Cardiac Paraganglioma associated with SDHB mutation and elevated 3-methoxytyramine level

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Introduction

The succinate dehydrogenase (SDH) protein complex catalyzes the oxidation of succinate. SDHB is one of four protein subunits forming SDH, the others being SDHA, SDHC and SDHD. Germline mutations in SDH proteins can cause familial paraganglioma [1]. SDHB along with SDHC and SDHD are tumour suppressor genes. Paragangliomas caused by mutations in SDHB have a higher incidence of malignancy when compared to those caused by SDHD, which are largely benign [2][3]. Paragangliomas caused by SDHB mutation are usually extra-adrenal.

Background

We report a rare case of a primary cardiac paraganglioma. A 49 year old male was found to have the SDHB mutation, having been screened following his 10 year old niece developing a phaeochromocytoma. He underwent routine biochemical testing which revealed elevated 3-methoxytyramine levels, one of the metabolites of catecholamines.

Clinical findings

The patient demonstrated no hypertension and did not volunteer any symptoms of catecholamine excess such as sweating, headache or tremor, but on direct questioning had experienced palpitations.

Biochemistry:

Biochemistry revealed persistently high serum and urine 3-MT (3xULN), and normal adrenaline and nor-metadrenaline metabolites.

Imaging:

MRI head and neck, ^{123}I- MIBG scans were normal, whilst CT chest showed indeterminate lung nodules. ^{18}F- FDG-PET scan showed an area of high uptake suggestive of a paraganglioma in the thorax.

Cardiac MRI showed a 3.7x2.9cm lesion in the inter-atrial groove extending superiorly between the junction of the right upper pulmonary vein and left atrium, compressing the SVC to the right

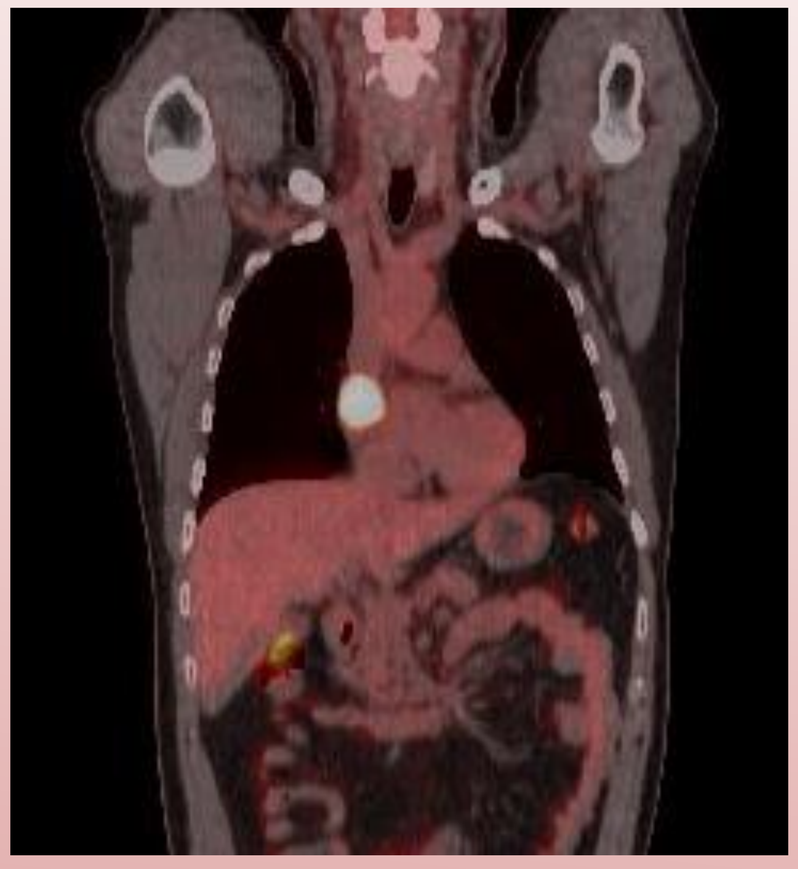
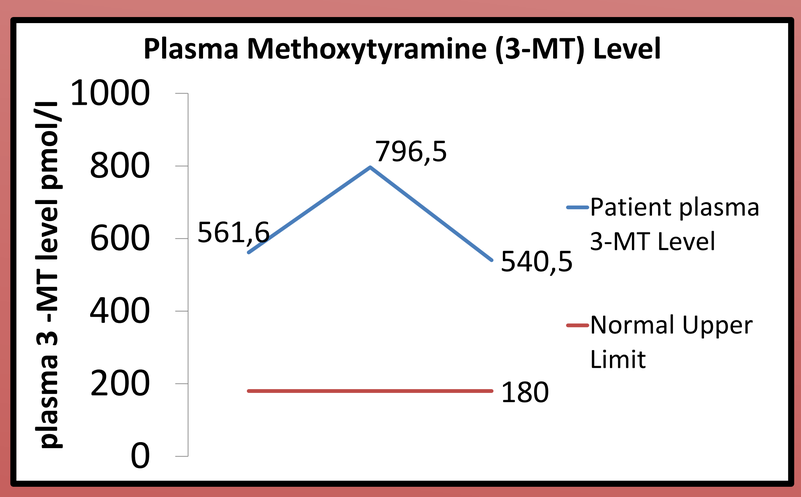


