Molecular Oncologic Imaging

https://dergipark.org.tr/tr/pub/moi

Von Hippel Lindau Syndrome with Renal Cell Carcinoma, Pancreas Serous Cystadenoma, Hemangioblastoma and NET Demonstrated on Ga-68 DOTATATE PET-CT and FDG PET-CT Imaging

Pınar Pelin ÖZCAN[1] Zehra Pınar KOÇ[2] Gökçe YAVAN[3] Mustafa Musa DİRLİK[4]

[1]Mersin University, Faculty of Medicine, Department of Nuclear Medicine, Mersin, Turkey, ppelinozcan@gmail.com
[2]Mersin University, Faculty of Medicine, Department of Nuclear Medicine, Mersin, Turkey, zehrapinarkoc@gmail.com
[3]Mersin University, Faculty of Medicine, Department of Nuclear Medicine, Mersin, Turkey, gyavan95@gmail.com
[4]Mersin University, Faculty of Medicine, Department of General Surgery, Mersin, Turkey, mdirlik@mersin.edu.tr

Cite this study: Ozcan, P. P., Koc, Z. P., Yavan, G. & Dirlik, M. M. (2022). Von Hippel Lindau Syndrome with Renal Cell Carcinoma, Pancreas Serous Cystadenoma, Hemangioblastoma and NET Demonstrated on Ga-68 DOTATATE PET-CT and FDG PET-CT Imaging. Molecular Oncologic Imaging, 2(1), 12-16

Keywords
68 Ga PSMA PET-CT
FDG PET-CT
Von Hippel Lindau Syndrome

Research Article
Received: 10.03.2022
Accepted: 14.04.2022
Published: 15.04.2022

ABSTRACT

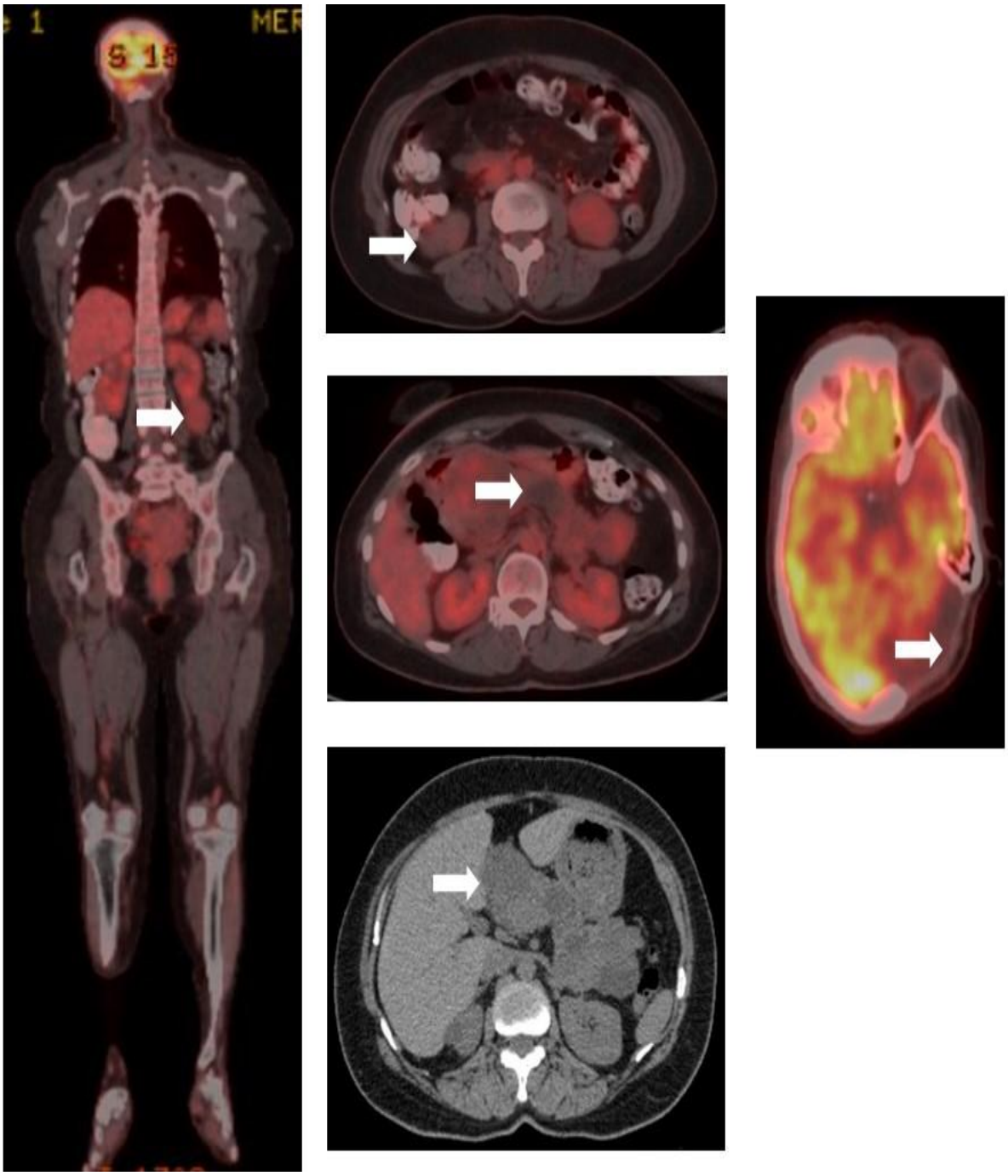
Von Hippel–Lindau disease (VHL), also known as Von Hippel–Lindau syndrome, is a rare genetic disorder with multisystem involvement (1). Imaging plays an important role in the evaluation of cancer patients. In recent years, much attention has been focused on 68Ga-PSMA PET-CT. We hereby, report the rare case of a Von Hippel Lindau Syndrome with renal cell carcinoma, pancreas cystadenoma, hemangioblastoma and pancreatic NET demonstrated on both Ga-68 PSMA and FDG PET-CT imaging.

