Case Report

A 20-week pregnant was referred for a routine scan which revealed a female fetus with complete agenesis of the corpus callosum and two interhemispheric cysts. An amniocentesis revealed a normal karyotype and normal SNP array. No other obvious structural defects were detected and in particular the eyes appeared normal. Follow-up scans revealed asymmetrical sulcation, delayed cortical development and unilateral ventriculomegaly, which were confirmed on MRI. These findings raised suspicion on the possibility of Aicardi syndrome. This was confirmed postnatally, due to typical findings on fundoscopy, seizures and confirmation of the prenatal brain findings.

Conclusion

Most cases with isolated agenesis of the corpus callosum have a favorable prognosis. However, even when the findings seem isolated and extensive genetic workout is normal, Aicardi syndrome cannot be ruled out, as its genetic basis is still unknown.