CARDIOVASCULAR FLASHLIGHT

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Succinate dehydrogenase gene mutation with cardiac paraganglioma: multimodality imaging and pathological correlation

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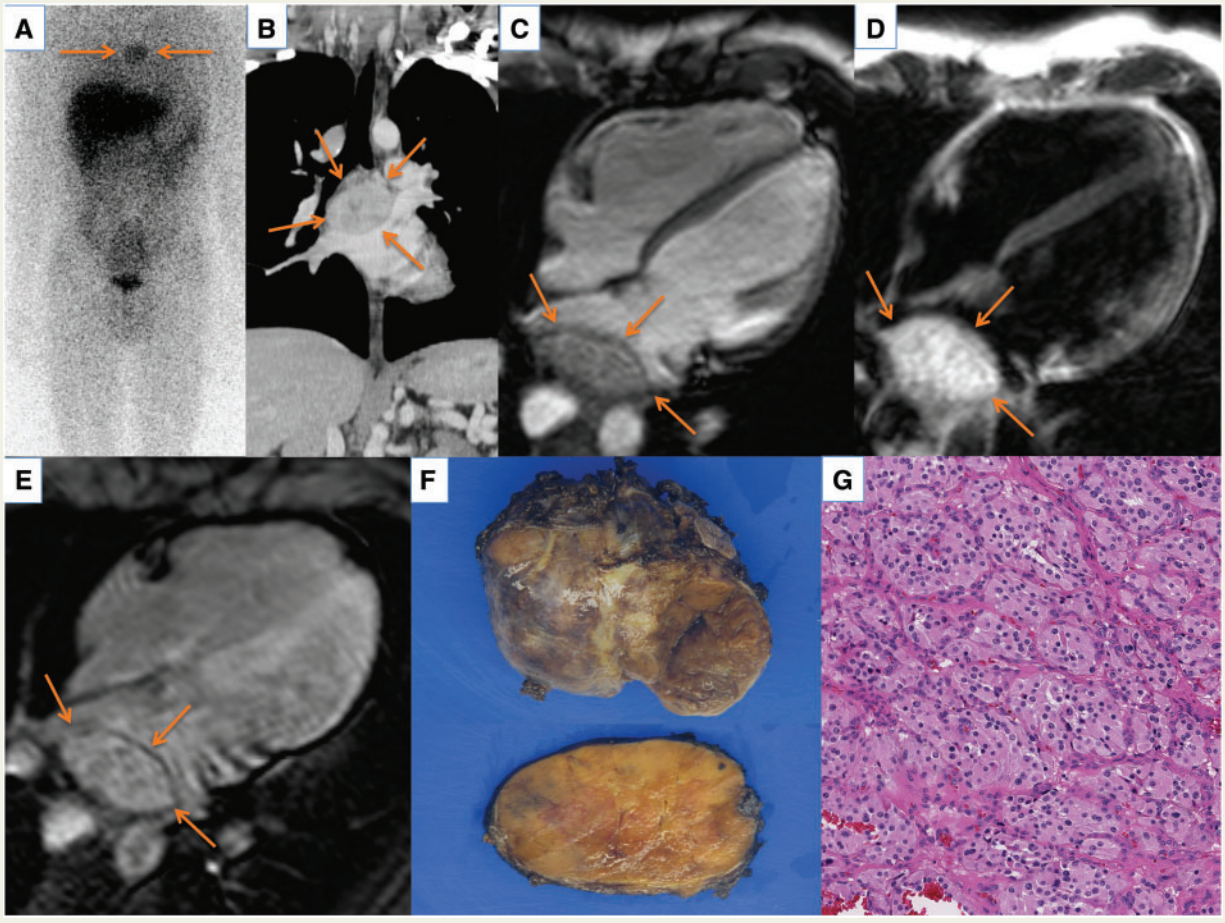
A previously well 28-year-old man presented with several-months of intermittent palpitations, dyspnoea and anxiety. Physical-examination was unremarkable except for a raised blood-pressure of 145/90 mmHg. Plasma and 24-h urinary-catecholamine and metanephrine levels were significantly elevated. Abdominal Computed-Tomography (CT) showed normal adrenal-glands, but total body I-131 metaiodobenzyl-guanidine (MIBG) scintigraphy revealed abnormal focal uptake in the posterior-mediastinum, concerning for a cardiac-paraganglioma (*Panel A*). CT of the chest demonstrated a circumscribed subcarinal mass behind the left atrium (*Panel B*). Echocardiographic images were extremely limited due to poor acoustic windows. Cardiac-magnetic-resonance (CMR) cine-imaging showed a 5.0 x 3.7 cm mass compressing the posterior aspect of the left-atrium (*Panel C*, Supplementary material online, *Video S1*). The mass was hyper-intense on T2-imaging (*Panel D*) and perfused briskly after gadolinium administration (*Panel E*, Supplementary material online, *Video S2*) suggestive of a vascular-tumour. The patient was initiated on alpha-blockade followed by a beta blocker, prior to surgical-excision. An encapsulated 5.1 x 4.2 x 2.2 cm yellow-brown homogenous-mass with focal areas of haemorrhage was removed (*Panel F*). A 'zellballen' pattern was seen on haematoxylin and eosin staining (*Panel G*). Immunohistology was positive for chromogranin-A, synaptophysin, and S-100 immunostains, consistent with cardiac-paraganglioma.

Paragangliomas are extra-adrenal tumours that arise from the autonomic-paraganglia. Pheochromocytomas can be considered intra-adrenal paragangliomas and are indistinguishable at the cellular-level. The majority of paragangliomas appear to be sporadic. However, at least one-third are associated with inherited-syndromes including succinate-dehydrogenase mutations, multiple-endocrine-neoplasia,



neurofibromatosis, and von Hippel-Lindau disease. Genetic-testing in this patient was positive for a known deleterious mutation in succinate-dehydrogenase subunit-B (c.88delC).

Cardiac-paraganglioma are rare and usually located near the left-atrium. Approximately 90% are benign and most are cured by complete excision. There are no reliable histological criteria to differentiate benign from malignant paragangliomas and malignancy is determined by tumour behaviour rather than histological appearance. Diagnosis requires a combination of biochemical and genetic-testing aided by multi-modality-imaging.

Supplementary material is available at *European Heart Journal* online.