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Rhomboencephalosynapsis - prenatal diagnosis and outcome

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Objective

The purpose of this study was to define the ultrasound features in a group of fetuses with suspected rhomboencephalosynapsis (RES) and to evaluate the outcome of these pregnancies.

Methods

This is a retrospective study of a cohort of fetuses diagnosed with suspected RES between 2013 and 2018. The ultrasound and MRI examinations were reviewed. The results were compared with postnatal clinical evaluation, imaging and pathology examinations.

Results

Seven fetuses with suspected RES were included in the study. Three fetuses were diagnosed with isolated RES. Four were diagnosed with RES and associated malformations, including ventriculomegaly, hydrocephalus and agenesis of the corpus callosum. The average gestational age of diagnosis was 21.0 (range 15-33.4 weeks). MRI was performed in 2 fetuses and amniocentesis was done in one. Six patients had a termination of pregnancy. One postmortem was performed with findings of RES and multiple CNS malformations, abnormal head structure and micrognathia. There was one livebirth in a fetus with suspected RES, dysgenesis of the corpus callosum and ventriculomegaly. This child is now 66 months old with mild motor and posture problems and normal intelligence.

Conclusion

Rhomboencephalosynapsis is a rare subgroup of congenital cerebellar malformations with a spectrum of severity. Information on long-term outcome is limited. In our study, the vast majority of patients chose to terminate the pregnancy and only one patient delivered with relatively good results.