The Imaging Gamut of Von-Hippel-Lindau

A 34-year-old man presented with progressive abdominal pain, occasional headaches, and excessive sweating for over a year. A contrast-enhanced computed tomography (CT) of the abdomen revealed heterogeneously enhancing and similar solid-cystic lesions in both adrenal glands and kidneys indicating pheochromocy and renal cell carcinomas, respectively, along with a few simple cortical cysts. Subcentimetric cysts were also seen in the pancreatic head [Figure 1]. Suspecting Von-Hippel-Lindau (VHL) syndrome on the basis of these findings, we performed magnetic resonance imaging (MRI) of the brain and spine. It revealed a cystic T1 hypointense and T2 hyperintense medullary lesion with an enhancing nodule, suggesting a brainstem hemangioblastoma. Intensely enhancing nodules were also seen within both cerebellar hemispheres, suggesting cerebellar hemangioblastomas [Figure 2]. On positron emission tomography imaging, the cerebellar and adrenal lesions were somatostatin receptor-expressing, whereas renal lesions were non-somatostatin receptor-expressing. Genetic testing revealed a heterogenous

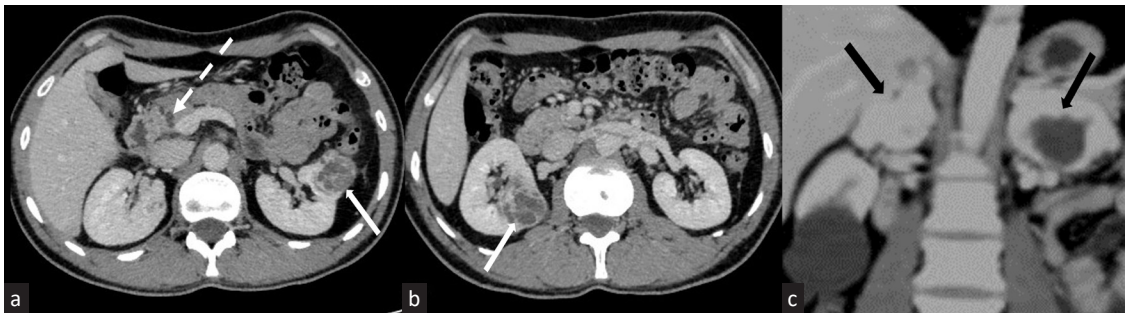

Figure 1: Contrast-enhanced CT images of a 34-year-old man with VHL. (a-b) Bilateral complex renal cystic lesion with heterogeneously enhancing solid internal components: renal cell carcinoma (solid white arrows) with nonenhancing subcentimeter hypodense lesions of cystic attenuation in the pancreatic head; simple pancreatic cysts (dashed white arrow). (c) Heterogeneously enhancing soft tissue density mass with hypodense internal areas involving bilateral adrenal glands: pheochromocytomas (black arrows). CT: Computed tomography, VHL: Von-Hippel-Lindau.

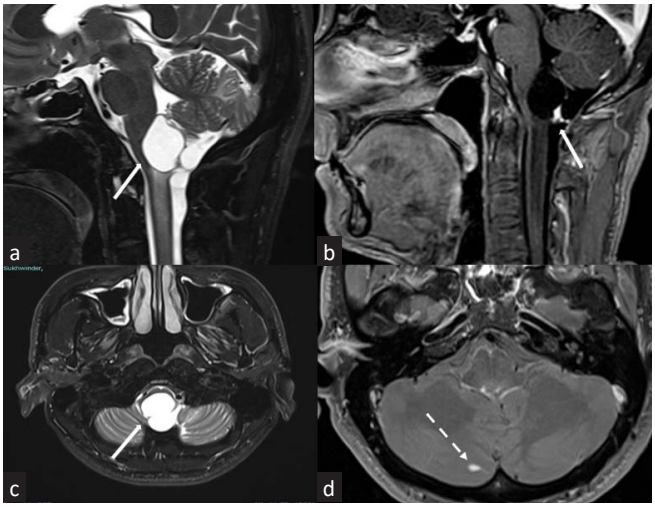

Figure 2: MRI images of brain and cervical spine of a 34-year-old man with VHL. (a-c) Sagittal and axial T2 weighted and postcontrast images reveal a well-defined T2 hyperintense cystic lesion with hyperenhancing mural nodule within the dorsal aspect of the medulla: hemangioblastoma (solid white arrows). (d) Axial postcontrast images show a small intensely enhancing area in the right cerebellar hemisphere: cerebellar hemangioblastoma (dashed white arrow). VHL: Von-Hippel-Lindau, MRI: Magnetic resonance imaging.

nonsense variation in exon 3 of the VHL gene, confirming the diagnosis. He underwent laparoscopic bilateral adrenalectomy and is on follow-up for the renal lesions.

VHL syndrome develops due to a mutation on the short arm of chromosome number 3.[1–3] It causes various tumors involving different organs.[2–4] The syndrome can be diagnosed in patients with a VHL variant gene if they have one VHL manifestation. In patients without the VHL variant gene or affected first-degree relatives, diagnosis can be established if they have at least two manifestations, one being a hemangioblastoma. Other manifestations include renal cell carcinoma, pancreatic neuroendocrine tumor, pheochromocytoma, and endolymphatic sac tumor.[5] Imaging is often the first diagnostic workup performed in most patients. The physician/radiologists need to be cognizant that such a constellation of lesions on CT/MRI points to a diagnosis of VHL.

Conflicts of interest: There are no conflicts of interest.

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References

1. Ganeshan D, Menias CO, Pickhardt PJ, Sandrasegaran K, Lubner MG, Ramalingam P, *et al*. Tumors in von Hippel-Lindau syndrome: From head to toe-comprehensive state-of-the-art review. Radiographics 2018;38:982.
2. Leung RS, Biswas SV, Duncan M, Rankin S. Imaging features of Von Hippel-Lindau disease. Radiographics 2008;28:65–79; quiz 323.
3. Shanbhogue KP, Hoch M, Fatterpaker G, Chandarana H. Von Hippel-Lindau disease: Review of genetics and imaging. Radiol Clin North Am 2016;54:409–22.
4. Mourão JLV, Borella LFM, Duarte JÁ, Dalaqua M, Fernandes DA, Reis F. Imaging manifestations of Von Hippel-Lindau disease: An illustrated guide focusing on the central nervous system. Radiol Bras 2022;55:188–92.
5. Louise M Binderup M, Smerdel M, Borgwadt L, Beck Nielsen SS, Madsen MG, Møller HU, *et al*. von Hippel-Lindau disease: Updated guideline for diagnosis and surveillance. Eur J Med Genet 2022;65:104538.

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