

## Prevalence of Methylenetetrahydrofolate Reductase A1298C Variant in Tunisian Childhood Acute Lymphoblastic Leukemia

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**Abstract :** Background: Acute lymphoblastic leukemia (ALL); a common blood cancer characterized by the interaction between genetic and environmental factors. Methylenetetrahydrofolate reductase (MTHFR) is an essential folate metabolic enzyme in the processes of DNA synthesis and methylation. A common functional variant of the MTHFR gene, the A1298C, which induces disturbances in folate metabolism, may affect susceptibility to ALL. Objective: The present study aimed to assess the prevalence of MTHFR polymorphism A1298 > C in Tunisian children with ALL. Materials and Methods: A total of 28 Tunisian ALL children were enrolled in this study. Genomic DNA was extracted from whole venous blood collected in ethylenediaminetetraacetic acid (EDTA). Genotyping was carried out with restriction fragment length polymorphism (RFLP) using MboII restriction enzyme. Genotype distribution and allele frequency of MTHFR A1298C was calculated in ALL patients. Results: The A1298C variant of MTHFR was found in 11(19.6%) heterozygous and one homozygous patient (3.5%). Conclusions: This result highlights that A1298C polymorphism of MTHFR is common in Tunisian childhood ALL and suggests that this variant may have a potential role in leukemogenesis. Genotyping of large samples and different ethnicities are required to validate these findings.

**Keywords :** methylenetetrahydrofolate reductase, acute lymphoblastic leukemia, A1298C variant, prevalence

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