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Neurofibromatosis Type 1 and Pheochromocytoma: Case Report

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Abstract **Case Report**

Neurofibromatosis type 1 (NF1) is a genetic disorder associated with neurofibromin 1 (NF1) gene mutation, which predisposes for various benign and malignant tumors. Some of these tumors that frequently observed in NF1 are pilocytic astrocytomas, gastrointestinal stromal tumors, pheochromocytomas and juvenile myelomonocytic leukemia. Pheochromocytomas account for 4% of incidentally discovered adrenal tumors and 0.1 to 0.2% of hypertensive patients. Neurofibromatosis type 1 is a rare cause of pheochromocytoma. We discuss through this case and a review of the literature.

Keywords: Neurofibromatosi type1, pheochromocytoma, case report.

OBSERVATION

A 40 years woman was admitted in endocrinology department with history of positive symptomatic triad (headaches, palpitations, sweating attacks) over 20 years.

There was no family history of NF1. She was also presenting in Dermatology department for various skin signs witch neurofibromatosis type 1has been diagnosed one year before. On admission she had multiple neurofibromas over the entire skin, café-au-lait spots on the trunk and extremities and skinfold freckling. Ophthalmic examination revealed multiple Lisch nodules

Urinary catecholamines were markedly increased. Magnetic resonance imaging (MRI) revealed a solid round tumor approximately 16 cm in diameter, located in right adrenal gland.

The pre-operative treatment with a α-blocker (phenoxybenzamine) was performed. Right adrenalectomy was performed.

Anatomopathology revealed pheochromocytoma. Urine catecholamines and their metabolites returned to normal ranges on post-operative day.

DISCUSSION

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen's disease, is a common autosomal dominant genetic disorder resulting from mutation in NF1, with a prevalence of one case in 3000 births irrespective of sex or ethnic origin [1-6].

It is characterized by skin lesions called café-au-lait spots and cutaneous neurofibromas [1-6]. The risk of benign and malignant tumors increases with NF1[7].

Pheochromocytoma is a catecholamine secreting tumor arizing from the chromaffin cells of the sympathoadrenal system, cause of hypertension wich it's curable by resection of the tumor [1, 4, 6, 8]. They are classified as sporadic or familial. Most of the pheochromocytomas are sporadic [4].

Familial predisposition concerns patients with multiple endocrine neoplasia type 2, neurofibromatosis Type 1 (NF-1), von Hippel-Lindau disease and familial carotid body tumors [4].

Pheochromocytoma (PHEO) is a complication occurring in 0.1-5.7% of patients with neurofibromatosis type 1 (NF1) [8, 9]. IT was described in1910 by Suzuki who has recognized the association of pheochromocytoma with NF1 [8].

According to the recommendations, screening is recommended for only symptomatic pheochromocytoma [1, 9].

Gutmann *et al.* have demonstrated that pheochromocytoma is resulting to a lack of neurofibromin expression, which can participate in the pathogenesis of NF1-pheochromocytoma [2] on the other side the absence of NF1 gene expression have been reaveled in patient without NF1 [2] on the other side loss of heterozygosity and neurofibromin have been observed in pheochromocytomas from patients with and without NF1[2, 10].

Surgical removal is the only definitive therapy for pheochromocytoma [2, 4, 6]. It was first described by Sargent in 1914, and replaced, since 1992 by laparoscopic adrenalectomie (laparoscopic transabdominal and the endoscopic retroperitoneal procedure) [6].

Currently laparoscopy is expanded to large,bilateral, and metastatic tumors [6, 8]. The only absolute contraindication to laparoscopic approach is large neoplasic lesions with involvement of the surrounding anatomical structures, in this case conversion can be an opption[4, 6].

CONCLUSION

The current strategy of exploring only symptomatic pheochromocytoma leads to an underestimation of their prevalence; as a result we highlight the importance of routine screenig of all NF1 patients to prevent morbidity and mortality associated with undiagnosed pheochromocytoma.

The authors declared no potential conflicts of interest

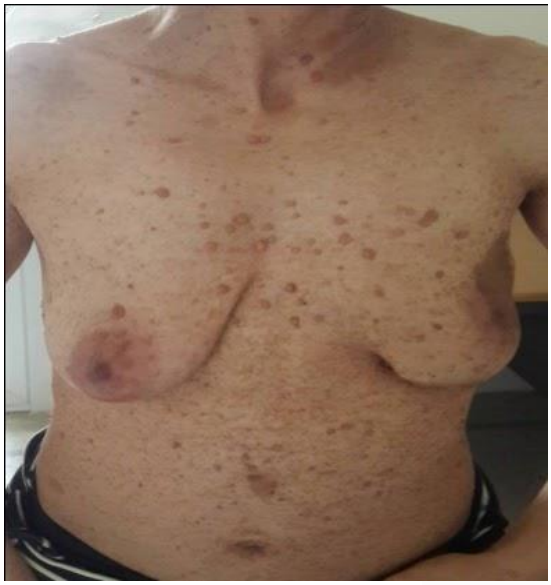

Fig-1: clinical examination out come café-au-lait spots and neurofibroma

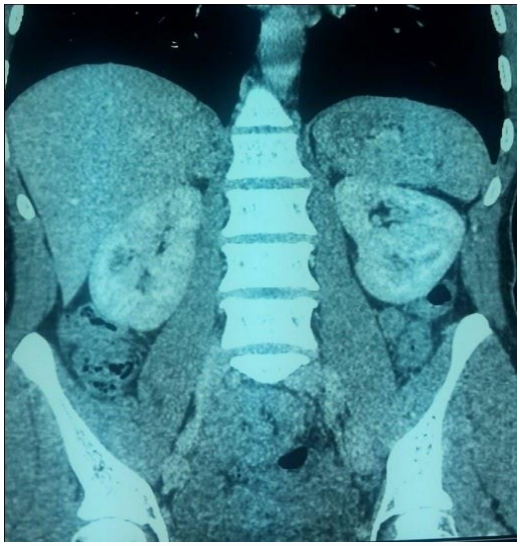

Fig-2: MRI showed right adrenal masse

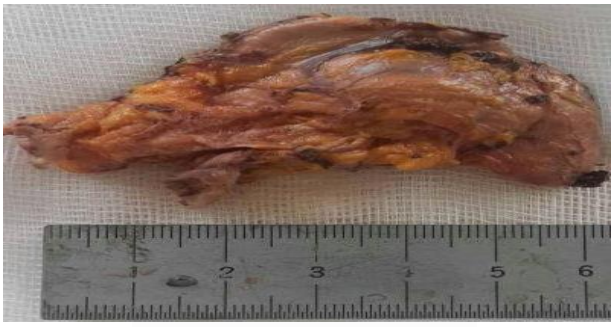

Fig-3: Removal adrenal masse

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