Von Hippel-Lindau disease case reports

2022

A 35-year-old African-American male with von Hippel Lindau syndrome developed Sinonasal renal cell-like adenocarcinoma (SNRCLA). He underwent surgical resection followed by adjuvant radiation and has no recurrence one year from diagnosis. A review of the literature yielded two similar cases in the setting of VHL. In this case with associated VHL syndrome, next-generation sequencing detected MST1R mutation, a possible driver. Sinonasal renal cell-like adenocarcinoma is an emerging tumor associated with VHL syndrome and it is hoped that future studies shed light on the underlying biology of this unique tumor [1].

2020

Different manifestations detected with 68Ga-DOTATOC PET/CT in patients with Von Hippel-Lindau disease: A case report [2].

A 55-year-old male and a 37-year-old female, who were diagnosed as having VHL syndrome with a positive family history. The male patient presented with upper abdominal discomfort 2 years prior to the current study, and was diagnosed in another hospital as having a space-occupying lesion at the head of the pancreas. After undergoing hemangioblastoma resection of the right cerebellar hemisphere 1 month ago, he was admitted to the hospital for recent aggravation of upper abdominal discomfort to receive further diagnosis and treatment. The female patient previously underwent right ovarian cystectomy and T5-T6 intramedullary hemangioblastoma resection. She was diagnosed at another hospital as having pancreatic cancer, and was admitted to the hospital for recent aggravation of upper abdominal discomfort to receive further diagnosis and treatment.

The diagnosis and treatment of VHL syndrome is currently relatively difficult. It poses a substantial threat to patients and their families. The early and timely diagnosis and treatment of VHL syndrome can improve patients' prognosis and rates of survival [3].

1)
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2)
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3)
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