1. Introduction

Von Hippel–Lindau disease (VHL) is characterized by visceral cysts and benign tumors with potential for subsequent malignant transformation. It is a type of phacomatosis that results from a mutation in the Von Hippel–Lindau tumor suppressor gene on chromosome 3p25.3.

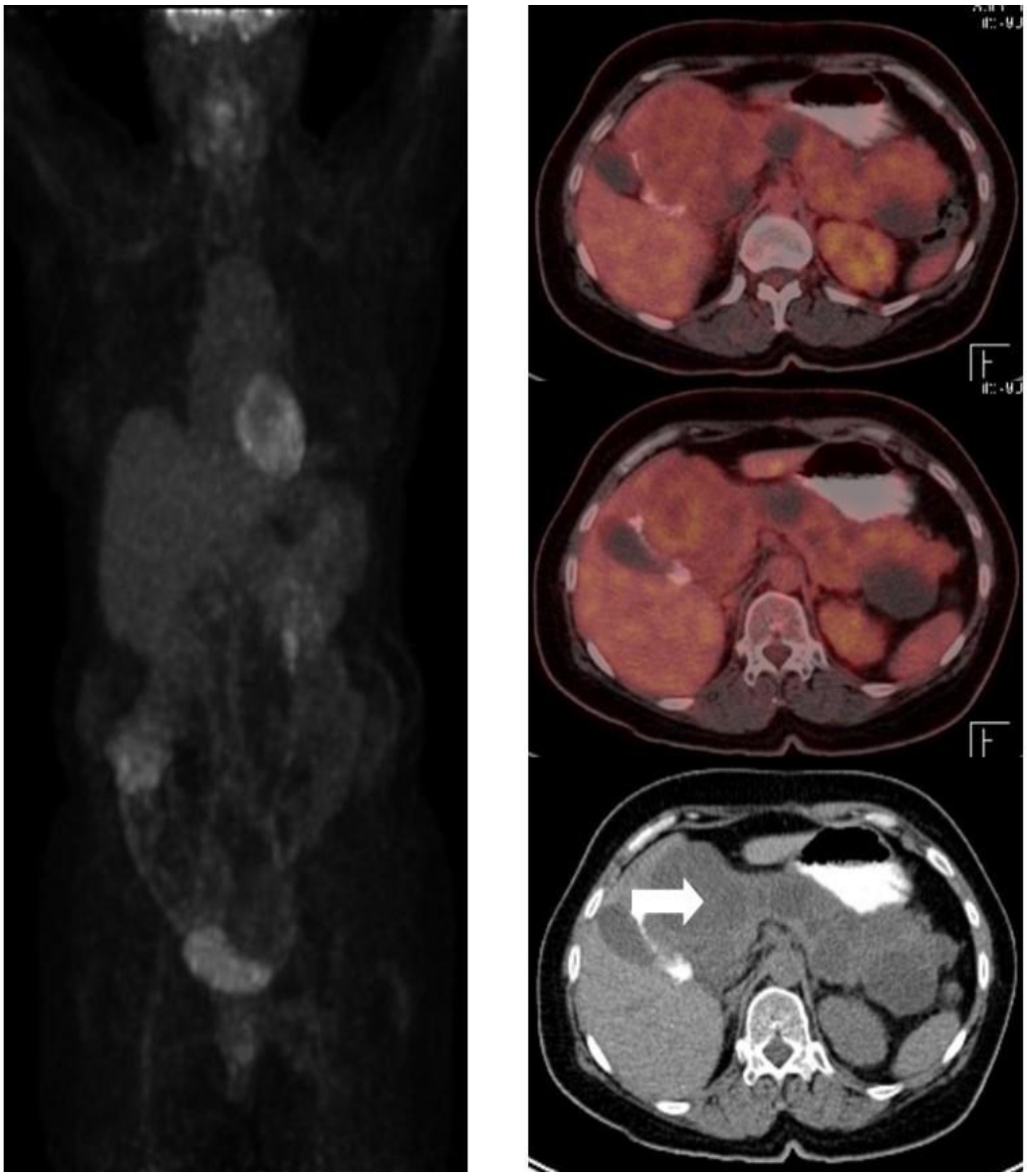
Conditions associated with VHL disease include angiomatosis, hemangioblastomas, pheochromocytoma, renal cell carcinoma, pancreatic cysts (pancreatic serous cystadenoma), endolymphatic sac tumor, and bilateral papillary cystadenomas of the epididymis (men) or broad ligament of the uterus (women) (2,3).

