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FOLATE - RELATED GENETIC POLYMORPHISMS AND RISK OF SPINA BIFIDA

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Background: Neural tube defects (NTD) are one of the most common and severe congenital defects. The defect usually leads to death or life-long handicap in surviving children. Periconceptional supplementation of folic acid is known to reduce the frequency of NTD by 50-70% (folate-dependent defects). Polymorphisms of genes encoding folate pathways-related enzymes and proteins (5,10-methylenetetrahydrofolate reductase – MTHFR, methionine synthase reductase – MSR, and transcobalamin II – TCII) have been demonstrated to be risk factors for having a child with NTD. Objective: The aim of the study was to determine the total frequency of known folate-related genetic polymorphisms – risk factors for having a child with NTD in mothers of children with such defect. Methods: The studied population included 110 mothers of children with nonsyndromic spina bifida (the most common NTD type in Poland). PCR-RFLP method was used to detect the occurrence of following risk factor – polymorphic genotypes: 677TT, 1298CC and 677T+1298C (MTHFR gene), 66GG (MSR gene), 776GG (TCII gene). Results: At least 1 of the above-mentioned genotypes was found in 55 (50%) of the mothers. In 18 participants 2 risk factors and in 1 mother 3 risk factors were found. Conclusion: The known folate-related genetic risk factors for having a child with NTD can be find in around 50% of mothers from affected families. Testing for the five genetic polymorphisms listed above should be routinely used in genetic counseling of NTD – families as the polymorphisms probably occur in the majority of mothers having a child with folate-related NTD