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**Radiology**

# Radiologic Hallmarks of Von Hippel–Lindau Syndrome: A Case of Multiorgan Involvement

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**Abstract** **Case Report**

Von Hippel–Lindau (VHL) syndrome is a rare autosomal dominant disorder resulting from germline mutations in the VHL tumor suppressor gene on chromosome 3p25.3. It predisposes affected individuals to the development of benign and malignant tumors and cysts in multiple organs, including the central nervous system, kidneys, adrenal glands, pancreas, and inner ear. Early radiologic recognition is essential for timely diagnosis, surveillance, and management.

**Keywords:** Von Hippel–Lindau syndrome, pheochromocytoma, pancreatic cystadenoma, renal cysts, CT imaging, hereditary tumor syndrome.



## INTRODUCTION

The von Hippel-Lindau (VHL) syndrome is a rare autosomal dominant genetic disorder characterized by the development of tumors and cysts in multiple organs, including the central nervous system, retina, kidneys, adrenal glands, pancreas, and inner ear [1]. This condition results from germline mutations in the tumor suppressor gene VHL located on chromosome 3p25.3, leading to a loss of function of the VHL protein (pVHL), a key regulator of the hypoxia pathway [2].

Clinical manifestations of VHL are highly variable and may include retinal and cerebellar hemangioblastomas, clear cell renal carcinomas, pheochromocytomas, endolymphatic sac tumors, and pancreatic cysts [1]. Early detection and regular surveillance are essential to prevent serious complications such as vision loss or neurological deficits and to optimize therapeutic management [3].

## CLINICAL PRESENTATION

A 44 years old male patient was presented to the ER with a hypertensive peak, headache and profuse sweating. The interrogation found that the patient had suffered from abdominal pain, weight loss and episodes of anxiety. A secondary hypertension was suspected, and a thoraco- abdomino-pelvic CT scan was demanded.

### RADIOLOGICAL PRESENTATION

The Ct was able to find a lesion in the right adrenal gland, spontaneously hypodense, showing intense and heterogeneous enhancement after contrast administration, containing cystic (necrotic) areas (figure 1).

The pancreas shows multiple cystic formations throughout its extent, of variable sizes and shapes, spontaneously hypodense, containing thin septations and parietal calcifications in some areas, without visible enhancement after contrast injection. There is no pancreatic ductal dilatation or peripancreatic fat infiltration (figure 1).

Multiple cortical and subcapsular renal cystic formations, with both hyperdense and hypodense contents, thin walls, and no detectable mural nodules or internal septations. These lesions show no enhancement after contrast administration (figure 1).

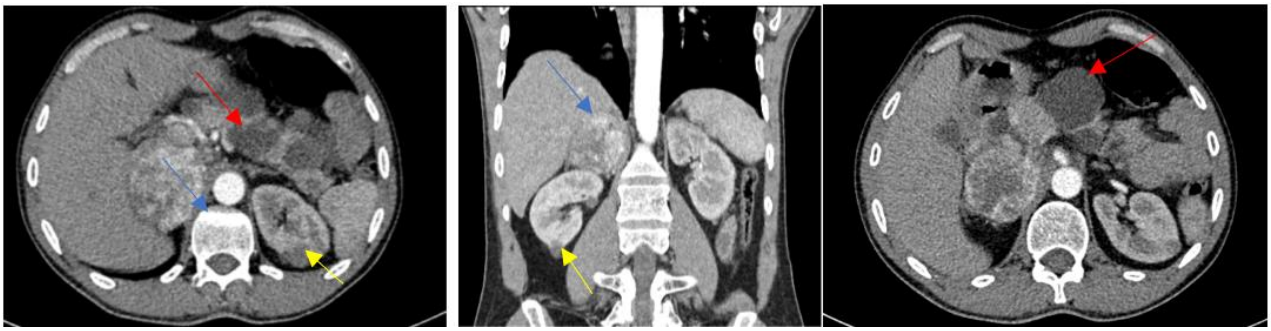

**Figure 1: Axial and coronal images of an abdominal CT scan after enhancement showing a pheochromocytoma (blue arrow), pancreatic cysts (red arrow) and renal cysts (yellow arrow)**

Findings are suggestive of a right adrenal lesion consistent with a locally invasive pheochromocytoma, associated with multiple pancreatic cystic lesions consistent with cystadenomas and bilateral renal cysts and nodules, overall compatible with Von Hippel-Lindau syndrome.

## CONLUSION

From a radiological perspective, Von Hippel-Lindau (VHL) syndrome represents a prototypical example of a multisystemic hereditary disorder where imaging plays a central role in both diagnosis and longitudinal management.

Radiologists must maintain a high index of suspicion when encountering multifocal cystic and solid lesions across abdominal organs, especially in young adults, to suggest the possibility of an underlying genetic syndrome. Accurate lesion characterization, assessment of local invasion, and systematic follow-up are key to preventing complications and guiding surgical or interventional planning.

Ultimately, the radiologist's role extends beyond image interpretation—it is pivotal in early detection, multidisciplinary coordination, and long-term surveillance, thereby improving the overall prognosis of patients with VHL syndrome.

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