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Fetal intracranial calcifications, not just congenital TORCH infection

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Objective

To report a case of Aicardi-Goutières syndrome (AGS) diagnosed in our centre to illustrate another cause of fetal intracranial calcifications, apart from TORCH infections.

Methods

Case report and review of the literature.

Results

Our patient was a Spanish 37-year-old pregnant woman, gravida 4, para 1. Both members of the couple had no exceptional medical history. First-trimester screening and second-trimester anomaly were normal. At the routine third-trimester sonographic examination (34 weeks of pregnancy), mild bilateral ventriculomegaly was detected and the patient was referred to our centre for a prenatal ultrasonographic consultation. The sonographic findings were moderate bilateral ventriculomegaly with an hyperechogenic periventricular halo and periventricular calcifications, enlarged cisterna magna and cerebral white matter hyperechogenic lesions like calcifications. The cerebellum was hypoplastic and it was displaced due to the enlargement of the cisterna magna. The estimated fetal weight was 2200 grams (6^{th} centile) with normal Doppler measurements. Maternal serologic tests were performed again, including CMV, and they were all negative. Moreover, we performed an amniocentesis with the following results: normal fetal karyotype (46, XX), and the polymerase chain reaction for CMV, Listeria, Toxoplasma, Parvovirus, hepatitis, herpes, Zika and Chikungunya were all negative. Fetal magnetic resonance imaging was performed and it confirmed the ultrasound findings. Additionally, we obtained amniotic fluid for array-CGH testing and a sequence variation of homozygous RNASEH2B gene (c.476G>T) was found. This pathogenic finding is associated with Aicardi-Goutières syndrome. At 37 weeks of gestation a 2420 grams (5^{th} centile) girl was born by vaginal delivery. At the present moment, the infant is 2 months old and is on medical follow-up by pediatrics and rehabilitation. She needs chronic oxygen therapy daily and is taking antiepileptic medication. Aicardi-Goutières syndrome (AGS) is a rare, genetic, early-onset and progressive encephalopathy with an autosomal recessive inheritance pattern. Nowadays, there are 7 genes whose mutations are related to this condition: ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1 and IFIH1. To the best of our knowledge, there are only 6 cases published with prenatal diagnosis of this syndrome. Prenatal imaging findings suggests a CMV congenital infection and the diagnosis in utero requires genetic testing like in our case.

Conclusion

AGS is a rare genetic and progressive encephalopathy. Prenatal imaging findings of this syndrome mimics a congenital CMV infection. The prenatal diagnosis of this condition is hard to achieve because of the genetic heterogeneity.