

Epidemiology/Genetics Abstracts

Title: IDENTIFIED GAPS IN THE LITERATURE: THE ASSOCIATION BETWEEN THE HYPERHOMOCYSTEINEMIA (MTHFR) MUTATION AND ANENCEPHALY

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Background: Studies report that the decline in spina bifida was temporally associated with folic acid fortification of U.S. grain supplies, but the association between fortification and the prevalence of anencephaly remains unclear and the genetic role of the male partner has not been investigated. Early prenatal diagnosis and subsequent pregnancy termination affect the low birth weight prevalence of neural tube defects, but many U.S. birth defects surveillance systems track malformations only among live births or beyond a specific gestational age, thus excluding elective terminations following a neural tube defect in early pregnancies and rendering the effectiveness of folic acid consumption for NTD prevention more difficult to establish.

Method: A systematic literature review was conducted using the 1970-2008 MEDLINE database with relevant search terms (hyperhomocystinemia, NTD, spina bifida, folic acid, etc.) and limited to humans and English language. Of the 378 publications, 110 were abstracted and systematically coded.

Results: The folic acid supplementation literature yielded two commonly cited recommendations: a) individuals with elevated plasma homocystine / MTHFR mutation should consume 100-400mcg folic acid daily to decrease cardiovascular disease and other adverse medical conditions; and b) all women of childbearing age should consume 400mcg of folic acid daily to reduce their risk of having an infant affected by a neural tube defect. Few studies investigated possible associations between neural tube defects and MTHFR. The existing studies have sampling and methodology limitations including: a) MTHFR testing of children with spina bifida; b) MTHFR testing of only mothers and neural tube defect offspring; c) use of national surveillance databases that exclude early pregnancy termination data; and d) no data from male partners.

Conclusion: The authors recommend studying the known adult MTHFR population to determine their pregnancy outcome history both to capture the early pregnancy diagnosis and terminations and to investigate the male genetic role.