Case Reports

Bilateral adrenal nodules and pheochromocytoma associated with Neurofibromatosis (NF-1)

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Introduction:

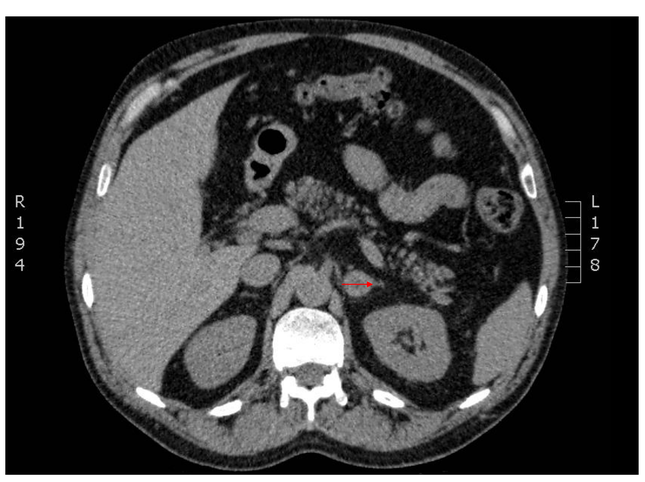
Neurofibromatosis type 1 described by Von Rackling Hausen in 1882, has an autosomal dominant inheritance with complete penetrance, caused by a mutation of the NF1 gene on chromosome 17 (1,2). Patients with Neurofibromatosis type 1 are at increased risk of developing pheochromocytoma. We report two cases of unilateral and bilateral Phaeochromocytoma in patients with NF-1 respectively.

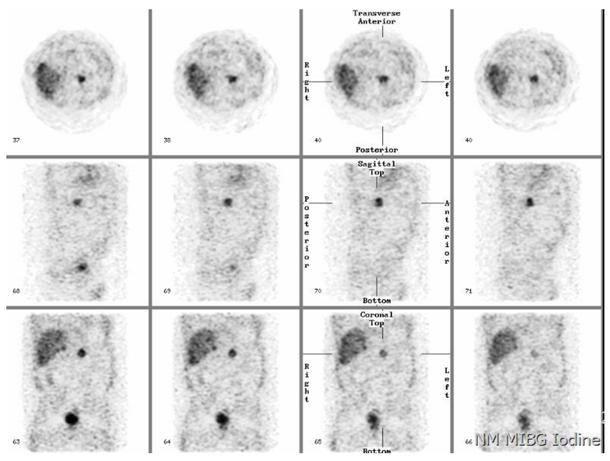
Case 1

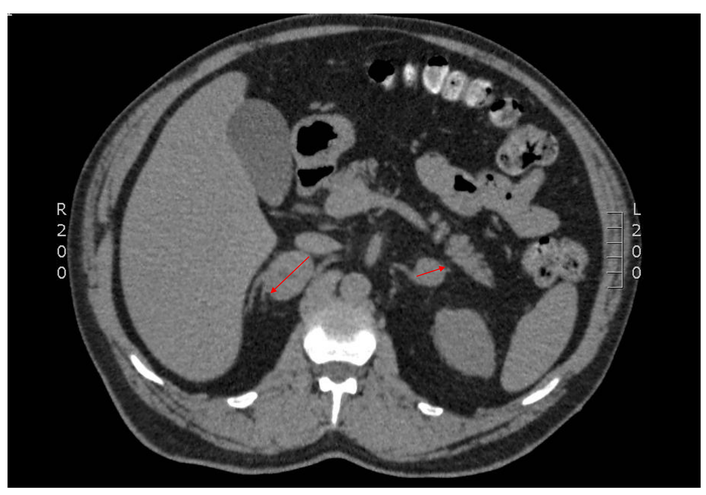
54 male known NF1, hypertensive referred with 32mm incidental left adrenal nodule while being investigated by Gastroenterology for weight loss. 24 hr urinary metanephrines were elevated to 3000nmol/24hrs .MIBG was in keeping with left pheochromocytoma. He was managed perioperatively with phenoxybenzamine and propranolol and underwent left laparoscopic adrenalectomy with good recovery. Post surgery urinary metanephrines were negative with no evidence of recurrence.

Case 2

64 y male, known NF-1 ,hypertensive, depression and osteoarthritis referred with episodic symptoms of palpitation, chest pain and breathlessness. Biochemically confirmed Phaeochromocytoma with 24 hr urinary metanephrines of 18595 nmol/24hrs. CT revealed 4.2cm right adrenal nodule and 2cm left adrenal nodule ,confirmed on MIBG. His BP was optimised with alpha and beta blockade and had bilateral adrenalectomy followed by hydrocortisone and fludrocortisone replacement therapy







Discussion:

The diagnostic criteria of NF -1 developed by National Institutes of Health Consensus Conference in 1987 is widely used in clinical practise [3-5].

Hypertension in patients with NF-1 may be caused by renal artery stenosis, coarctation of the aorta and by pheochromocytoma [6] or it may be "essential". [7]

Phaeochromocytoma has been clinically identified in 0.1 -5.7% of patients with NF-1 and in 20-50% of NF-1 patients with hypertension, compared to 0.1% of all hypertensive individuals with the mean age at the time of diagnosis of 42 years(8).Course of Pheochromocytoma is similar to those of isolated disease

Persons with NF-1 are also at increased risk for malignant conditions, especially malignant peripheral nerve sheath tumor (MPNST), leukemia and rhabdomyosarcoma [9].

Although it is a rare association, screening and careful evaluation of the patients with NF-1 and hypertension must be highlighted as undiagnosed Phaeochromocytoma carries high risk of life threatening cardiovascular complications perioperatively.

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Adrenal and steroids

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