, moderate fetal VM and severe fetal VM (fetal hydrocephalus).[4,7,15]

The aim of the case presentation was to illustrate the supremacy of prenatal investigations in the prevention of the severe congenital defects.

CASE REPORT

We present the case report of a 32-year-old primiparous women, at 18 weeks of pregnancy, who was referred in a

private clinic from Bucharest, Romania, in march 2019, for a routine prenatal ultrasound examination.

The ultrasound examination was performed with a GE Voluson E10 ultrasound machine.

After the pregnant woman was informed about the ultrasound investigation, with her informed prenatal consent for sonogram, the ultrasound scan was performed transabdominally by an experienced sonographer.

Table 1: Obstetrics report.

2D genetic	Value	m1	m2	m3	m4	m5	m6
Dist.							
D	1.80 cm	3.09	2.80	0.20	0.36	0.52	3.85
Distance ratio							
D1	3.10 cm	3.10					
D2	2.09 cm	2.19					
D1/D2	1.42	1.42					

The ultrasound scanning highlighted a single fetus 18.3 weeks old (chronological) and 19.2 weeks old (biometric), in evolution, with estimated fetal weight 2,580 g±38g.

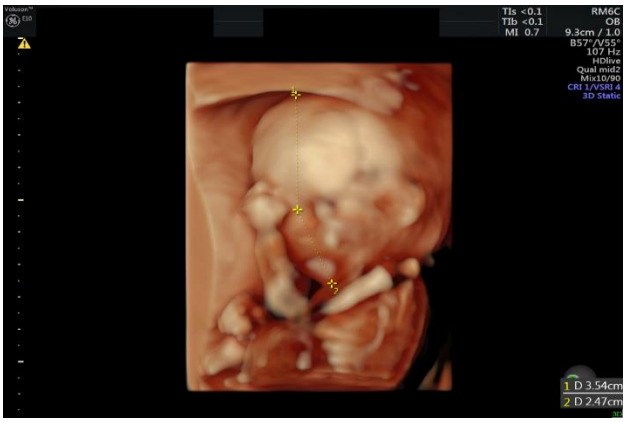

Figure 1: Ultrasound image of the fetal head, 3D static: neurocranium height (D1) and viscerocranium height (D2).

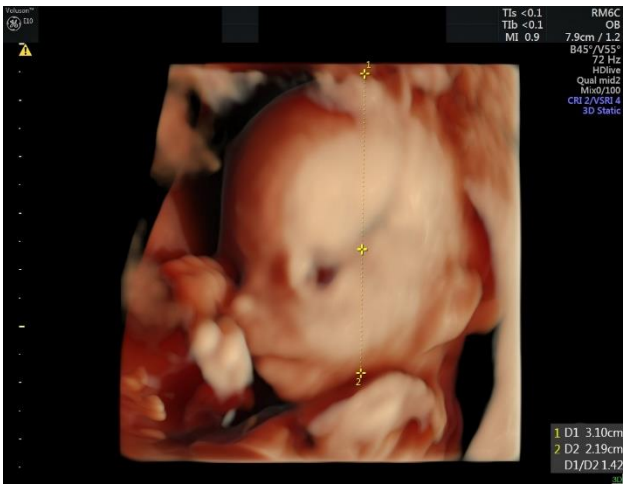

Figure 2: Ultrasound image of the fetal head, 3D static: neurocranium height (D1), viscerocranium height (D2) and distance ratio D1/D2.

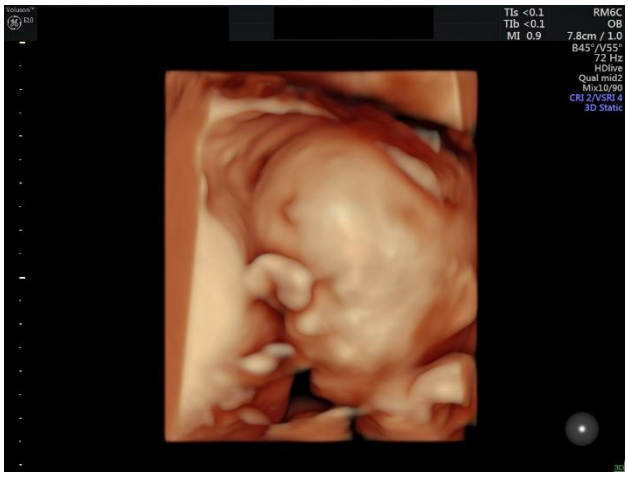

Figure 3: Ultrasound image of the fetal head, 3D static: hypoplastic nasal bone.

