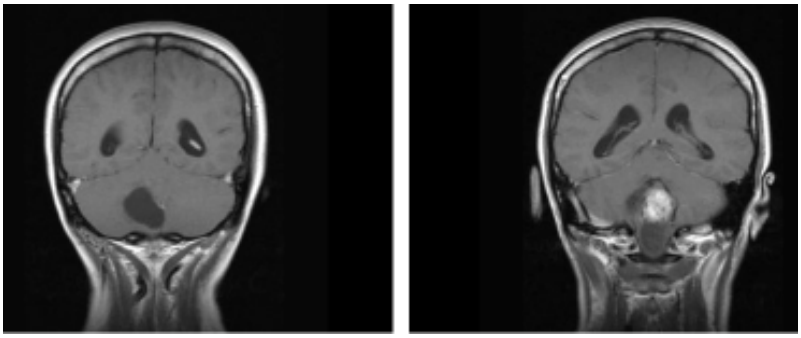
Putting Together Lesions in the Brain, Retina, Kidney and Pancreas

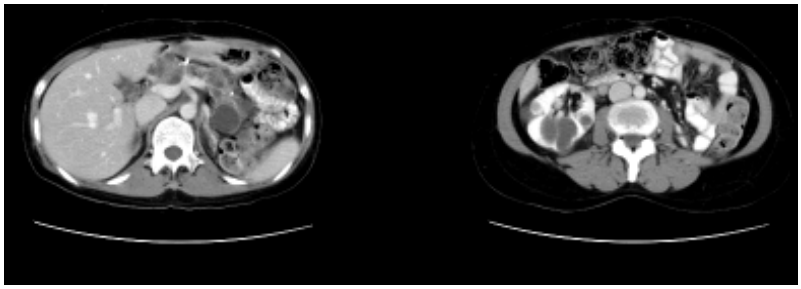
Deidre Anne De Silva,[1] *MBBS, MRCP, FAMS*, Andrew BS Pan,[2] *MBBS, MRCP, FAMS*

Quiz

A 40-year-old Malay lady presented with a 6-month history of headache. She had a past history of a left nephrectomy 6 years ago. Neurological examination revealed generalised brisk limb reflexes and right ataxia. Additional clinical findings were a ballotable right kidney and bilateral retinal capillary hemangiomas. Brain magnetic resonance imaging (MRI) and abdominal computed tomographic (CT) imaging are shown below.



MRI brain.



CT abdomen.

What is the diagnosis?

a. Autosomal dominant polycystic kidney disease
b. Autosomal recessive polycystic kidney disease
c. Von Hippel-Lindau disease
d. Multiple endocrine neoplasm type II
e. Vascular metastases

Answer

The brain MRI shows a lesion in the posterior cranial fossa involving the right cerebellar hemisphere and vermis extending to the brain stem. The lesion consists of solid and cystic components. Of note, there are multiple signal flow voids within the lesion representing blood vessels. The radiological appearance is consistent with a haemangioblastoma. The abdominal CT shows multiple cytic lesions in the pancreas and right kidney, as well as enlarged lymph nodes.

The diagnosis is Von Hippel-Lindau disease. This is evidenced by the combination of a posterior fossa haemangioblastoma, retinal haemangioblastomas, right kidney cystic lesions, pancreatic cystic lesions and a previous left nephrectomy. The histology of the left kidney lesion was confirmed as renal cell carcinoma.

The most common features of Von Hippel-Lindau disease are central nervous system and retinal haemangioblastomas, renal cysts and carcinoma, pancreatric cysts and neuroendocrine tumours and pheochromocytoma in the adrenal gland.[1] The diagnosis in this patient was confirmed with genetic analysis which revealed a non-sense mutation in the loci of the Von Hippel-Lindau tumour suppressor gene on the short arm of chromosome 3.[2]

REFERENCES

1. Shuin T, Yamasaki I, Tamura, K, Okuda, H, Furihata M, Ashida S. Von Hippel-Lindau disease: molecular pathological basis, clinical criteria, genetic testing, clinical features of tumors and treatment. Jpn J Clin Oncol 2006;36:337-43.
2. Kaelin WG Jr. Molecular basis of the VHL hereditary cancer syndrome. Nat Rev Cancer 2002;2:673-82.

[1] Singapore General Hospital Campus, National Neuroscience Institute, Singapore
[2] Pan Neurology, Epilepsy and Sleep Disorders Clinic, Mount Elizabeth Medical Centre, Singapore
Address for Correspondence: Dr Deidre Anne De Silva, Department of Neurology, Singapore General Hospital, Outram Road, Singapore 169608.
Email: deidre.a.de.silva@sgh.com.sg