

TYPE I NEUROFIBROMATOSIS WITH PHEOCHROMOCYTOMA

Saikat Datta¹, Sharmistha Bhattacharjee²

¹ Department of Medicine, North Bengal Medical College, Sushrutanagar, Darjeeling, India

² Department Community of Medicine, North Bengal Medical College, Sushrutanagar, Darjeeling, India

Correspondence to: Sharmistha Bhattacharjee (sharmistha.bhattacharjee@gmail.com)

DOI: 10.5455/ijmsph.2013.220420134

Received Date: 26.03.2013

Accepted Date: 22.04.2013

ABSTRACT

Type 1 neurofibromatosis is a common neurocutaneous syndrome with various common and uncommon associations. The present case represents an uncommon association of type 1 neurofibromatosis and pheochromocytoma, which is probably due to mutation of NF-1 gene.

KEY-WORDS: Type 1 Neurofibromatosis; Pheochromocytoma; Mutation; NF-1 Gene

Introduction

The neurofibromatoses are one of the common types of autosomal dominant neurocutaneous disorders which can be divided into neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2) and 'segmental neurofibromatosis'^[1], with each types having their own special characteristics and association with other malignancies. Intracerebral tumours occur in 1.5-8% of cases of NF1.^[2] Other complications include scoliosis and pseudoarthrosis.^[3] The present association of NF1 with pheochromocytoma assumes importance because of the fact that <1 percent of all cases of NF1 is associated with pheochromocytoma.^[4]

Case Report

A thirty five year old female patient presented in ER with features of right sided hemiplegia and a blood pressure of 210/110 mm Hg. CT scan of brain revealed non-haemorrhagic infarction of recent origin in the left MCA territory. She was managed conservatively with antihypertensives and aspirin, and her blood pressure was normalized. But subsequent clinical examinations revealed excessive fluctuations of her blood pressure (fluctuations about 70 mm Hg systolic and 30 mm Hg diastolic). The fluctuations were more on the day abdominal palpations were done. Also, she had multiple firm nodules and rounded hyperpigmented macular spots on her body,

suggesting multiple neurofibromata with Café-au-lait spots (Figure 1).



Figure 1: Multiple Neurofibromata with Café-au-lait Spots



Figure 2: Right Adrenal Gland SOL as seen in CT

All her routine laboratory parameters including serum calcium, renal function tests and thyroid profile were within normal limits. There was no family history of similar skin lesions, she never had any history of tinnitus or deafness, and her developmental milestones were normal. She underwent ultrasound of abdomen which

revealed a round SOL in the right suprarenal region. Non-contrast CT scan of abdomen revealed a large soft tissue density (45 HU) SOL measuring 57mmX67mm in right suprarenal region with areas of necrosis (Figure 2). The SOL was separate from kidney and had extended anterolaterally upon the posterosuperior aspect of right lobe of liver, compressing it but not involving it; inferiorly extended up to the upper pole of right kidney, medially up to the IVC but separate from it. Right adrenal gland was not seen separate from the SOL. A 24 hour urinary VMA estimation showed 47.7 mg/24 hrs (normal reference < 6mg/ day). Hence a diagnosis of Type I neurofibromatosis with right sided pheochromocytoma was made.

Discussion

With an incidence of one in 3000 to 4000 individuals, neurofibromatosis type 1 (NF1) is one of the most common autosomal dominant genetic diseases and very likely the most frequent disorder with increased cancer risk.^[5] Although type 1 neurofibromatosis is an autosomal dominant condition, about 50% of cases are due to new mutations. Its gene is situated on chromosome 17q11.2 (NF-1 gene). Sporadic pheochromocytomas may also occur due to germline mutations in the RET, VHL, NF1, SHDB, SDHC, SDHD, or SDHAF₂ genes.^[6] But association of pheochromocytoma with type 1 neurofibromatosis is reported to be 0.1%-5.7% of all cases of neurofibromatosis.^[7] The above report case probably represents such a rare association due to mutation of NF1 gene.

Conclusion

Type 1 neurofibromatosis is one of the most common phakomatoses encountered worldwide. Although they are characterized by their associations with intracerebral tumours, in specific scenario, other rare associations should be kept in mind, and clinical clues should be always sought for in these circumstances.

References

1. Grant R. Neurocutaneous syndromes. In: Donaghy M, editor. Brain's diseases of the nervous system. 12th ed., New York, Oxford, 2009;3.
2. Huson SM, Compston DAS, Harper PS. A genetic study of von Recklinghausen neurofibromatosis in south-east Wales. II Guidelines for genetic counseling. *J Med Genet*.1989;26:712-21.
3. Akbarnia BA, Gabriel KR, Beckman E, Chalk D. Prevalence of scoliosis in neurofibromatosis. *Spine*.1992;17(8 Suppl):S244-8.
4. Huson SM. Neurofibromatosis 1: a clinical and genetic overview. In: Huson SM & Hughes RAC, Eds., *The Neurofibromatoses: A Pathogenetic and Clinical Overview*. London. Chapman and Hall Medical,1994.
5. Wimmer K. Neurofibromatosis: the most frequent hereditary tumor predisposition syndrome. *Wien Med Wochenschr*. 2005;155(11-12):273-80.
6. Neumann HPH. Pheochromocytoma. In: Longo D, Fauci AS, Braunwald E, editors. *Harrison's principles of Internal Medicine*. 18th ed. Vol 2. New York. McGraw-Hill Professional;2011.
7. Walther MM, Herring J, Enquist E, Keiser HR, Linehan WM. Von Recklinghausen's disease and pheochromocytomas. *J Urol*. 1999;162: 1582-6.

Cite this article as: Datta S, Bhattacharjee S. Type I neurofibromatosis with pheochromocytoma. *Int J Med Sci Public Health* 2013; 2:770-771.

Source of Support: Nil

Conflict of interest: None declared