Neurofibromatosis type 1 associated with pheochromocytoma

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Introduction:
Neurofibromatosis 1 (NF1) or von Recklighausen disease, is an autosomal condition caused by heterozygous mutations of the NF1 gene. Patients with NF-1 are at an approximately fourfold higher risk of developing tumors than the general population. Pheochromocytoma may occur in about 1% of these patients.

Case report:
A 24-yr-old woman was admitted to our hospital for further examinations of a 6 cm right adrenal mass, that was incidentally discovered by abdominal ultrasonography during examinations for asthenia and weight-loss and confirmed by abdominal computed tomographic scans and magnetic resonance imaging.

She had multiple café-au-lait spots on the trunk and extremities and skinfold freckling.

Bilateral opthalmic examination revealed no Lisch nodules. Urinary catecholamines were markedly increased. The treatment with β-blockers and alpha-blockers kept the patient asymptomatic. Right suprarenallectomy was successfully preformed and the anatomopathological examination of the surgical sample confirmed the diagnosis of pheochromocytoma.

Discussion:
We report a patient of unilateral pheochromocytoma in the background of neurofibromatosis type 1. NF1 is due to mutation of neurofibromin gene on chromosome 17[1]. NF1 is characterized by cutaneous pigmented abnormalities and neoplastic growth of neural crest-derived cells. In 1910, Suzuki for the first time described the association between pheochromocytoma and type I neurofibromatosis[2]. Pheochromocytoma is seen in about 1% of NF1 patients but the incidence increases to 20–50% in cases of NF1 with hypertension. A high index of suspicion is required for identification of such rare association. Data from observational studies suggest that all patients with NF1 and hypertension should undergo a biochemical testing for pheochromocytoma.[4]

Conclusion:
Our case highlights the role of screening for pheochromocytoma in all patients of neurofibromatosis with deserves attention especially with the presence of hypertension.

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