A new diabetic patient with an abdominal mass

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A 47-year-old woman was referred to the new patient diabetic clinic with typical symptoms of hyperglycaemia. On routine abdominal examination, a mass was found on the left side of the abdomen which was ballottable and bimanually palpable. There was a history of 'benign retinal tumour' several years ago which did not need active treatment. Further enquiry revealed this to have been a retinal angioma. Routine laboratory tests on blood and urine were unremarkable except for findings of diabetes mellitus. She went on to have ultrasound scan followed by computed tomography (CT) of the abdomen (figure).

Questions

- 1 What is the main abnormality seen on the CT scan and what is the radiological diagnosis?
- 2 What other abnormalities are present on the CT scan?
- 3 What condition does the patient have?
- 4 What other abnormalities may occur in this condition?

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Figure CT scan of the abdomen

Answers

QUESTION 1

There is a large solid tumour arising anteriorly from the lower half of the left kidney, with a necrotic centre. There are two solid tumours of the right kidney, with the anterior tumour predominantly necrotic. These appearances are consistent with bilateral hypernephromas.

QUESTION 2

There are multiple bilateral simple renal cysts, multiple pancreatic cysts and a small left adrenal mass.

QUESTION 3

The combination of retinal angioma and bilateral hypernephromas indicates the diagnosis of von Hippel-Lindau syndrome.

QUESTION 4

von Hippel-Lindau syndrome is a rare genetic disorder with multi-organ involvement. The most common features are retinal angiomas (51%), haemangioblastoma of the central nervous system (46%), and renal lesions (33%).¹ The pancreatic features of the condition include pancreatic cysts, endocrine deficiency,² exocrine deficiency,³ metastatic islet cell tumour,⁴ and acute pancreatitis.⁵ The other features are phaeochromocytomas (24%), epididymal cystadenomas and, rarely, carcinoid tumour of the bile duct⁶ and deafness due to bilateral endolymphatic sac tumours.⁷

Discussion

von Hippel-Lindau syndrome is an autosomal dominant inherited disease with a prevalence of 1:40 000.¹ The von Hippel-Lindau (VHL) gene has been localised and cloned at human chromosome 3p25-p26.⁸ The VHL gene product has been found to suppress growth of renal cell carcinoma lines *in vitro*.⁹ The association of von Hippel-Lindau disease with renal cell carcinoma may be explained either by mutation of or loss of both alleles of the VHL gene or by disturbance in the function of the VHL gene product.⁹⁻¹⁰

Although commonly referred to as cerebello-retinal haemangioblastomatosis, there are other significant clinical associations like renal cell carcinoma and phaeochromocytoma. Our

patient was known to have retinal angiomas. Subsequent CT scan of the brain did not show evidence of intracranial haemangioblastoma. Both of the renal tumours were biopsied under CT guidance and histological examination showed adenocarcinoma. Urinary normet-adrenaline screening for phaeochromocytoma was negative.

Although pancreatic cysts are common, pancreatic endocrine or exocrine deficiency is uncommon. Our patient presented with symptoms of diabetes mellitus which was treated by dietary means. There was no clinical suggestion of steatorrhoea. Central nervous system lesions in von Hippel-Lindau disease present as haemangioblastomas and metastatic renal carcinomas. The haemangioblastomas are mainly intracranial (74%), the rest being located in the spinal cord.¹¹ Retinal angiomas present as both capillary and cavernous haemangiomas. The renal lesions include renal angiomas, renal cysts and renal carcinomas. Renal carcinomas are frequently bilateral and, if not diagnosed early, tend to metastasize. Our patient went on to have bilateral nephrectomy and renal replacement therapy with chronic ambulatory peritoneal dialysis. Parenchymal sparing surgery is sometimes used in selected patients with renal carcinoma.¹²

The initial presentation of von Hippel-Lindau syndrome could be to a medical or surgical speciality but more awareness of the syndrome and its associations will lead to early diagnosis. The finding of retinal angiomas or CNS haemangioblastomas should lead to radiological assessment of the kidneys. Screening of first degree relatives will help in early identification of affected individuals.

Formal referral to a medical genetics service will help detailed screening of the patients and their relatives. Medical genetics clinics normally maintain computer registers of such families and are best placed to ensure their adequate prolonged follow-up.

Final diagnosis

von Hippel-Lindau syndrome.

Keywords: von Hippel-Lindau syndrome; renal cell carcinoma; diabetes mellitus

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Fever and dyspnoea in a young man with a rash

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A 36-year-old male smoker presented with a 7-day history of fever, chills and malaise. He admitted to increasing dyspnoea over the past 2 days. There was no history of cough, nausea or vomiting. The patient had developed crops of erythematous pruritic vesicles on his face, trunk, and extremities 5 days prior to admission. He admitted to being exposed to a child with a similar rash, and his own three sons had developed a similar rash.

On examination he appeared to be severely ill. Skin examination revealed multiple vesicular lesions with crusting, mostly on his chest, back, and extremities. An arterial blood gas revealed pH 7.42, pO_2 6.63 kPa, pCO_2 4.03 kPa, and O_2 saturation of 87% on room air. He was febrile with a temperature of 39°C. His lungs were clear to auscultation bilaterally. Routine blood tests were normal except for neutrophil leucocytosis (white blood cells of $10 \times 10^9/l$ with 89% neutrophils, 3.3% lymphocytes, 6.4% monocytes, 0.1% eosinophil, and 0.8% basophil). His chest X-ray is shown in the figure.

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Figure Chest X-ray

Questions

- 1 What is the differential diagnosis?
- 2 What treatment would you advise?
- 3 What are the risk factors and other complications associated with this condition?