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Neural Tube Defects and 19 bp Deletion Within Intron-1 of Dihydrofolate Reductase Gene

Nejat AKAR, Ece AKAR, Yonca EĞİN, Gülhis DEDA, Saadet ARSAN, Mesiha EKİM
Department of Pediatrics, Faculty of Medicine, Ankara University, Ankara - TURKEY

Abstract: Aims: Dihydrofolate reductase (DHFR) is necessary for the reduction of the ingested folates before they are used in the body metabolism. Thus, the DHFR enzyme has an important role in folate supplementation and the DHFR gene is a strong candidate for a teratogenic locus for neural tube defects (NTDs). There is a 19 bp deletion within the first intron of the DHFR gene, which may have an effect on folate reduction. We thus studied this mutation in Turkish spina bifida patients and their mothers to determine whether there is an association with the occurrence of NTD. Materials and Methods: The case-control study included 69 meningomyelocele (MMC) patients and 104 mothers who gave birth to NTD babies. One hundred sixty eight women, consecutively selected, who admitted to the laboratory, were included as controls. One hundred thirty-six newborns without MMC were also included. Results: DHFR gene 19 bp deletion in homozygous state was significantly higher in the MMC group compared to control newborns [Odds Ratio 2.4 (CI 95% 0.95-6.08)]. It also brought 2.4- fold risk [Odds Ratio 2.4 (CI 95% 1.04-5.6)] in NTD mothers compared to controls. Conclusions: Our data revealed that DHFR gene 19 bp deletion homozygosity and other possible mutations in the DHFR gene should be studied further especially in folate-supplemented mothers with NTD recurrence.

Key Words: Neural tube defects, spina bifida, dihydrofolate reductase

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