VHL, PBRM1, and SETD2 Genes in Kidney Cancer: A Molecular Investigation

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Abstract : Kidney cancer is the most lethal urological cancer accounting for 3% of adult malignancies. VHL, a tumorsuppressor gene, is best known to be associated with renal cell carcinoma (RCC). The VHL functions as negative regulator of hypoxia inducible factors. Recent sequencing efforts have identified several novel frequent mutations of histone modifying and chromatin remodeling genes in ccRCC (clear cell RCC) including PBRM1 and SETD2. The PBRM1 gene encodes the BAF180 protein, which involved in transcriptional activation and repression of selected genes. SETD2 encodes a histone methyltransferase, which may play a role in suppressing tumor development. In this study, RNAs of 30 paired tumor and normal samples that were grouped according to the types of kidney cancer and clinical characteristics of patients, including gender and average age were examined by RT-PCR, SSCP and sequencing techniques. VHL, PBRM1 and SETD2 expressions were relatively down-regulated. However, statistically no significance was found (Wilcoxon signed rank test, p > 0.05). Interestingly, no mutation was observed on the contrary of previous studies. Understanding the molecular mechanisms involved in the pathogenesis of RCC has aided the development of molecular-targeted drugs for kidney cancer. Further analysis is required to identify the responsible genes rather than VHL, PBRM1 and SETD2 in kidney cancer.

Keywords : kidney cancer, molecular biomarker, expression analysis, mutation screening

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