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IMAGE | PANCREAS

Von Hippel-Lindau Disease Presenting as Obstructive Jaundice

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CASE REPORT

A 34-year-old woman who had been diagnosed with type 2 diabetes mellitus 4 years ago presented with painless progressive jaundice without cholestatic symptoms for the last 1 month. Her family history included the fourth-degree consanguineous marriage of the patient and the death of 8 family members, from either renal or brain cancers which were never investigated. Abdominal computed tomography showed complete replacement of pancreatic parenchyma with multiple cysts of varying sizes compressing the common bile duct (CBD) in pancreatic head region with intrahepatic biliary radicle dilatation with bilateral renal cysts (Figure 1). Magnetic resonance cholangiopancreatography showed multiple simple cysts with a normal main pancreatic duct (Figure 2). Screening magnetic resonance imaging of the brain and spine showed hemangioblastoma in the superior vermis and spinal cord at D9 level (Figure 3). Based on these findings, the patient was diagnosed with von Hippel-Lindau (VHL) disease. Endoscopic retrograde cholangiography revealed dilated proximal CBD with extrinsic compression in mid-CBD (Figure 4). A plastic biliary Cotton-Leung stent was placed. The findings were disclosed to the patient and surgery to remove the cysts causing obstructive jaundice was recommended. As the patient was unwilling to undergo surgery, she has to have her stent replaced every 3 months.

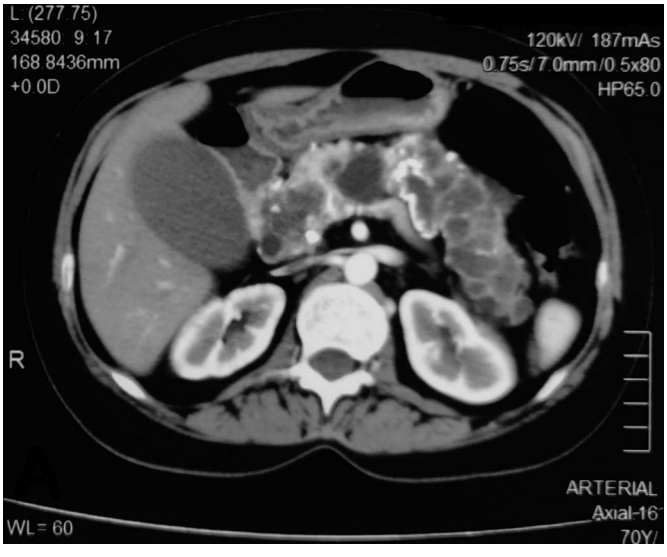

Figure 1. Replacement of pancreatic parenchyma with multiple cysts causing common bile duct (CBD) compression in head region with resultant upstream CBD dilatation.

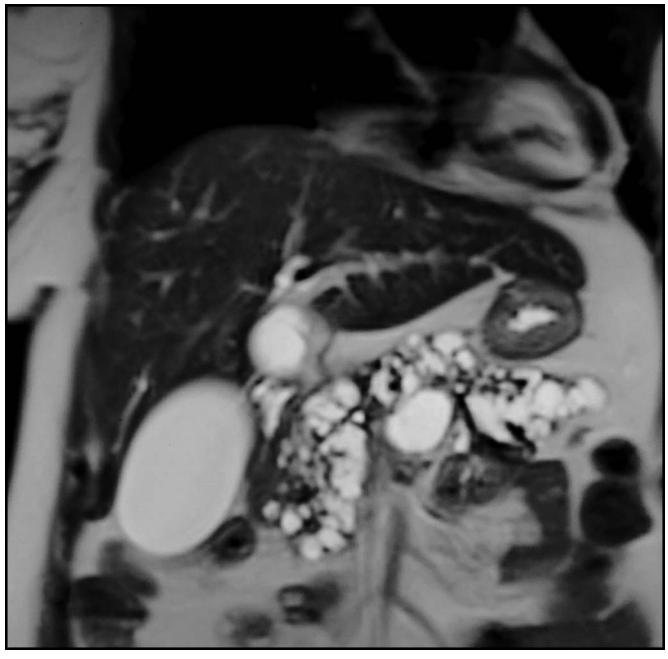

Figure 2. Magnetic resonance cholangiopancreatography showing enlargement of the entire pancreas with multiple simple cysts of varying size causing the common bile duct compression in the pancreatic head.

