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Mutations in MTHFR Gene Associated with Mental Retardation and Cerebral Palsy Combined with Mental Retardation in Erbil City

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Abstract : Folate metabolism plays a crucial role in the normal development of the neonatal central nervous system. It is regulated by MTHFR gene polymorphism. Any factors, which will affect this metabolism either by hereditary or gene mutation will lead to many mental disorders. The purpose of this study was to investigate whether MTHFR gene mutation contributes to the development of mental retardation and CP combined with mental retardation in Erbil city. DNA was isolated from the peripheral blood samples of 40 cases suffering from mental retardation (MR) and CP combined with MR were recruited, sequence the 4, 6, 7, 8 exons of the MTHFR gene were done to identify the variants. Exons were amplified by PCR technique and then sequenced according to Sanger method to show the differences with MTHFR reference sequences. We observed (14) mutations in 4, 6, 7, 8 exons in the MTHFR gene associated with Cerebral Palsy combined with mental retardation included deletion, insertion, Substitution. The current study provides additional evidence that multiple variations in the MTHFR gene are associated with mental retardation and Cerebral Palsy.

Keywords : methylenetetrahydrofolate reductase (MTHFR) gene, SNPs, homocysteine, sequencing
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