An unusual presentation of Multiple Endocrine Neoplasia 1 (MEN1)

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Case history:
A 36 year-old man was referred to our department for further evaluation of a large adrenal and multiple liver mass lesions. These were found on an abdominal ultrasound scan requested because of a three-week history of mild, episodic abdominal pain. On admission, he was generally well and otherwise asymptomatic. He had no history of weight loss and, on direct questioning, described having suffered from mild night sweats for over 10 years. There was no significant family history of any medical problems. There were no findings on examination.

Investigations and method:
His diagnostic work-up included a detailed metabolic evaluation, cross-sectional and radionuclide imaging studies and, subsequently in the light of the results, genetic testing.

Results and treatment:
Hyperparathyroidism was evident, with a serum corrected calcium 3.04mmol/L (2.15-2.65), PTH 16pmol/L (1.6-6.9) and a twenty-four hour urine calcium excretion of 11.1mmol (2.5-7.5). Ultrasound of the neck showed three enlarged parathyroid glands (two ~1.3cm and one 8mm) and a normal thyroid. Two twenty-four hour urine collections for metanephrines (nmol/24hrs) are shown below (urine volumes of 1,979ml and 2,264ml respectively).

Plasma noradrenaline was raised at 22.2nmol/L (<4.14). Plasma glucagon was 65pmol/L (<50) and other fasting gut peptides were normal, as was serum calcitonin.

<table>
<thead>
<tr>
<th></th>
<th>Metadrenaline</th>
<th>Normetadrenaline</th>
<th>3-methoxytyramine</th>
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</thead>
<tbody>
<tr>
<td>1st Collection</td>
<td>974</td>
<td>70,318</td>
<td>3,255</td>
</tr>
<tr>
<td>2nd Collection</td>
<td>1,434</td>
<td>71,690</td>
<td>3,222</td>
</tr>
<tr>
<td>Normal Range</td>
<td>&lt;2,000</td>
<td>&lt;4,400</td>
<td>&lt;2,500</td>
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Whole-body FDG-PET with CT showed metabolically active 8.6x6.3cm inhomogenous adrenal and 2.2x1.7cm pancreatic tail masses, together with a metabolically inactive 6.3x3.5cm extracapsular low density mass antero-lateral to the right lobe of the liver. The margins of the adrenal mass were irregular and infiltrative although no definite invasion of the IVC or liver was demonstrated. Only the adrenal lesion showed significant uptake of radiolabeled MIBG.

Genetic testing identified a previously described mutation in intron 4 of the MEN1 gene.

Conclusions and points for discussion:
This case describes an unusual de novo presentation of multiple endocrine neoplasia type 1 (MEN1). Phaeochromocytomas have been reported in MEN1 but are very uncommon. The main management dilemma at present is the abdominal surgical strategy. Should the pancreatic tail lesion be resected or observed? Should the subcapsular mass be biopsied or...
resected? If biopsy is preferred, should this be done prior to or at the time of a right adrenalectomy?