عنوان مقاله:

Examining the Role of Polymorphisms in Exon 25 of the PKD1 Gene in the Pathogenesis of Autosomal Dominant Polycystic Kidney Disease in Iranian Patients

محل انتشار:

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نویسندگان:

Morteza Bagheri - *Nephrology and Kidney Transplant Research Center, Urmia University of Medical Sciences, Urmia, Iran Cellular and Molecular Research Center, Cellular and Molecular Medicine Institute, Urmia University of Medical .Sciences, Urmia, Iran*

Khadijeh Makhdoomi - *Nephrology and Kidney Transplant Research Center, Urmia University of Medical Sciences, .Urmia, Iran*

Ali Taghizadeh Afshari - *Nephrology and Kidney Transplant Research Center, Urmia University of Medical Sciences, .Urmia, Iran*

Ahmad Ali Nikibakhsh - *Nephrology and Kidney Transplant Research Center, Urmia University of Medical Sciences, .Urmia, Iran*

Isa Abdi Rad - *Nephrology and Kidney Transplant Research Center, Urmia University of Medical Sciences, Urmia, Iran & Cellular and Molecular Research Center, Cellular and Molecular Medicine Institute, Urmia University of Medical .Sciences, Urmia, Iran*

خلاصه مقاله:

Background: Autosomal dominant polycystic kidney disease (ADPKD) is a highly prevalent life-threatening monogenic disorder with high morbidity and mortality. Roughly 1:400-1000 individuals are affected with this disease worldwide. The development of ADPKD is largely attributed to mutations in the polycystic kidney disease (PKD)1 and PKD2 genes. However, the pathogenicity of the different polymorphisms in PDK1 in the development of ADPKD remains unclear. The aim of this study was to further elucidate the role of the polymorphisms in exon 25 of the PDK1 gene in relation to the pathogenesis of ADPKD in Iranian patients. Methods: The genomic DNA of 36 Iranian patients with ADPKD was isolated using the standard salting out method. The PCR products were directly sequenced and analyzed. Results: The frequencies of CAG>GAG, ATG>GTG, GTC>GTA, and GTG>ATG polymorphisms in exon 25 of the PKD1 gene were 34 (94.44%), 33 (91.67%), 26 (72.22%), and 5 (13.89%), respectively. The most frequent polymorphism associated with ADPKD was the homozygous CAG→GAG which causes an amino acid change of Q[Gln] to E[Glu] at codon 3005. Conclusions: Our data suggests that there is potentially a common polymorphism of PDK1 among the Iranian population with ADPKD. This may aid in the diagnosis and genetic screening of at-risk .patients for ADPKD

کلمات کلیدی:

.ADPKD, PKD1 Gene, Polymorphism