Z605 Two mutations of the PKD2 gene in Korean with autosomal dominant polycystic kidney disease

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Autosomal dominant polycystic kidney disease (ADPKD) is one of the most frequently inherited disorders, with an incidence of 1/1000 individuals. ADPKD is a systemic disorder mainly characterized by the progressive formation and enlargement of cys genetically heterogeneous that can be caused by an alteration in at least three different gts in the kidneys, which are associated with decline in renal function. The disease is aenes: PKD1, the major locus at 16p13.3, PKD2 at 4q21-23 and the much rarer PKD3, not yet mapped. Recently the PKD2 gene has been positionally cloned and consist of 15 exons with an open reading frame of 2,904 bp and a 3'untranslation region (UTR) of 2,086 bp. We report a systematic mutation screening of all 15 exons of PKD2 gene, using single strand conformation polymorphism (SSCP) analysis followed by nucleotide sequencing. From the analysis with the PKD2 gene in 28 Korean PKD patients, two mutations were identified. One is an A to G substitution that might be a missense mutation, changing a threonine to an alanine at codon 419 in exon 5 (T419A). The other is a four base pair deletion in exon 6 (1436del4), resulting in a frameshift leading to a premature translation stop and giving rise to a predicted production of truncated protein.

Z606 Ser20Gly Mutation of *Amylin* in Essential Hypertensives

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Essential hypertension is a complex disease with strong genetic influences. Insulin resistance, which is frequently observed in patients with essential hypertension, is a confounding factor in hypertension, diabetes, and odesity, among which there has been suggested common genetic factor. Amylin, which is consist of at least three exons, is a recently discovered peptide hormone composed of 37 amino acids that is cosecreted with insulin by pancreatic beta cells. Amylin has been reported to have important action for glucose metabolism. Thus, molecular abnormalities of amylin may be lead to insulin resistance. Recently, S20G mutation in the *amylin* gene was found in Japanese NIDDM patients. This finding allowed us to examine the role of amylin in the pathogenesis of essential hypertension. To screen for the presence of mutation within the franking region and exons of *amylin* gene, we performed the SSCP analysis on Korean 83 normotensives and 87 hypertensives, respectively. One mutation type with amino acid S20G substitution were observed within exon3 of *amylin* gene. The allele frequencies of the mutation were 2.3 and 1.2% in the hypertensive and normotensive groups, respectively. Our data suggest that *amylin* gene may not play a major role in susceptibility to essential hypertension or insulin resistance.