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Association of neural tube defects in children of mothers with MTHFR 677TT genotype and abnormal carbohydrate metabolism risk: a case-control study

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ABSTRACT. Abnormalities in maternal folate and carbohydrate

metabolism have both been shown to induce neural tube defects (NTD) in humans and animal models. However, the relationship between these two factors in the development of NTDs remains unclear. Data from mothers of children with spina bifida seen at the Unidad de Espina Bífida del Hospital Infantil Virgen del Rocío (case group) were compared to mothers of healthy children with no NTD (control group) who were randomly selected from patients seen at the outpatient ward in the same hospital. There were 25 individuals in the case group and 41 in the control group. Analysis of genotypes for the methylenetetrahydrofolate reductase (MTHFR) 677CT polymorphism in women with or without risk factors for abnormal carbohydrate metabolism revealed that mothers who were homozygous for the MTHFR 677TT polymorphism and at risk of abnormal carbohydrate metabolism were more likely to have offspring with spina bifida and high levels of homocysteine, compared to the control group. The increased incidence of NTDs in mothers homozygous for the MTHFR 677TT polymorphism and at risk of abnormal carbohydrate metabolism stresses the need for careful metabolic screening in pregnant women, and, if necessary, determination of the MTHFR 677CT genotype in those mothers at risk of developing abnormal carbohydrate metabolism.

Key words: Spina bifida; Neural tube defects; Folic acid; Diabetes; MTHFR

