

A case of Neurofibromatosis type 1 with Pheochromocytoma and Gastrointestinal Stromal Tumour (GIST)

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A 69 year old female with newly diagnosed pheochromocytoma was seen in clinic. Her past medical history included hypertension which was well controlled on Atenolol.

She first presented with abdominal discomfort to the general surgeons in 1990, when an abdominal computed tomography (CT) scan revealed a large incidental right suprarenal mass. Biopsies were deemed difficult due to adherent bowels to the mass and follow-up CT scans suggested a benign stable adenoma. Urinary catecholamines or metanephrines had never been checked.

In June 2006 she re-presented with peritonitis and a laparotomy revealed a thickened wall cyst at the duodeno-jejunal junction. An elective excision of the cyst was performed in April 2007 and during the procedure it was very difficult to achieve haemodynamic stability because of variations in systolic blood pressure of between 50 – 200 mmHg. An unplanned resection of the mass was concomitantly performed.

Histology of the cyst showed a (23 X 20 X 20 mm) malignant Gastrointestinal Stromal Tumour (GIST) and histology of the suprarenal mass confirmed pheochromocytoma (67 X 50 X 25 mm; weighing 80 grams).

She had numerous truncal and upper limb neurofibromata, café-au-lait macules, axillary freckling and relative macrocephaly. Her mother and the eldest of her two daughters had a similar phenotype. One daughter had learning difficulties.

Although unlikely in this age group we considered the possibility of MEN IIB, but genetic screening for RET proto-oncogene was negative. She was normocalcaemic although serum PTH was 5.2 pmol/L (0.5 - 4.4). Thyroid ultrasound showed a multinodular goitre with no dominant nodule.

Although the patient had neurofibromatosis type 1, her pheochromocytoma only came to light 17 years later, during surgical intervention for an abdominal cyst. The coincidence of both gastrointestinal stromal tumours (GIST) and pheochromocytoma in neurofibromatosis type 1 is very rare^[1].

Clinical studies indicate that GISTs are identified in 5 - 25% of patients with type 1 neurofibromatosis^[2-5]. Gastrointestinal symptoms are not uncommon in patients with pheochromocytoma. We recommend that such symptoms in patients with NF-1 may need further investigations to rule out the possible co-existence of GISTs.

References:

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