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ABSTRACT

Expression Profile in von Hippel-Lindau Disease Associated Sporadic Clear Cell Renal Cell Carcinoma

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The renal cell carcinoma (RCC) is the ninth most common type of cancer in world. The most frequent histology is clear-cell RCC, and can be sporadic or associated with hereditary syndromes, like von Hippel-Lindau Disease (VHL), an autosomal dominant syndrome that results from germline mutations in the VHL gene. The aim of this study was to analyze the expression profiles of ccRCCs from VHL patients compared with sporadic. Seven sporadic ccRCC paired with normal kidney tissue, and 21 paired ccRCC from six VHL patients were included. Total RNA was extracted using commercial kits, and microarray experiments were performed using Human Gene 1.0 ST Array (Affymetrix). Partek Suite Software was used for statistical analysis of differential expressed genes between sporadic and VHL tumors, and hierarchical clustering. We found 60 differentially expressed genes which discriminate VHL from sporadic tumors: 45% of them associated with protein binding, 30% related to nucleic acid binding, and 27.5% involved in ion binding. The hierarchical cluster analysis showed that normal samples (VHL + sporadic) grouped together, but in two distinct clusters; and separate of tumor samples (VHL + sporadic). We could define a molecular profile that distinguish normal kidney from VHL patients from normal kidney from patients with sporadic ccRCC. In conclusion, our results highlight new mechanisms of ccRCC carcinogenesis in VHL, and define genes that are exclusively expressed in normal and tumor tissues trigged by germline VHL mutations.