Von Hippel-Lindau syndrome with pancreatic adenocarcinoma (with videos)

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A 56-year-old male, who has been experiencing recurrent abdominal pain for 10 years and aggravating for 6 months, presented to our hospital. He had a history of pancreatitis for 10 years and was found to have hypertension, diabetes, and kidney stones for 1 year. Initial laboratory tests showed a CEA level of 5.42 ng/mL (normal: 0–5 ng/mL) and a CA-199 level of 320 U/mL (normal: 0–39 U/mL). Enhanced computed tomography of the abdomen revealed polycystic lesions in the liver, pancreas, and bilateral kidneys. A suspicious low-density area was identified in the pancreatic body [Figure 1a]. EUS confirmed the presence of a solid hypoechoic lesion in the body of the pancreas [Figure 1b]. Color Doppler confirmed that the cystic lesion in the neck of pancreas compressed the portal vein and the splenic vein [Figure 1c and video 1]. Color Doppler confirmed that the main pancreatic duct did not communicate into the cyst in the neck of pancreas [Figure 1d and video 2]. We punctured the solid lesion with a 22-G needle guided by EUS for three passes and got enough tissue core. Cytological results of the samples were grade IV, which was

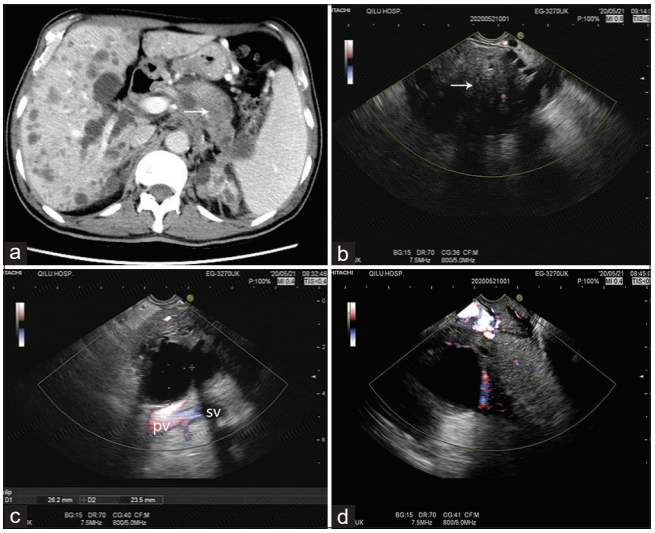

Figure 1. (a) Enhanced computed tomography of the abdomen revealed a suspicious low-density area in the body of the pancreas. The pancreatic cervical cyst compressed the SV (arrow). (b) Hypoechoic lesion in the body of the pancreas (arrow). The boundary of the solid lesion was not clear and echo attenuation was obvious in the rear. (c) EUS color Doppler: The cystic lesion compressed PV and SV. (d) The cyst did not communicate into the main pancreatic duct. PV: Portal vein; SV: Splenic vein

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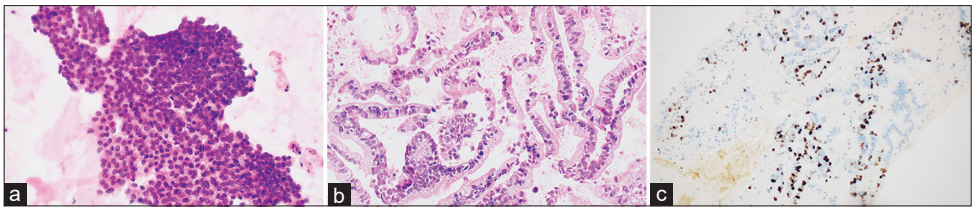

Figure 2. (a) Cytological result was grade IV, which was neoplastic. Intraductal papillary mucinous tumor with moderate dysplasia was not excluded. (b) Histopathological analysis confirmed the diagnosis of a highly differentiated carcinoma. (c) Immunohistochemistry showed that the positive rate of Ki 67 was 25%

neoplastic. Intraductal papillary mucinous tumor with moderate dysplasia was not excluded [Figure 2a]. Histopathological results of the samples confirmed the diagnosis as highly differentiated carcinoma [Figure 2b]. Immunohistochemistry showed that the positive rate of Ki 67 was 25% [Figure 2c]. The results of gene detection of the peripheral blood lymphocytes showed that Von Hippel-Lindau (VHL) gene test was positive, while CDKN1B, MEN1, and NF1 gene tests were negative. The final diagnosis was VHL syndrome with pancreatic adenocarcinoma. After multidisciplinary discussions among experts from pancreatic surgery, oncology, and imaging department, it was concluded that surgery was not recommended for this patient. Therefore, chemotherapy was administered and the patient was stable when followed up after 2 months.

VHL syndrome is a rare autosomal dominant inherited multisystem disease, which is characterized by benign or malignant lesions of multiple organs. Abdominal organs are often involved. Pancreatic lesions can be the only abdominal manifestation of VHL disease (VHLD) in some cases.[1] Pancreatic lesions, including cysts, may precede any other manifestations of VHLD by several years, and recognition of them permits earlier diagnosis in patients being screened for VHLD.[2,3] Computed tomography, magnetic resonance imaging, and EUS can help to detect lesions[4], but only EUS easily aid to achieve pathological diagnosis of the pancreas. Once the diagnosis has been made, long-term surveillance and screening of kindred should be scheduled.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal his identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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