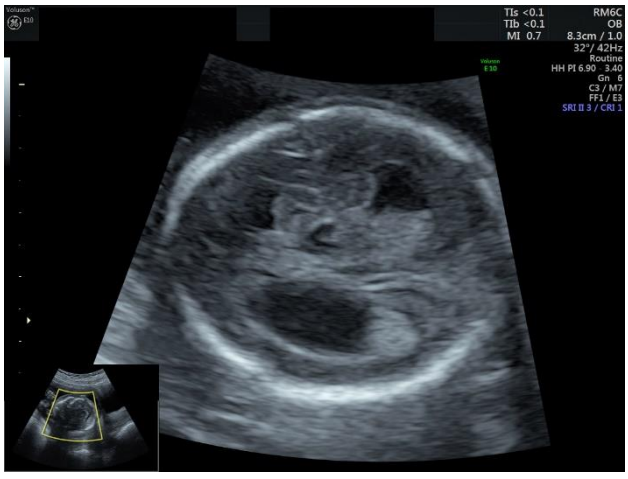

Figure 4: Ultrasound image of the fetal head: bilateral fetal ventriculomegaly, inequality of the cerebral hemispheres, no visible cavum septum pellucidum, agenesis of corpus callosum.

A detailed second-trimester ultrasound evaluation of the fetus revealed several major abnormalities of the fetal

head: neurocranium height (D1): 31 mm, viscerocranium height (D2):21 mm, (Figure 1, 2), D1/D2 ratio: 1.41 (Figure 2) and (Table 1); hypoplastic nasal bone:3.5 mm (Figure 3); posterior horn of lateral ventricle 10.4 mm (ICp 0.58); anterior horn of lateral ventricle: 9.16 mm (ICa 0.51), solid hypoechoic mass on the external wall of the left ventricle: 14.8/5.6 mm; no visible cavum septum pellucidum; agenesis of corpus callosum (Figure 4); third ventricle visible and moderately enlarged: ventriculomegaly with evolution to bilateral hydrocephalus; choroid plexus hyperplasia with bilobed aspect (Figure 5).

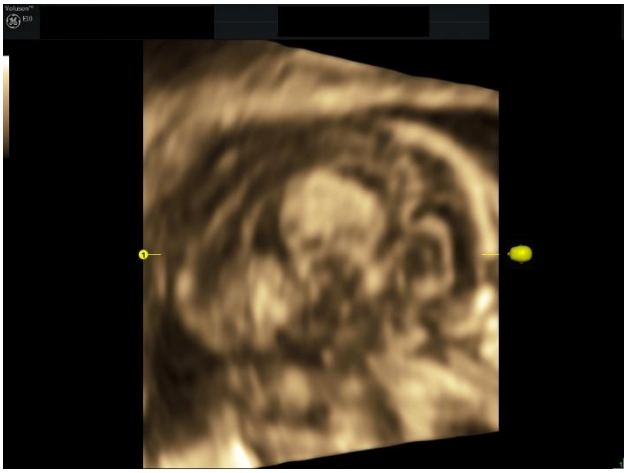

Figure 5: Ultrasound image of the fetal head: moderate fetal ventriculomegaly, agenesis of corpus callosum, choroid plexus hyperplasia.

The parents were informed about the major severity of anomalies and decide to terminate the pregnancy. Anatomopathological examination confirmed the prenatal diagnosis.

DISCUSSION

VM is a prenatal malformation characterized by the enlargement of the lateral ventricles, associated with increases in total brain volume.[13] The rate of association with other cerebral and extracerebral anomalies ranges from 45% to 80%.[16] The association between additional abnormalities and worse outcome is commonly known. On the other hand, the prevalence of delay in the development remains higher, even if cases of associated anomalies are excluded.[17]

Genetic counselling after the prenatal diagnosis of fetal ventriculomegaly is challenging, because four out of five of fetuses with prenatal diagnosis of isolated VM survive, and three out of five of these, present delayed neurodevelopment.[18,19]

In our case report, the prenatal ultrasound diagnosis of VM was crucial, because it was possible to avoid the appearance of a newborn severely affected congenitally, through an accurate early diagnosis.

The limitations of the study were given by the fact that the couple, for the moment, did not accept to perform genetic tests.

CONCLUSION

Prenatal ultrasound examination of the fetal brain is important in the evaluation of the fetal neurodevelopment. In our case, prenatal ultrasound examination was decisive in the early prenatal diagnosis and optimized management of the malformed fetus with VM.

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