عنوان مقاله:

Genetic Diagnosis of a Lethal Form of Autosomal Recessive Polycystic Kidney Disease

محل انتشار:

مجله بین المللی کودکان, دوره 6, شماره 2 (سال: 1397)

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خلاصه مقاله:

Background Autosomal recessive polycystic kidney disease (ARPKD; OMIM number 263200) is a severe early onset hereditary form of polycystic kidney and liver disease. Case Report In the current study, we present a consanguineous couple with a history of an affected son with polycystic kidney disease (PKD), hepatic failure and epileptic seizures who died at the age of 8 months. Both parents were heterozygote for a missense mutation in PKHD1 gene (NM_170724, c.9107T> G, p.V3036G). Conclusion Unlike previous studies which showed the association between missense mutations of PKHD1 gene and mild phenotype of ARPKD, we have demonstrated the presence of a certain heterozygote missense mutation in parents of a patient affected with lethal form of disorder. Such phenotypic variations should be considered in genetic counseling of families especially those seeking prenatal .diagnosis

کلمات کلیدی:

ARPKD, Gene, Mutation