The detection of tumours specific to VHL disease is important in the disease's diagnosis. In individuals with a family history of VHL disease, one hemangioblastoma, pheochromocytoma or renal cell carcinoma may be sufficient to make a diagnosis. As all the tumours associated with VHL disease can be found sporadically, at least two tumours must be identified to diagnose VHL disease in a person without a family history (4,5).



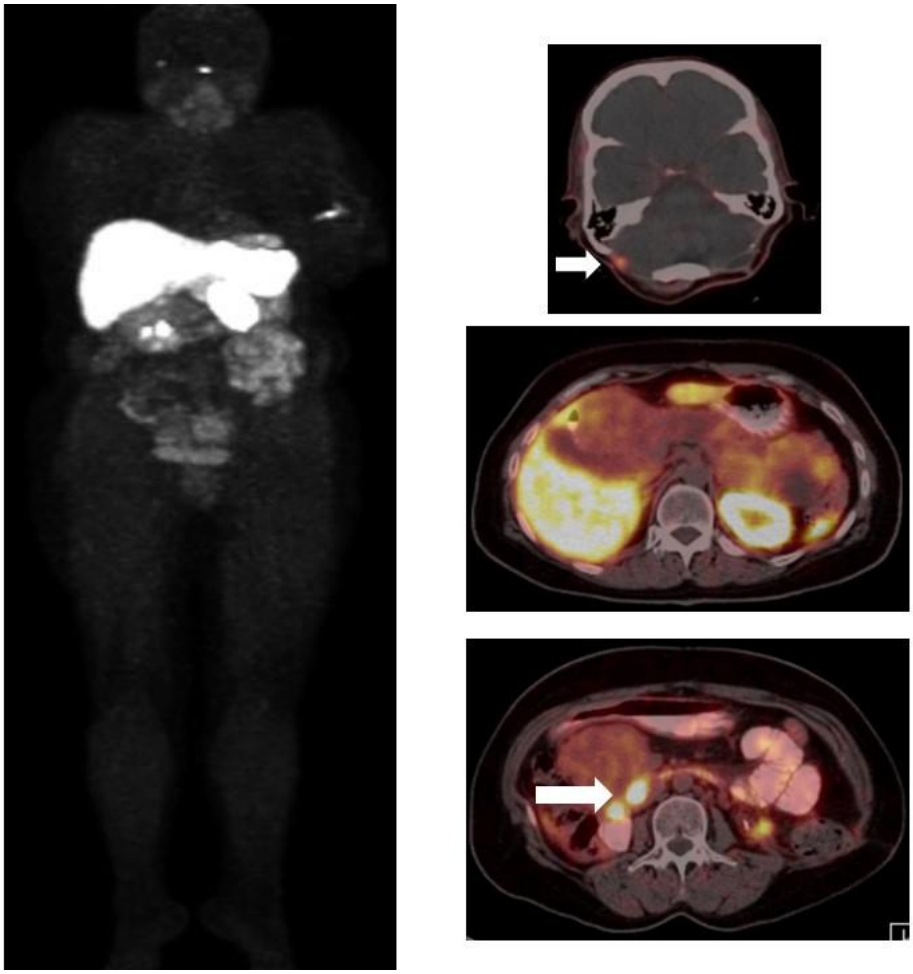
2. Figure 1:

