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Smith-Magenis syndrome after ultrasonographic detection of semilobar holoprosencephaly at 12 weeks

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Objective

To show our experience on the ultrasonographic diagnosis of malformations of the central nervous system (CNS) at 12 weeks from a clinical case.

Methods

Literature review based on a clinical case.

Results

27 year-old primigravid with no family history, no gynecological-obstetric history of interest. MF: 4 / 28-30. No surgical interventions. No allergies. BMI 35. Smoking 15-20 cigarettes / day. The patient attended at 12 weeks for an ultrasound, so as to perform first trimester combined test. LMP: 12/13/2016 - EDD: 09/19/2017. At 9 weeks the analytical results were B-HCG 0.759 MoMc and PAPP-A 0.731 MoMc. During ultrasound sesion, a CRL value of 57.8 mm was detected, resulting in pregnancy according to the patient's LMP (12 + 2 weeks), a BPD of 25.6 mm (according to 13 + 5 weeks) and a Nuchal Translucency of 2.2 mm (P 90-95). In the morphological examination, presence of 4 limbs, cardiac activity and positive fetal movements, difficult assessment of the central nervous system (CNS) with possible holoprosencephaly and was therefore referred to Fetal Medicine. At the fetal Medicine review, a morphological ultrasound was performed with 12-week CRL (61 mm). Ductus venosus with absence of a wave, the heart can not be evaluated in detail due to limitation of BMI (poor transmission). In the CNS assessment, image compatible with semilobar holoprosencephaly and flat profile with hypoplastic / absent nasal bone. Posterior total occlusive placenta. Voluntary termination of pregnancy (VTP) meaning and terms were explained and a genetic study was recommended. The patient decided VTP and genetic study of fetal sample resulting diploid QF-PCR for Chromosomes 13, 18, 21 and XY sex chromosomes. The result of CGH-arrays concluded that there was a 3.4 Mb deletion in 17p11.2 (RAI1 gene) that coincides with a recurrent microdeletion described as the cause of Smith-Magenis syndrome that may include distinctive physical features and multiple congenital malformations (brachycephaly, hypoplastic nasal bone, prognathism, psychomotor and growth retardation among others). The genetic study in the mother was normal.

Conclusion

Malformations of the CNS are among the most common types of major congenital anomalies. Ultrasound examination is an effective modality for prenatal diagnosis of these anomalies. Holoprosencephaly affects 1 in 8,000 live births and is the most common structural anomaly of the developing forebrain, resulting in facial dysmorphism and neurologic impairment. Given the increasing relative contribution of genetic diseases to perinatal morbidity and mortality, proper recognition of holoprosencephaly could help make an early genetic diagnosis in order to improve the management.