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Mutated methylenetetrahydrofolate reductase as a risk factor for spina bifida

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Abstract

Periconceptional folate supplementation reduces the risk of neural-tube defects. We studied the frequency of the 677C→T mutation in the 5,10-methylenetetrahydrofolate reductase (MTHFR) gene in 55 patients with spina bifida and parents of such patients (70 mothers, 60 fathers), 5% of 207 controls were homozygous for the 677C→T mutation compared with 16% of mothers, 10% of fathers, and 13% of patients. The mutation was associated with decreased MTHFR activity, low plasma folate, and high plasma homocysteine and red-cell folate concentrations. The 677C→T mutation should be regarded as a genetic risk factor for spina bifida.