F111 Two Novel Polymorphisms in the Coding Region of the Vasopressin Type 2 Receptor Gene

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The AVPR2 gene encodes the vasopressin V2 receptor. Disruption of the AVPR2 gene causes X-linked congenital nephrogenic diabetes insipidus(NDI). So far, many different mutations and one molecular heterogeneity (codon 331$^{AGC \rightarrow AGT}$) have been reported. In this study, we performed PCR-directed sequencing of the full length of the AVPR2 gene in five NDI patients and found two novel polymorphisms in coding region of the AVPR2 gene which do not alter the encoded amino acids. That is, the codon 42 $^{GCG \rightarrow GCA}$ and the codon 309 $^{GTA \rightarrow GTG}$ were found in one patient and two unrelated patients, respectively. Polymorphisms in coding region are very rare and can be used as informative markers in disease diagnosis. Our two novel polymorphisms will be used as informative markers for diagnosis of NDI.

F112 Cellular Localization of Three Novel Mutant Vasopressin V2 Receptors In Nephrogenic Diabetes Insipidus

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The AVPR2 gene encodes vasopression type 2 receptor. Disruption of the AVPR2 gene causes X-linked congenital nephrogenic diabetes insipidus(NDI). So far, about 70 different mutations have been reported in this disease. Most of them lead the receptor trapped in cytoplasm. As a consequence, the abnormal receptor can not reach plasma membrane for binding to vasopressin. This study was focused on the cellular localization of three novel mutant vasopressin type 2 receptors (A98P, L274P, P322L) which we previously reported. AVPR2 cDNA was obtained from inner medullary collecting duct cells of a nomal individual by RT-PCR and was cloned into pKF 18k/19k plasmid. Site directed mutations were introduced into them with Mutan-super Express Km. Resulting mutant AVPR2 clones were re-introduced into the pQBI25 expression vector including GFP reporter gene and were expressed in cos-7 cells. Cellular localization of the expressed proteins was traced by confocal laser scanning microscopy. These results provide a clue to fully understand pathogenesis of NDI caused by three novel mutations.