A 49 years old women patient with known history of Von Hippel-Lindau disease underwent FDG PET-CT imaging for metabolic characterisation of pancreatic mass. In PET-CT imaging, approximately 7x5 cm in size at the pancreatic tail at the posterior level of the stomach, 24 mm in the corpus and 7x7.5 cm in the head of the pancreas with low FDG affinity (SUVmax: 4.4) and possibly cystic lesions were observed. There was a craniotomy defect in the left occipital cortex. In addition, two mass lesions of approximately bigger one 4 cm with heterogeneous density and low FDG uptake was detected in the lower pole of the left kidney and right kidney. The pathology results of the patient who was operated for the masses detected in the right and left kidney was reported as clear cell carcinoma.



3. Figure 2

In the FDG PET-CT imaging performed for the follow-up of cystic lesions in the pancreas and for the restaging of renal cell carcinoma, cystic lesions in the pancreatic head, body and tail level, which did not show significant changes in the follow-up, and irregular FDG activity secondary to chronic pancreatitis in the surrounding parenchyma were detected. Ga-68 DOTATATE imaging was recommended to the patient with suspected NET.



4. Figure 3

In Ga-68 DOTATATE imaging, besides hypometabolic pancreatic serous cystadenomas that may accompany VHL syndrome, 2 prominent hypermetabolic (SUVmax. 128) nodular lesions with a size of 18 and 8 mm in the vicinity of the right paravertebral area at the level of the pancreatic head-uncinate process were detected, consistent with the diagnosis of low grade NET. In addition, a lesion compatible with a possible hemangioblastoma known to accompany VHL syndrome with a millimetric size (SUVmax: 9.84) located peripherally at the level of the right cerebellar cortex was also detected in the imaging.

Von Hippel-Lindau disease is a familial cancer syndrome with a variety of malignant and benign tumors. There are few reports in the literature. Renal and pancreatic tumors are the most common visceral manifestations of von Hippel-Lindau disease. Since, multipl VHL manifestations such as hemangioblastomas or pancreatic neuroendocrine tumors are known to overexpress somatostatin reseptors (6-8). Bilateral epididymal cystadenomas showing intensely increased Ga-68 DOTA peptid activity in a VHL patient was reported in a case report (9). Data from off-label use of Octreotide-LAR in a 64-year-old female patient with severe VHL phenotype showed that 9 months of octreotide treatment led to resultant clinical stability and decreased tumor volume. The authors recommended alternative treatment strategy based on the information of somatostatin receptor expression on VHL associated hemangioblastomas (10). Endolymphatic sac and duct that can be either sporadic or associated with von Hippel-Lindau disease. Endolymphatic sac tumor with increased Ga-68 DOTATATE uptake was reported in a case report by Papadakis GZ and collagues (11). Cell surface expression of somatostatin receptors by this tumor, suggests the potential application of somatostatin reseptor imaging using Ga-68-DOTA peptids in the work-up. VHL's spectrum of manifestations is broad with 40 different lesions in 14 different organs (12). Renal cysts and renal cell carcinoma are the most common visceral VHL manifestations and and the most common causes of death in VHL disease (12-13). The introduction of 68Ga-DOTA-conjugated-peptides into clinical practice enabled somatostatin reseptör-imaging with PET, and evolves as the imaging standard of

reference for the detection and characterization of SSTR-positive tumors, with high potential in VHL-disease (14-15). Ga-68 DOTATATE PET/CT confirmed SSRT-overexpression in renal solid tumor in a case report (14). In this current case report both FDG PET-CT and Ga-68 DOTATATE findings of the patient was presented. FDG PET-CT imaging demonstrated low FDG uptake in renal tumors and no uptake in NET, pancreatic cysts and hemangioblastoma. During Ga-68 DOTATATE imaging the patient had a history of right nephrectomy and left partial nephrectomy for bilateral renal clear cell carcinoma. Ga-68 DOTATATE PET/CT demonstrated intensely uptake in NET, and hemangioblastoma.

