

A Case of Von Recklinghausen's Neurofibromatosis

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A young male presented at our clinic for routine consultation. Multiple skin nodules were observed on the face and hypo pigmented macules on the dorsum of both hands. We examined the patient and skin nodules between 2 and 15 milimeters appeared on his trunk, abdomen, upper limbs, dorsal and lumbar regions. (Panel A and B). Scoliosis of the vertebral spine and several "café au lait" macules along with axillary freckling were present in several body areas. (Panel B). This clinical picture depicts a well known medical⁽²⁾ disorder such as Neurofibromatosis type 1,

a Neurocutaneous syndrome caused by neurogenic tumors arising from neural sheath cells located along peripheral and cranial nerves.^{(3) (1)} Neurofibromatosis type 1, also known as Von Recklinghausen disease, is an inherited autosomal dominant disorder with an incidence of 1 in 3000. NF type 1 is the most prevalent type (90 per cent of all cases) and it is caused by mutations of the NF1 gene located at chromosome 17 (q 11.2) and coding for a tumor suppressor gene, neurofibromin.⁽⁴⁾ These patients should be offered genetic counseling and scheduled for regular follow-ups.⁽⁵⁾



Informed Consent and Conflict of Interest

We obtained a verbal informed consent from the patient for continuing medical education purposes and reflected on his Medical file.

The author declares having no conflict of interest.

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