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Analysis of folate pathway gene variants in mothers of Down syndrome children

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Introduction: Folates are essential nutrients required for synthesis of DNA, RNA, amino acid metabolism, formation of SAM, histones and lipids. Folates are reduced and methylated in the liver with the help of enzymes encoded by genes such as MTHFR, MTRR and RFC I. Variants in the genes encoding these enzymes lead to *hypomethylation*, resulting in non-disjunction which in turn increases the risk for Down syndrome. The present study assessed the effect of these gene variants and other factors among mothers of Down syndrome children.

Methodology: A total of 150 mothers having Down syndrome children and 150 mothers having normal children were enrolled in the study. Chromosomal analysis was done to confirm trisomy 21 in children. PCR/RFLP was carried out in mothers for genotyping of MTHFR, MTRR and RFC I.

Findings: Out of the 150 cases, 56 were females and 94 were males with age ranging from two days to 17 years. Mean maternal age was 27.2±5.2 years and 28.8±4.9 years in Down syndrome mothers and control mothers, respectively. Further, analysis showed that intake of folic acid during conception helps in reducing risk, while, intake of drugs for having male child, parity and alcohol intake significantly increases the risk for Down syndrome child. Genotypic frequency of MTHFR 1298 A>C was significantly different among cases and controls ($\chi^2=4.78$, $p=0.02$), indicating presence of C instead of A allele in MTHFR 1298 is associated with risk for the Down syndrome. However, other variants (MTRR and RFC I) did not show any association with Down syndrome.

Conclusion: In the present study, parity, drugs and alcoholism and presence of MTHFR 1298 A>C variants is associated with increased risk for Down syndrome among mothers.

Biography

Amandeep Kaur is pursuing her PhD and has experience in Cytogenetics and Molecular Genetics. She has worked as Project Fellow under a three-year project on Down syndrome. She has recently completed another project as Principal Investigator in a project sanctioned to her by DST, India under women scientist scheme.

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