

## Neurodevelopmental outcomes of children with prenatal diagnosis of isolated vermian hypoplasia

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**Objective.** Magnetic resonance (MR) helps in the definition of malformations when posterior fossa anomalies are suspected. What is still under debate is how to counsel parents about less severe conditions. The aim of our study was to evaluate neurodevelopmental (ND) outcomes of children with isolated vermian hypoplasia.

**Materials and Methods.** Prospective study of children born with a confirmed diagnosis of vermian hypoplasia, prenatally evaluated at our centre from 2009 to 2021. All fetuses underwent MR, amniocentesis was offered to investigate molecular and standard karyotype. Neurological follow-up was performed using age-appropriate developmental tests (GRIFFITHS II, WPPSI-III, WISC-IV) or by submitting a questionnaire to cases living far from our centre.

**Results.** 31 pregnancies were included, of which 9 (29%) were terminated on parental request: MR autopsy and pathologists confirmed the findings.

22 cases were considered for ND evaluation: fetal MRI in 21 (95%), and amniocentesis in 13 (59%) were performed. Six cases were lost at postnatal follow-up; 5 had syndromic features (Noonan, Turner, Joubert, X-linked disorder, one still under genetic investigation); one was post haemorrhage. 10 cases were defined isolated: the ND outcome at scholar age was normal in 4 children (40%), mildly impaired in 4 (40%) and moderately impaired in 2 (20%), one diagnosis of autism spectrum disorder and one moderate learning disability.

**Conclusions.** Although tending towards a favourable prognosis, there is a fair variability of neurological outcomes when cerebellar vermian hypoplasia is isolated, which should be considered in parental counselling. Long-term follow-up is necessary to detect scholar age motor and language outcomes.