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Hereditary Paraganglioma-Pheochromocytoma Syndrome Associated with Renal Cell Carcinoma in a Patient with Succinate Dehydrogenase B Mutation

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Introduction

Succinate dehydrogenase, a mitochondrial enzyme involved in the citric acid cycle, has been implicated not only in hereditary paraganglioma and pheochromocytoma, but in a variety of additional tumors including renal cell carcinoma (RCC), gastrointestinal stromal tumors (GIST), thyroid cancer, and pituitary adenomas.

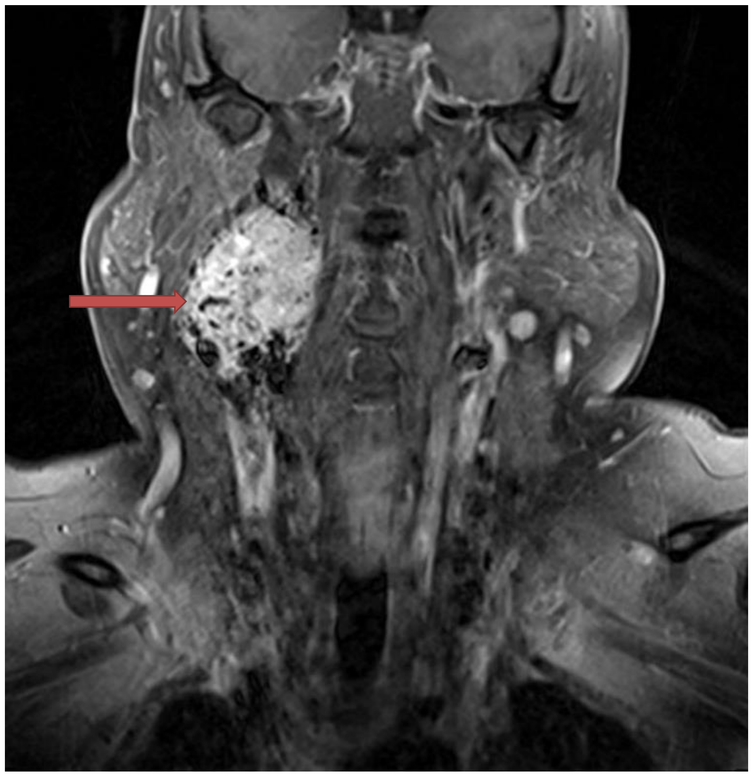

Figure 1: Right carotid body paraganglioma (red arrow)

Case Description

A 51 year old woman presented with a right neck mass. MRI neck displayed a carotid body tumor (Figure 1), and initial biochemical evaluation was most consistent with a non-secretory neck paraganglioma: 24 hour urinary norepinephrine 71 mcg/24 hrs (nl 15-100 mcg/24 hrs), epinephrine less than 2 mcg/24 hrs (nl 2-24 mcg/24 hrs), dopamine 626 mcg/24 hrs (nl 52-480 mcg/24 hrs), metanephrine 73 mcg/24 hrs (nl 90-315 mcg/24 hrs), normetanephrine 420 mcg/24 hrs (nl 122-676 mcg/24 hrs). I123-metaiodobenzylguanidine (MIBG) scintigraphy was without abnormal radiotracer uptake in the chest, abdomen, or pelvis. The patient underwent embolization followed by surgical removal of the carotid body paraganglioma.

Genetic screening was positive for a mutation in the succinate dehydrogenase B subunit (SDHB). CT chest and abdomen obtained for tumor staging noted a 1.4 cm solid exophytic lesion arising from the left kidney (Figure 2). The patient underwent partial nephrectomy and pathology was consistent with a succinate dehydrogenase-deficient renal cell carcinoma. Follow up MR neck and CT abdomen 6 months after surgery showed no evidence of recurrent carotid body paraganglioma or RCC respectively.

Figure 2: Left renal cell carcinoma (red arrow)

Discussion

All patients diagnosed with paraganglioma/ pheochromocytoma should undergo genetic screening. This is particularly important because the presence of a mutation in SDH, von Hippel-Lindau (VHL), NF1, RET, amongst others, could have major clinical implications given the increased risk of malignancies including RCC, GIST, papillary thyroid cancer and neuroendocrine tumors. Understanding the genetic basis of this patient's paraganglioma allowed for early detection and treatment of the coexisting RCC. Identification of a specific mutation will additionally guide future radiologic screening and serial biochemical testing. Identification of a mutation necessitates genetic screening and counseling of other family members, as early diagnosis and treatment are imperative in reducing morbidity and mortality.