

The importance of ultrasonographic markers in early diagnosis of serious congenital malformations incompatible with postnatal life: a case report



Dinu-Florin Albu , Cristina-Crenguta Albu , Stefan-Dimitrie Albu
University of Medicine and Pharmacy Carol Davila, Alco San Clinic - Fetal Medicine Dept.
Bucharest, Romania

A 25-year-old female comes for a ultrasound examination in the second trimester of pregnancy. It is the first pregnancy of a Caucasian non consanguineous couple.

For investigation, Voluson E8 3D/4D life ultrasound device is used.

Results: The ultrasound investigation reveals, ovoid-oblong shaped skull, with biparietal cranial diameter: 41.7 mm, occipito-frontal cranial diameter: 57 mm and head circumference: 155.8 mm, positive "lemon sign", recurved cerebellum, positive "banana sign", lateral ventricle with 14 mm lower horn (Fig. 1 - 5), nuchal translucency of 4.7 mm, dysmorphic spine (Fig. 6 – 10), in thoracolumbar area T7, L4, L5 dysraphism of vertebral arches with meningocele, positive "saucer sign".



The following diagnosis set: Malformed fetus 18.6 weeks of pregnancy, Holoprosencephaly, Thoracolumbar Spina Bifida. The prognosis of the condition is outlined and specialized genetic investigations are recommended, but the parents decide interruption of pregnancy.



Conclusion: Note the significance of ultrasonography investigation in early diagnosis of serious congenital malformations.