

## CASE REPORT

## AUTOIMMUNE HYPOTHYROIDISM IN PATIENT WITH NEUROFIBROMATOSIS-1

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A 53 year old woman, diagnosed with Neurofibromatosis I with multiple neurofibromas over trunk, upper limb & lower limb, she had café au lait spots on her abdomen skin. She was admitted to the tertiary care setup with the complains of cold intolerance, numbness in the limbs, high blood pressure & constipation, patient also had complain of weight gain, lab revealed high TSH, low Free thyroid hormones & positive anti thyroglobulin antibodies. Case was diagnosed with autoimmune hypothyroidism. This is the first case reported with such association of these two diseases.

**Keywords:** Neurofibromatosis, café au lait spots, hypothyroidism, Autoimmune hypothyroidism

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## INTRODUCTION

Recklinghausen disease which is another name of neurofibromatosis type-1 (NF 1) is an inherited disorder of autosomal dominant type. It usually has neurofibromas, lisch nodules in eyes, café-au-lait spots and inguinal freckling and difficulty in learning.<sup>1</sup> The prevalence of NF 1 is only 1 in 3500.<sup>1</sup> The mutation responsible for NF 1 gene is located on chromosome 17q11.<sup>2</sup> The NF1 gene encodes for neurofibromin, which acts as a tumor suppressor protein.<sup>3</sup>

Patients who have NF 1 have also shown to develop some other tumours like optic pathway Gliomas, astrocytomas, brainstem gliomas, chronic myeloid leukaemia and rhabdomyosarcoma.<sup>4</sup> NF 1 has also been linked with been linked with thyroid carcinomas in some reports. In one report presence on neurofibroma adjacent to thyroid gland with papillary thyroid carcinoma has also been documented.<sup>4</sup> But the presence of NF 1 along with hypothyroidism which is secondary to lack of dietary intake has not been documented up till now.

This report discusses the possible association of neurofibromatosis with hypothyroidism and possible risk of developing thyroid carcinoma in future.

## CASE REPORT

A 53 year old woman presented to the department of ASH, with the symptoms of weight gain, decreased heat tolerance, decreased sensations in the limbs, on clinical examination patient had diastolic blood pressure of 110 mmHg at three consecutive readings, heart rate was found to be 66/min, neurological examination revealed decreased reflexes in the knee joint of the body, skin revealed yellow discoloration, furthermore patient also had neurofibromatosis type 1, which she developed at the age of 5, family history revealed no such problem regarding skin.

Lab investigation showed TSH was 22.5IU/L & free thyroxine was 50 mg/dl, in addition patient also had anti thyroglobulin positive.



**Figure: Cutaneous manifestations of Neurofibromatosis in the patient**

## DISCUSSION

In 1987, seven cardinal diagnostic criteria for NF1 were established.<sup>5</sup> If any two of the following seven criteria are met, a diagnosis of NF1 is made: (a) two or more neurofibromas on or under the skin or one plexiform neurofibroma, (b) freckling of the groin or the axilla (arm pit), (c) six or more café-au-lait spots measuring 5 mm in the greatest diameter in

prepubescent individuals and over 15 mm in the greatest diameter in post-pubescent individuals, (d) skeletal abnormalities such as sphenoid dysplasia or thinning of the cortex of the long bones of the body, (e) two or more Lisch nodules (hamartomas of the iris), (f) optic glioma, or (g) a first-degree relative with NF1. Similar, NF 1 has no association with the primary hypothyroidism, therefore the primary aim of this case report is to report the quarry of significant association of hypothyroidism with the Neurofibromatosis-1.

## REFERENCES

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