and was thought to have a malabsorption syndrome. However, her condition was not definitively diagnosed and she progressed to become malnourished and severely emaciated. Due to the rarity of WD, the specific PAS staining was never performed, which lead to the delay in her diagnosis.

A recently published paper described a case of WD diagnosed by the capsule endoscopy [5], this non-invasive and innovative technique could be utilized in future to follow-up these patients and to verify the resolution of macroscopic findings. We decided to follow up our patient using capsule endoscopy rather than perform endoscopic procedures, which are very uncomfortable and alternatively requires sedation. Capsule endoscopy allowed us to document easily the progressive improvement of lesions and villous regrowth, which are two very important prognostic parameters.

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A rare cause of obstructive jaundice - VHL syndrome

Von Hippel-Lindau syndrome(VHL), is a rare genetic disorder characterized by visceral cysts and benign tumors in multiple organ systems. It is an autosomal dominant condition associated with hemangioblastomas of the central nervous system and retina, renal cysts, renal cancer, pheochromocytoma, pancreatic cysts, tumours of the endolymphatic sac in the ear and epididymal cystadenomas. Pancreatic involvement, especially as isolated organ involvement, is not common in patients with VHL.[1, 2] Most pancreatic lesions are simple cysts and rarely cause symptoms or develop into malignant tumors. Extrahepatic biliary obstruction secondary to pancreatic cysts is a rare occurrence in VHL syndrome.[3,4] We describe a patient with VHL who had biliary obstruction due to pancreatic cysts.

Case Report

A 30 year old female patient presented with jaundice for 25 days, gradual onset and progressive, with generalised pruritus and pale coloured stools. There was no history of fever, vomiting, abdominal pain, abdominal distension, melena, hematemesis, pedal oedema, night blindness, bleeding gums or diarrhoea. The patient gave a history of having undergonea brain surgery, the details not available. There was history of visceral cystic lesions in her elder sister. On physical examination, scratch marks werepresent all over the body. The right kidney was palpable. A cystic lesion was palpable in epigastrium.

Investigations showed a haemoglobin of 11.6 g/dL (reference range: 12-18 g/dL), a total leukocyte count of 8 x109 /L (reference range: 4-11 x109 /L) and platelets 274 $x10^3$ /mm^3 (reference range: 150-400 $x10^3$ /mm^3). Her liver function tests revealed a bilirubin count of 25.7 mg/dL (reference range: 0-1.0 mg/dL), direct bilirubin of 18 mg/dL, aspartate aminotransferase (AST) 42 IU/L (reference range: 10-21 IU/L), alanine aminotransferase (ALT) 52 IU/L (reference range: 2-15 IU/L) and serum alkaline phosphatase of 609IU/L (reference range:20-140IU/L). Serum amylase and lipase values were normal.

Retinal hemangiomas were observed on fundus examination **(Figure 1)**. Ultrasound examination showed dilated intrahepatic biliary radicles, hepatic ducts and supra pancreatic common bile duct. There were multiple cysts in both the pancreas and the kidneys. The hepatic vein and the portal vein were normal. Contrast enhanced computed tomography showed intrahepatic and extrahepatic biliary dilatation with multiple cysts in pancreas causing compression at the lower end of the common bile duct. Multiple cysts were seen in both the kidneys **(Figure 2)**. Magnetic resonance imaging of abdomen showed dilated intrahepatic and extrahepatic bile duct, distended gall bladder, multiple cysts in pancreas and kidney. Magnetic resonance imaging of the brain showed hemangioblastomas in right superior cerebellar hemisphere and left parietal region **(Figure 3)**. Diagnosis of Von Hippel Lindau syndrome was made with these finding. The patient was adviseda hepaticojejunostomy.

Discussion

An individual is diagnosed with VHL if at least one of the followin gcriteria are met[5]:

1. The individual has at least two of the manifestations stated below.
2. The individual has at least one of the manifestations stated below, and a pathogenic mutation in VHL gene or at least one first-degree relative with VHL.

Diagnostic VHL manifestations include:

1. The retina: Hemangioblastoma
2. The cerebellum, the medulla oblongata, or the spinal cord: Hemangioblastoma
3. The inner ear: Endolymphatic sac tumour (ELST)
4. The kidneys: Renal cell carcinoma

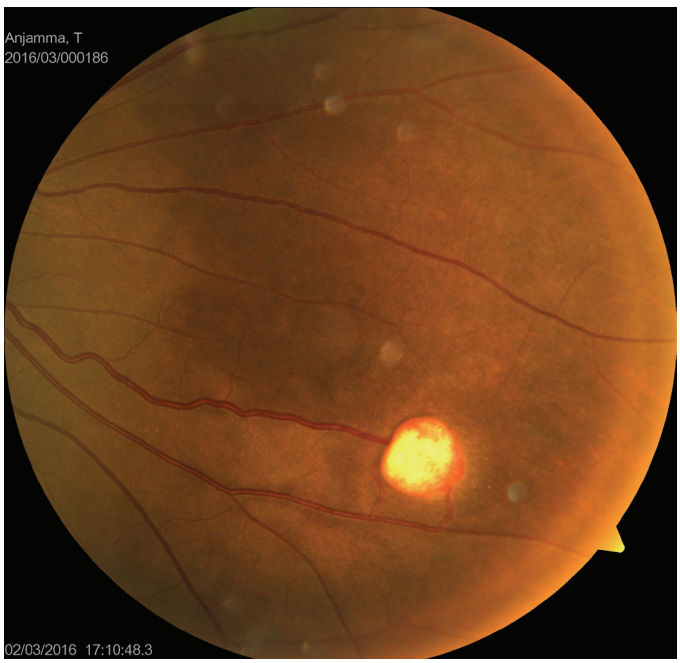

Figure 1: Image showing fundus with hemangioma in right eye.

