عنوان مقاله:

Introducing a recurrent mutation in a patient affected with Methylmalonic academia by Whole Exome Sequence

محل انتشار:

کنفرانس بین المللی ژنتیک و ژنومیکس انسانی (سال: 1400)

تعداد صفحات اصل مقاله: 1

نویسندگان:

Ghazaleh Malekizadeh - *Department of Molecular and Cell Biology, Faculty of Basic Science, University of Mazandaran, Babolsar, Iran*

Omid Jazayeri

Morteza Alijanpour - *Non-Communicable Pediatric Disease Research Center, Health Research Institute, Babol University of MedicalScience, Babol, Iran*

Majid Tafrihi - *Department of Molecular and Cell Biology, Faculty of Basic Science, University of Mazandaran, Babolsar, Iran*

خلاصه مقاله:

Background: Methylmalonic Acidemia (MMA) is a rare autosomal recessive metabolicdisorder, result from genetic defect in methylmalonyl-CoA mutase (MCM) enzyme. Thisenzyme is necessary in the catabolism of branched chain amino acids (BCAA) for thedegradation of odd-chain fatty acids, the amino acid valine, isoleucine, methionine, andthreonine, and cholesterol. MMA has wide range of clinical manifestations varying from nosigns or symptoms to severe lethargy and metabolic crisis in newborn infants. This disease iscaused by mutation in five mainly genes (MUT, MMAA, MMAB, MMADHC, MCEE). In thisstudy we reported a recurrent MMA causative mutation in ۲ years old boy.Materials and Methods: We performed whole exome sequencing method (WES), followed bySanger sequence in our patient. In silico analyses of the identified variant was performed usingweb-based bioinformatics programs.Results: WES identified the missense mutation c.A۹۷۶G (p.R۳۲۶G) in the MUT gene whichaffects the stability and enzymatic activity of MCM. The results of the Sanger sequence showedthat our patient is homozygous and his parents are carriers. Bioinformatics software programssuch as Polyphen, SIFT have predicted that this variant will be damaging.Conclusion: This pathogenic mutation has previously been reported in Iran and Ukraine.Considering that our patient is from the northern Iran and this mutation has been already reportedthe same region; Therefore we can conclude that this mutation is recurrent and prevalent in northof Iran. Additionally, our finding would be beneficial for prenatal diagnosis of MMA as well asestablishing a local variant database.

کلمات کلیدی:

Methylmalonic Acidemia, Whole exome sequencing, Iran, Mutation, Metabolic disorder