Clinical, Biochemical, Molecular and Therapeutic analysis of Maple Syrup Urine Disease in Upper Egypt.

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Abstract:

Maple syrup urine disease (MSUD) is an autosomal recessive inherited metabolic disorder caused by mutations in any of the genes encoding for the branched-chain keto dehydrogenase (BCKDH) components. This study screened MSUD patients throughout the whole Upper Egypt describing their symptoms, clinical and laboratory findings, genetic studies, and their treatment, with a 6-month follow-up for their responses. Screening identified three children with MSUD. Homozygous mutation in R195Q single nucleotide polymorphism (SNP) within the BCKDHA gene was found with the second MSUD patient. Follow-up for 6 months to assess the treatment regimens and progression of cases demonstrated that early treatment regimens including a dietary restriction of branched-chain amino acids with L-Carnitine administration could prevent MSUD-associated intellectual disabilities. It was concluded that R195Q SNP is pathogenic, and it may cause inherited forms of MSUD in some patients. MSUD cases have rarely been reported; so these findings will be highly useful for future cases of MSUD in the Upper Egyptian population.

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