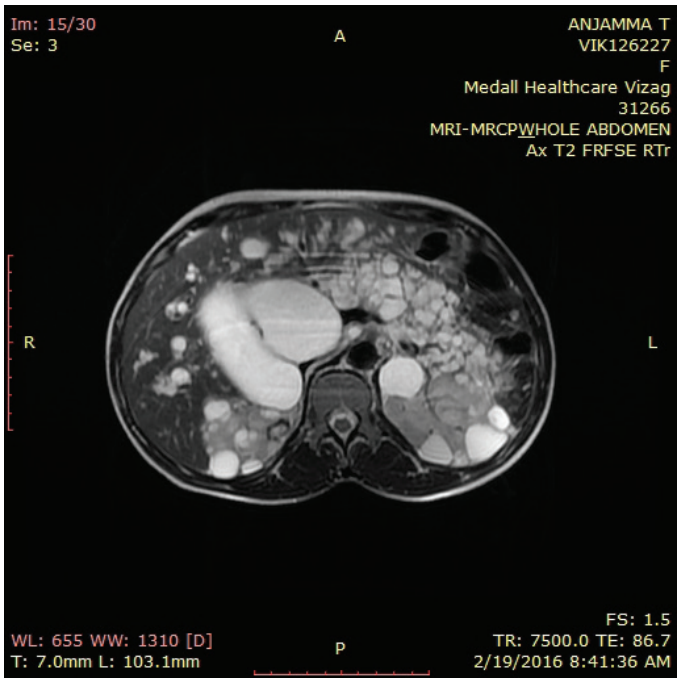

Figure 2: MRI abdomen showing dilated bileduct, cysts in pancreas and kidney.

5. Pheochromocytoma, paraganglioma, and/or glomus tumour
6. The pancreas: Neuroendocrine neoplasms and/or multiple cysts

Our patient was diagnosed with VHL syndrome on the basis of the presence of bilateral retinal angiomas, CNS hemangioblastomas,pancreatic and renal cysts.

Pancreatic involvement occurs in 17% to 56% of patients with VHL.[1,2] Though simple pancreatic cysts and cystadenomas represent the most common pancreatic lesions in VHL, literature also reports other common ones such as neuroendocrine tumors as well as the rarer ones such as adenocarcinomas, hemangioblastomas and metastases from renal cell cancer.[3] Most patients with pancreatic involvement remain asymptomatic, although some patients present with abdominal pain, with or without evidence of pancreatitis.

In conclusion, the management of obstructive jaundice in VHL disease depends largely on the cause of the obstruction and the operability of the lesion. Conservative management is advocated in the majority of VHL patients with pancreatic involvement. Surgery is indicated in symptomatic patients (obstructive jaundice, upper gastrointestinal bleed).

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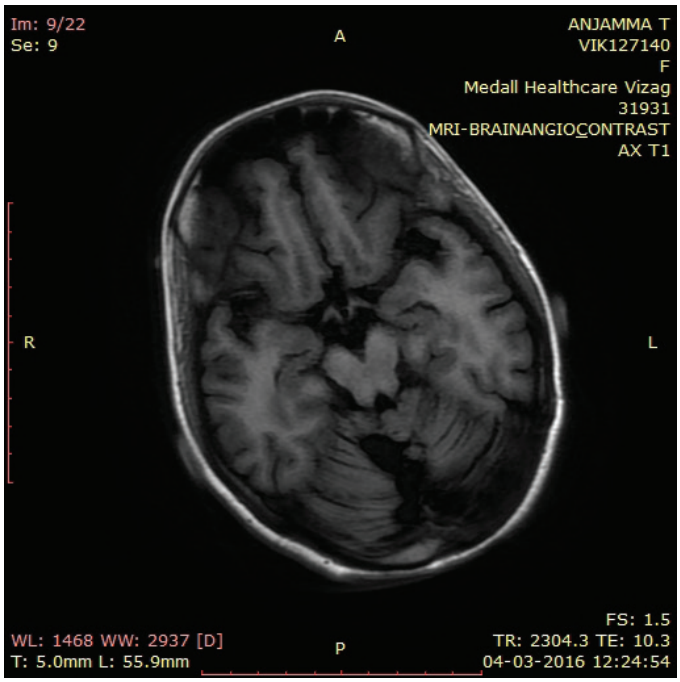

Figure 3: MRI brain showing solid cystic lesions with enhancing mural nodule in right superior cerebellar hemisphere and left parietal region s/o hemangioblastoma.

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