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Self-assessment corner

Abdominal tumours and neurofibromatosis

AJ Martin, RCN Williamson

A 50-year-old, black female nurse was found to be hepatitis B positive in a routine screen. She was entirely asymptomatic and had never been jaundiced. Liver function tests were normal, but an ultrasound scan showed dilatation of the bile duct plus a cystic lesion near the pancreatic tail. Duodenoscopy revealed a small tumour at the ampulla of Vater, and biopsy showed 'adenocarcinoma with areas of carcinoid differentiation'.

The patient had Type 1 neurofibromatosis and three years earlier had developed mild hypertension with several episodes of paroxysmal headache. A series of urinary vanillyl mandelic acid levels had been normal at that time, and the hypertension was well controlled with a beta-blocker.

Physical examination was unremarkable apart from the iris hamartomas ('Lisch nodules') and cutaneous neurofibromas typical of Type 1 neurofibromatosis. A gastrointestinal hormone screen showed a minimally increased plasma somatostatin level of 171 pmol/l (upper limit of normal (uln) 150), but an oral glucose tolerance test was normal. Urinary catecholamine levels were slightly raised but not diagnostic of phaeochromocytoma: noradrenalin 0.85 μmol/24 h (uln 0.59), adrenalin 0.75 μmol/24 h (uln 0.19), dopamine 2.1 μmol/24 h (uln 3.27). Abdominal computed tomography (CT) was performed (figure).

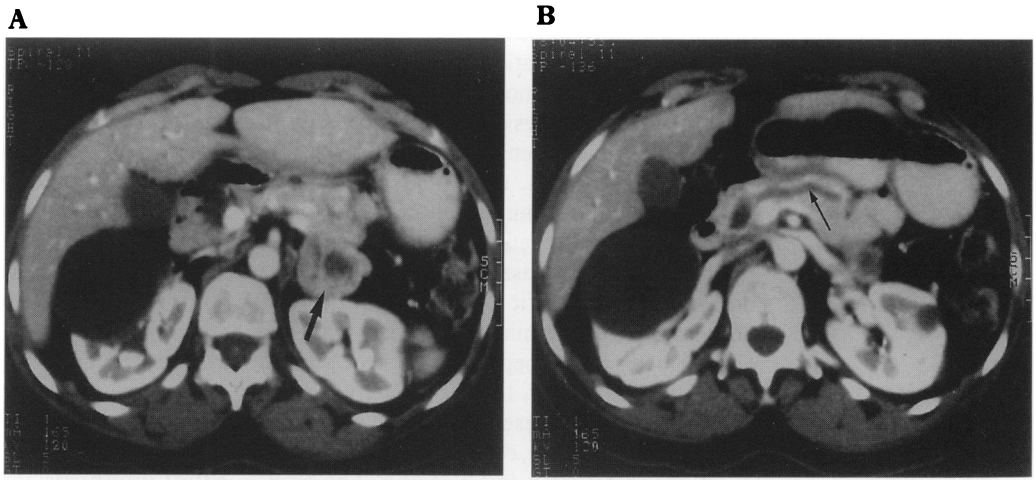

Figure Contrast-enhanced spiral CT scans

Questions

1 What do the CT scans show?
2 What is the most likely diagnosis?

Department of Surgery, Royal Postgraduate Medical School, Hammersmith Hospital, London, UK
AJ Martin
RCN Williamson

Correspondence to
Professor RCN Williamson,
Department of Surgery,
Hammersmith Hospital, Du Cane Road, London
W12 0NN, UK

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Answers

QUESTION 1
Image A shows a cystic lesion anterior to the upper pole of the left kidney while in B there is dilatation of the main pancreatic duct. The duodenal tumour cannot be identified and there is an incidental right renal cyst.

QUESTION 2
Type 1 neurofibromatosis with probable duodenal somatostatinoma and phaeochromocytoma.

Treatment

At operation, the left-sided lesion was found to arise from the adrenal gland and was excised under α-adrenergic blockade. Its histological appearances were typical of a phaeochromocytoma. The ampullary tumour was completely resected by pylorus-preserving proximal pancreatoduodenectomy. Histological features included a glandular architecture with intraluminal psammoma bodies and positive staining for somatostatin.

Discussion

The gastrointestinal features of Type 1 neurofibromatosis (box 1) include stromal tumours, abnormalities of the enteric innervation, and an association with a distinctive somatostatin-rich duodenal carcinoid[1]. Of about 60 such tumours described to date, more than half have been in Type 1 neurofibromatosis patients, and duodenal somatostatinoma is the commonest peri-ampullary tumour in Type 1 neurofibromatosis.[2] These tumours (box 2) are histologically unusual, and unlike their pancreatic counterparts they seldom produce the 'somatostatinoma syndrome' of diabetes, dyspepsia, diarrhoea and cholelithiasis. Rather, they are usually asymptomatic until they present with local complications such as jaundice, cholangitis, pancreatitis, duodenal obstruction and bleeding. The prognosis is good, even in the presence of metastases, and a complete resection is usually curative.[3]

The frequency of phaeochromocytoma in Type 1 neurofibromatosis is over 1%, more than ten times that in the general population.[4]

Type 1 neurofibromatosis: gastrointestinal features

Benign stromal tumours
- neurofibroma, leiomyoma
- in up to 25% of autopsies
- usually asymptomatic

Enteric neuronal hyperplasia
- significant dysmotility in 10% of patients
- may mimic Hirschsprung's disease

Duodenal somatostatinoma

Box 1

Duodenal somatostatinoma

- glandular architecture and psammoma bodies may lead to confusion with adenocarcinoma
- presents with local complications
- excellent prognosis after resection
- those associated with Type 1 neurofibromatosis are usually in the immediate peri-ampullary region and may have a predilection for black patients

Box 2

Phaeochromocytoma seems to be particularly common in Type 1 neurofibromatosis patients who also have duodenal somatostatinoma,[5] and it has been recommended that the pre- and per-operative management of patients with Type 1 neurofibromatosis suspected of having phaeochromocytoma or somatostatinoma should include the search for a second tumour. This triad may represent a form of multiple endocrine neoplasia, but to date there is no evidence that it is inherited as a specific trait as no cases have been reported in members of the same family.[1]

Final diagnosis

Type 1 neurofibromatosis with somatostatinoma and phaeochromocytoma

Keywords: neurofibromatosis, somatostatinoma, phaeochromocytoma

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