عنوان مقاله:

Detection of GAN gene Mutation in Leukodystrophy: A Case Report

محل انتشار:

دومین کنگره بین المللی و دهمین همایش ملی نوروژنتیک ایران (سال: 1396)

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خلاصه مقاله:

Introduction: Leukodystrophies comprise a clinically and genetically heterogeneous group of progressivehereditary neurological disorders mainly affecting the myelin in the central nervous system. Here, we present afamily with leukodystrophy followed by molecular genetic evaluation.Methods: A nine years old boy with peripheral neuropathy, polycyctic kidney and Precocious puberty, referredfor genetic counselling. EMG showed axonal neuropathy which has suggested demyelinating type. Parents werefirst cousin with no history of other patient in their first and second degrees' relatives. Genomic DNA wasevaluated through whole exome sequencing followed by bioinformatics analysis of data. Parents and healthychild were examined for the candidate gene variant.Results: One homozygote variant c.851+1G> A on gene GAN has been detected. The frequencies of this variantin normal population are very low. It has been reported as pathogenic and predicted to be damaging by severalonline prediction tools. GAN- related giant axonal neuropathy is inherited in an autosomal recessive manner.Parents and other healthy child were heterozygote for this variant.Conclusion: This case demonstrates that this major developmental error can be diagnosed with new .molecularmethod such as whole exome sequencing along with comprehensive clinical examination

کلمات کلیدی:

leukodystrophy, Neurogenetics, Whole Exome Sequencing