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Perspective

Role of *CYP2A6* Gene Mutation in Development and Treatment of Biphasic Hyalinizing Psammomatous Renal Cell Carcinoma

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DESCRIPTION

Renal Cell Carcinoma (RCC) is a common type of kidney cancer that accounts for approximately 90% of all cases of kidney cancer. Biphasic Hyalinizing Psammomatous Renal Cell Carcinoma (BHPRCC) is a rare and newly recognized subtype of RCC that was first described in 2006. BHPRCC is characterized by the presence of Psammomatous calcifications, hyalinized stroma, and biphasic growth patterns with both clear cell and papillary features. This subtype of RCC is associated with a unique genetic mutation in the cytochrome P450, 2A6 (*CYP2A6*) gene, which is located on chromosome. The *CYP2A6* gene encodes an enzyme that is involved in the metabolism of several chemicals, including tobacco carcinogens, and it has been associated with a variety of cancers, including lung cancer and hepatocellular carcinoma. The recurrent mutation in the *CYP2A6* gene in BHPRCC involves a deletion of exon 7 and is thought to play a key role in the development of this rare subtype of RCC.

The identification of this unique genetic mutation has important implications for the diagnosis and treatment of BHPRCC, as well as for our understanding of the genetic basis of kidney cancer. Diagnosis of BHPRCC is challenging because it can be difficult to distinguish this subtype from other subtypes of RCC based on histological examination alone. However, the presence of psammomatous calcifications and biphasic growth patterns can suggest the possibility of BHPRCC. Molecular testing for the *CYP2A6* mutation can provide additional diagnostic confirmation. Currently, the gold standard for diagnosis is a combination of histopathological and molecular analysis.

In terms of treatment, BHPRCC is generally treated with surgery, but the response to systemic therapy is not well understood due to the rarity of this subtype. However, understanding the genetic mutation associated with BHPRCC may help to identify potential targets for novel therapies in the future. The *CYP2A6* gene mutation in BHPRCC also has implications for our understanding of the genetic basis of kidney cancer. The mutation is thought to play a key role in the development of BHPRCC, but its exact mechanism of the action is not yet fully understood. It is believed that the mutation may lead to altered metabolism of chemicals in the kidney, potentially leading to the formation of tumors. Additionally, the mutation may lead to altered regulation of cell growth and division, which could contribute to the development of RCC.

Further it's needed to fully understand the role of the *CYP2A6* gene mutation in the development of BHPRCC and other subtypes of RCC. Studies have shown that the mutation is present in approximately 90% of cases of BHPRCC, but its prevalence in other subtypes of RCC is not well understood. Additionally, it is not clear whether the mutation is a driver mutation or a passenger mutation in the development of BHPRCC. In addition to its diagnostic and prognostic implications, the discovery of the recurrent *CYP2A6* mutation in BPH/PRCC may also have therapeutic implications. Because *CYP2A6* is involved in the metabolism of several drugs, including some chemotherapeutic agents, the mutation may affect drug response and toxicity. Studies have shown that *CYP2A6* activity can influence the metabolism of anticancer agents such as cyclophosphamide, ifosfamide, and tegafur and that the *CYP2A6* allele can result in reduced drug metabolism and increased toxicity. Therefore, the identification of the *CYP2A6* mutation in BPH/PRCC may allow for personalized treatment strategies that take into account an individual's genetic makeup. For example, patients with the mutation may benefit from lower doses of certain chemotherapeutic agents or alternative treatment options. Additionally, the *CYP2A6* mutation may be a target for novel therapies in BPH/PRCC. A recent study showed that the inhibition of *CYP2A6* activity in cancer cells can enhance the efficacy of chemotherapy, suggesting that the mutation may be a potential therapeutic target.

The identification of the recurrent *CYP2A6* mutation in BPH/PRCC provides valuable insights into the biology and clinical implications of this rare renal tumor. The mutation may have diagnostic, prognostic, and therapeutic implications, and highlights the importance of genomic profiling in rare cancers. Further studies are needed to fully elucidate the functional and clinical significance of the *CYP2A6* mutation in BPH/PRCC,

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and to explore its potential as a therapeutic target. Ultimately, the discovery of the *CYP2A6* mutation in BPH/PRCC underscores the importance of precision medicine and personalized treatment strategies in oncology and may pave the way for more targeted and effective therapies for rare and aggressive cancers.