REFERENCES

1. Von Hippel-Lindau disease, Genetic and Rare Diseases Information Center (GARD) – an NCATS Program". rarediseases.info.nih.gov. Retrieved 2018-04-17
2. Lindsay, Kenneth W; Ian Bone; Robin Callander; J. van Gijn (1991). Neurology and Neurosurgery Illustrated. United States: Churchill Livingstone. ISBN 978-0-443-04345-1
3. Frantzen, Carlijn; Links, Thera P.; Giles, Rachel H. (21 June 2012). "Von Hippel-Lindau Syndrome". Von Hippel-Lindau Disease. GeneReviews at NCBI. University of Washington, Seattle. Retrieved 30 March 2013
4. Maher ER; Glenn GM; Walther M; et al. (June 2011). "von Hippel-Lindau disease: a clinical and scientific review". European Journal of Human Genetics. 19 (6): 617–23
5. Friedrich, CA (December 1, 1999). "Von Hippel-Lindau syndrome. A pleomorphic condition". Cancer. 86 (11 Suppl): 2478–82
6. Leung RS, Biswas SV, Duncan M, et al. Imaging features of von Hippel-Lindau disease. Radiographics. 2008; 28:65–67
7. Ambrosini V, Campana D, Allegri V, et al. 68Ga-DOTANOC PET/CT detects somatostatin receptors expression in von Hippel-Lindau cerebellar disease. Clin. Nucl. Med. 2011; 36:64–65.
8. Oh JR, Kulkarni H, Carreras C, et al. Ga-68 Somatostatin Receptor PET/CT in von Hippel-Lindau Disease. Nucl. Med. Mol. Imaging. 2012; 46:129–133
9. Papadakis G.Z., Millo C, Sadowski SM, et. al. Epididymal cystadenomas in von Hippel-Lindau disease showing increased activity on 68Ga-DOTA-TATE PET/CT. Clin Nucl Med. 2016 October ; 41(10): 781–782
10. Sizdahkhani S, Michael J. Feldman MJ.,Martin G. Piazza MG., et. al. Somatostatin receptor expression on von Hippel-Lindau-associated hemangioblastomas offers novel therapeutic target. Scientific Reports | 7:40822 | DOI: 10.1038/srep40822
11. Papadakis G.Z., Millo C, Sadowski SM, et. al. Endolymphatic sac tumor showing increased activity on 68 Ga-DOTA-TATE PET/CT. Clin Nucl Med. 2016 October; 41(10): 783–784
12. Leung RS, Biswas SV, Duncan M, et al. Imaging features of von Hippel-Lindau disease. Radiographics. 2008; 28:65–67
13. Wood CG, Stromberg LJ, Harmath CB, et al. CT and MR imaging for evaluation of cystic renal lesions and diseases. Radiographics. 2015; 35:125–141.
14. Hofman MS, Lau WF, Hicks RJ. Somatostatin Receptor Imaging with 68Ga-DOTATATE PET/CT: clinical utility, normal patterns, pearls, and pitfalls in interpretation. Radiographics. 2015; 35:500–516
15. Papadakis GZ, Bagci U, Sadowski SM, et al. Ectopic ACTH and CRH co-secreting tumor localized by 68Ga-DOTA-TATE PET/CT. Clin Nucl Med. 2015; 40:576–578
16. Papadakis G.Z., Millo C, Sadowski SM, et. al. Kidney tumor in a von Hippel-Lindau (VHL) patient with intensely increased activity on 68 Ga-DOTA-TATE PET/CT. Clin Nucl Med. 2016 December; 41(12): 970–971

© Author(s) 2021. This work is distributed under https://creativecommons.org/licenses/by-sa/4.0/