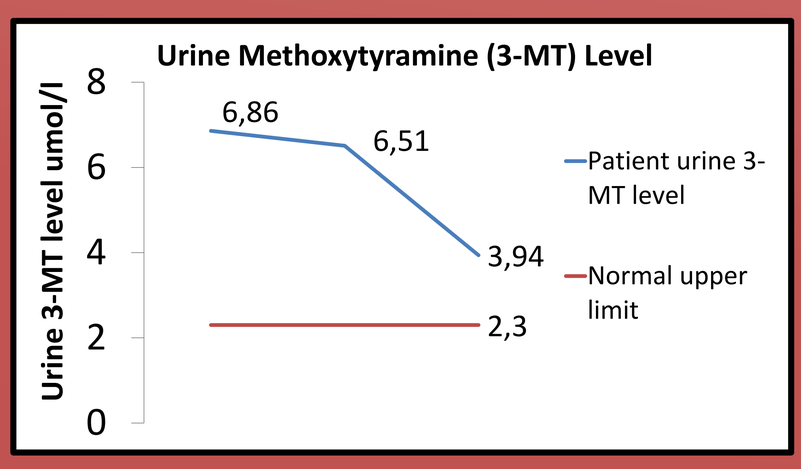
Fig 1: FDG-PET showing intense uptake in intra-atrial groove.

Management

The patient was alpha and beta blockaded in preparation for surgery to protect against significant catecholamine release when anaesthetised or during resection of the tumour.

Before the planned surgical intervention the patient presented as an emergency with chest pain, requiring urgent cardiac bypass surgery for resection of the cardiac tumour and atrial walls, as intraoperative exploration revealed tumour invasion into interatrial septum. Histology confirmed an intra-thoracic cardiac paraganglioma.





Discussion

Paragangliomas almost all produce catecholamines. These catecholamines are metabolized by the tumor within the chromaffin cells to form the ortho-methylated products, which are metanephrine (MN), normetanephrine (NM), and 3-methoxytyramine (3-MT) [4]. Sometimes the tumour secretes only these metabolites, and not the catecholamines making it a more sensitive way to detect these tumours. The catecholamine's and their metabolites are excreted in the urine. The persistently high 3-MT in the absence of high MN or NM in this case is an uncommon biochemical presentation of a paraganglioma.

Paragangliomas in the chest are uncommon, accounting for less than 2% of systemic paragangliomas [5]. Most cardiac tumours are metastasis, fewer then 5% being primary tumours, and of these myxomas are the most common [5]. Primary cardiac paragangliomas are an extremely rare finding. The association between SDH mutations and primary cardiac paraganglioma is reported in the literature, however the anatomical location of interatrial groove, as in this case, is unusual.

This patients ^{123}I- MIBG scan was normal, but this is not entirely surprising as there is evidence in the literature to support that false negative SPECT ^{123}I- MIBG is most commonly found in SDHB-related paraganglioma [6].

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BES2017 EP-057
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Poster presented at: SfE BES 2017 Harrogate
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