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### A Case Of Familial Paraganglionoma Syndrome

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This is a case of a 48-year-old man who was referred from his local hospital with raised urinary catecholamines whilst under investigation for resistant hypertension in 2002. Subsequent investigation revealed a left sided thoracic paraganglionoma, which was successfully resected. He developed an ipsilateral Horner's syndrome but subsequent urinary catecholamine screening was normal.

Initial genetic screening had focussed on excluding MEN 2 as the patient had a subcutaneous lesion on his R forearm that may have represented a neurofibroma. MEN 2 screening was negative and, on the advice of a dermatologist, no further investigation of the skin lesion was undertaken.

The patient had three brothers, one of whom was diagnosed with a renal tumour in 2003 and died in 2004.

Further genetic screening was undertaken of these individuals and both he and his brother had a balanced translocation of chromosome 6, but no evidence of mutation in the VHL gene. His most recent tests show a mutation in the SDHB gene confirming the diagnosis of familial paraganglioma syndrome (and refuting a diagnosis of VHL). His offspring are currently asymptomatic but have been offered genetic counselling and surveillance.

Earlier this year he was noted to be hypertensive and had developed a left sided chest discomfort similar to that which he had prior to his original diagnosis. Elevated urinary catecholamines indicated a recurrence, and while a MIBG scanning failed to localise the culprit lesion, an octreotide scan showed a new focus of abnormal uptake projected over the central mediastinum. He is due back at clinic imminently for discussion of these results.

This case highlights a rare cause of catecholamine excess and the importance of accurate genetic screening to allow tailored counselling and surveillance of the patient and his relatives.

