

Abstract

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Cerebral folate deficiency with developmental delay, autism, and response to folinic acid.

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BACKGROUND: The authors describe a 6-year-old girl with developmental delay, psychomotor regression, seizures, mental retardation, and autistic features associated with low CSF levels of 5-methyltetrahydrofolate, the biologically active form of folates in CSF and blood.

CONCLUSION: Folate and B12 levels were normal in peripheral tissues, suggesting cerebral folate deficiency. Treatment with folinic acid corrected CSF abnormalities and improved motor skills.

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