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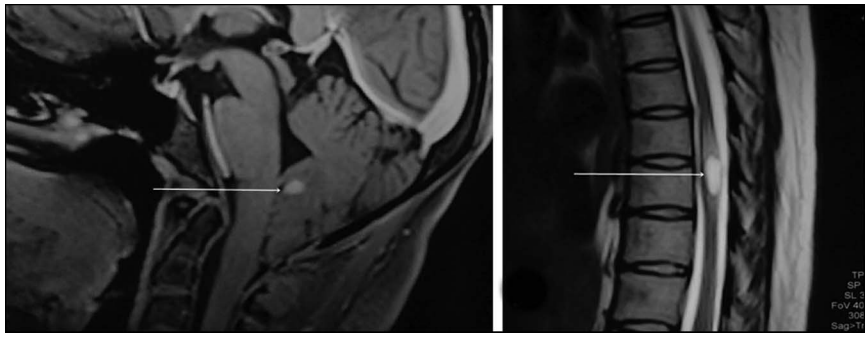

Figure 3. Magnetic resonance imaging of the brain and spine showing T2 hyperintense lesions in the superior vermis and D9 vertebrae body level with mild perilesioned edema.

VHL disease is an autosomal dominant multisystem disease associated with various tumors and cysts involving the central nervous system, retina, kidneys, adrenal glands, and pancreas. The loss of functional VHL protein leads to a high level of nondegraded hypoxia-inducible factor leading to accelerated tumor growth.[1] The diagnostic criteria for VHL disease are (i) greater than one CNS hemangioblastoma, (ii) CNS hemangioblastoma in combination with visceral manifestation of VHL disease, and (iii) any manifestation and known family history of VHL disease.[2] VHL disease is classified into 2 types: type 1 includes patients without pheochromocytoma and type 2 includes patients with pheochromocytoma. Pancreatic involvement can be seen in both types. The incidence of VHL disease is approximately 1 in 36,000 live births, and its penetrance is >90% by the age of 65 years.[3] The involvement of the pancreas in VHL disease is seen in 32.7%–77.2% cases, whereas isolated involvement of the pancreas is seen in approximately 7.6% of cases.[4] Pancreatic lesion can be solid or cystic, with solid lesions being neuroendocrine tumors and cystic lesions being simple cysts or serous cystadenomas with very low risk of malignancy in the cystic lesions.[5,6] Presentation of pancreatic lesion range from asymptomatic disease (30%–70%) to symptoms such as abdominal pain, obstructive jaundice, or gastrointestinal bleeding. The management of obstructive jaundice varies according to the cause of obstruction and operability of lesion.

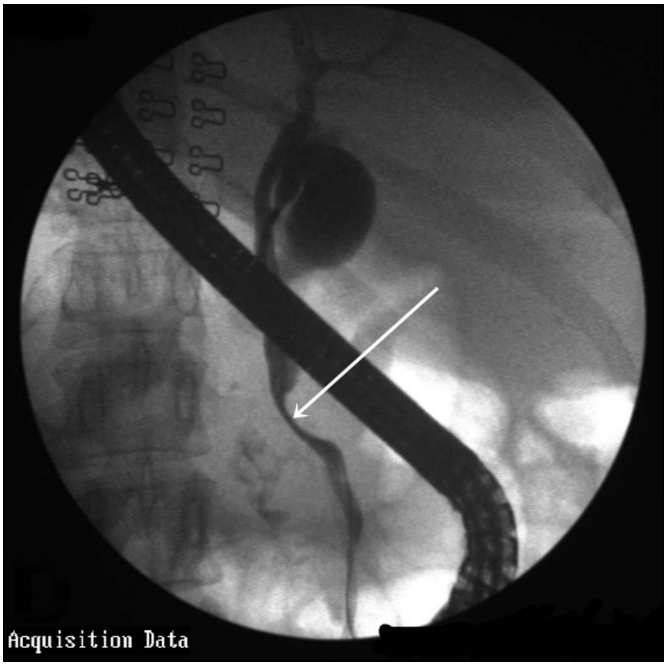

Figure 4. Endoscopic retrograde cholangiography revealed the mildly dilated proximal common bile duct with narrowing in the mid-distal.

DISCLOSURES

Author contributions: All authors contributed equally to this manuscript. S. Giri is the article guarantor.

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