عنوان مقاله:
A 3-year-old boy with lethargy, leg muscle degeneration and mental retardation

محل انتشار:
سومین کنگره بین المللی و پانزدهمین کنگره ملی ژنتیک ایران (سال: 1397)

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خلاصه مقاله:
Introduction: Maple syrup urine disease (MSUD) is an autosomal recessive disease characterized by disruption of the normal activity of the branched-chain α-ketoacid dehydrogenase (BCKAD) complex. MSUD can be caused by homozygous or compound heterozygous mutation in at least 3 genes: BCKDHA, BCKDHB, and DBT. MSUD presents in the neonate with feeding intolerance, failure to thrive, lethargy and maple syrup odor to urine. Methods: We analyzed a 3-year-old boy who was born of a consanguineous marriage visited at Bandar Abbas medical genetics laboratory (BMGL) of Hormozgan province Iran, suspected with MSUD. The mother's amniotic fluid(AF) sample was taken in 15th week of gestational age and DNA was extracted. Targeted NGS (Next Generation sequencing) was suggested followed by sanger sequencing for mutation confirmation in patient and family members (parents and sister). The fetus genotype was examined by sanger sequencing for the known mutation. Results: NGS analysis showed a c.C653G homozygote mutation in BCKDHB gene for patient. Sanger sequencing investigation indicated heterozygousity of parents for same mutation but his sister's homozygosity for wild type allele. Similarly, sanger sequencing on fetal cells showed that the fetus is also heterozygous for the c.C653G mutation.Conclusions: We have shown a case of MSUD type Ib. Homozygote c.C653G mutation has been reported as the causative defect for the patient. Fetus genotype analysis for the 3rd child of family showed heterozygosity for the mutation. We prevented the recurrence of a rare disease in Hormozgan province by early identification of c.C653G mutation in parents and its examination in their embryo.

کلمات کلیدی:
MSUD, BCKDHB, Mutation, NGS