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Discrepancy in the pathogenicity of a previously reported mutation in Maple Syrup Urine Disease

Fateme Golnabi1, Zahra Motezaker1, Tina Shirzade1, Maryam Abiri1,3* , Sirous Zeinali 2,1*

Dr. Zeinali's Medical Genetics Laboratory, Kawsar Human Genetics Research Center, Tehran, Iran
Department of Molecular Medicine, Biotechnology Research Center, Pasteur Institute of Iran, Tehran, Iran
Department of Medical Genetics, School of Medicine, Iran University of Medical Sciences, Tehran, Iran

Email:zeinalipasteur@yahoo.com*
mary_abiri86@yahoo.com*

Introduction:

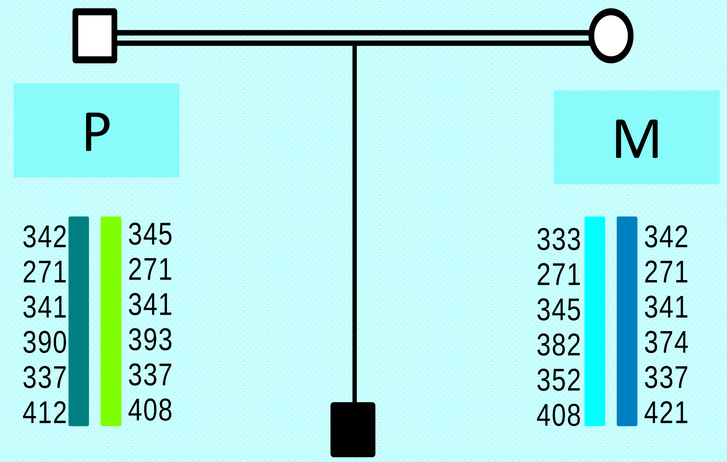
Maple Syrup Urine Disease (MSUD) is a rare autosomal recessive disorder of branched-chain amino acid (BCAA) Metabolism. The disease is mainly caused by mutations either in the BCKDHA, BCKDHB, DBT or DLD genes. Here we describe a variant in DBT gene which is apparently pathogenic, but in reality it couldn't be disease causing.

Materials and methods:

A family suspicious to MSUD was referred us to confirm the clinical diagnosis and mutation detection. In this study, autozygosity mapping was done using STR (short tandem repeat) markers linked to the above-mentioned genes. Then the candidate gene was subsequently sequenced. Heterozygosity of the identified variants was tested in parents.

Results:

Although Homozygosity mapping did not show homozygous haplotype for each of studied genes, direct sequencing of the 4 associated genes were done .Different variations including homozygous variant of c.1150 G>A (p.Gly384Ser) were observed in DBT gene in the affected child. The father was also homozygous for this variant but the mother was heterozygote.



Discussion:

Although Homozygosity mapping did not show homozygous haplotype for each of studied genes, direct sequencing of the 4 associated genes were done .Different variations including homozygous variant of c.1150 G>A (p.Gly384Ser) were observed in DBT gene in the affected child. The father was also homozygous for this variant but the mother was heterozygote.