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Posterior fossa malformations: diagnosis and neurodevelopmental outcome

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Objective

Posterior fossa malformations are commonly observed during prenatal US screening. These malformations encompass a heterogeneous spectrum of conditions characterized by progressive abnormal development of the sub-tentorial area and may result in a significant neurological deficit. This study aims to characterize non-cystic posterior fossa malformations using pre-natal MRI and identify the correlation with the post-natal diagnosis, the short-term APGAR score and the long-term neurodevelopmental outcome.

Methods

This historical prospective cohort study included 56 fetuses diagnosed with non-cystic posterior fossa malformations at Sheba Medical Center. The control group was based on 70 fetuses who underwent embryonic MRI, and no findings were found in their examination. We used patients' medical records and VABS-II questionnaire to assess the neurodevelopmental outcomes of these malformations.

Results

In 23/34 cases of living children, we achieved a long-term follow-up, by evaluating children's development in a range of ages from 1 year to 10 years. All the required information was gathered for all cases. Among the study group, 19 had normal neurodevelopmental outcomes, 4 had abnormal neurodevelopmental outcomes, there were 4 cases of neonatal death, 18 pregnancies were terminated, and 11 were not cooperative with the study.

Conclusion

From all cases with non-cystic posterior fossa malformations detected by fetal MRI, 82.6% had normal neurodevelopmental outcomes, and 17.4% had abnormal neurodevelopment. All patients with abnormal outcomes had additional anomalies, and some had chromosomal aberrations. Therefore, when the malformation is isolated, the risk of neurodevelopmental impairment is low.