Abstract

Neuropediatrics. 2007 Dec;38(6):276-81.

Folate receptor autoimmunity and cerebral folate deficiency in low-functioning autism with neurological deficits.

Ramaekers VT, Blau N, Sequeira JM, Nassogne MC, Quadros EV.

Division of Child Neurology, University Hospital Liège, Belgium.

BACKGROUND: Reduced folate transport to the CNS was identified in two autism spectrum disorders, i.e., Rett syndrome and infantile low-functioning autism with neurological abnormalities.

METHODS: Twenty-five patients with early-onset low-functioning autism with or without neurological deficits, were evaluated for serum folate, cerebrospinal fluid (CSF) 5-methyltetrahydrofolate (5MTHF), and serum FR autoantibodies of the blocking type to determine the significance of folate receptor (FR) autoantibodies with respect to folate transport across the blood-CSF barrier.

RESULTS: In spite of normal serum folate, CSF 5MTHF was low in 23 of 25 patients. The reduced CSF folate in 19 of these 23 patients could be explained by serum FR autoantibodies blocking the folate binding site of the membrane-attached FR on the choroid epithelial cells. Oral folinic acid supplements led to normal CSF 5MTHF and partial or complete clinical recovery after 12 months.

CONCLUSION: Serum FR autoimmunity appears to represent an important factor in the pathogenesis of reduced folate transport to the nervous system among children with early-onset low-functioning autism associated with or without neurological deficits. Early detection of FR autoantibodies may be a key factor in the prevention and therapeutic intervention among this subgroup of patients with autism.

PMID: 18461502