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Clinical Features of Neurofibromatosis Type 1 (Von Recklinghausen Disease)

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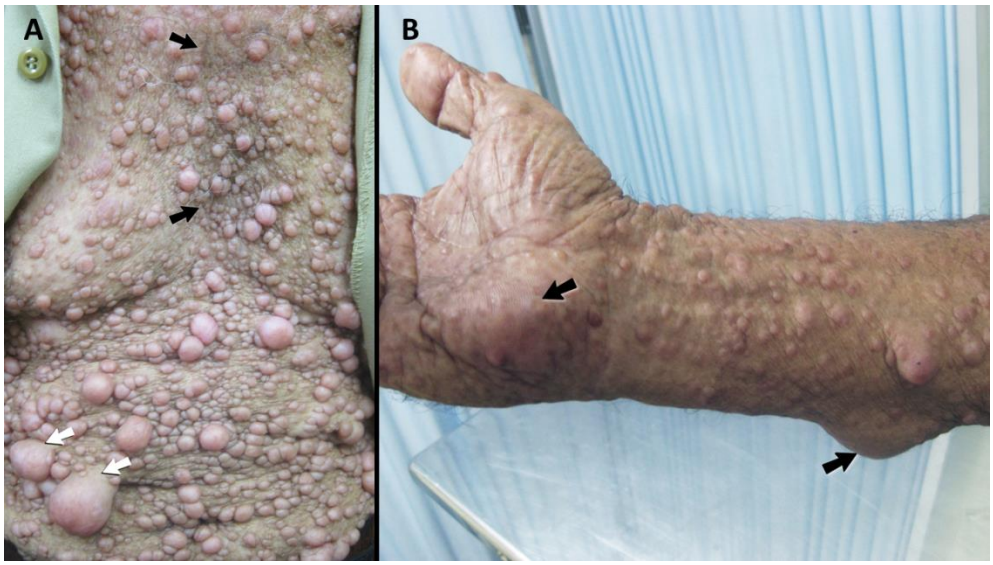
ABSTRACT

Neurofibromatosis type 1 (Von Recklinghausen disease) belongs to a group of genetic disorders, and it is considered one of the most common genodermatosis. This disease negatively interferes with the quality of life and highly influences both emotional and social patients' well-being. In this manuscript, I present a curious case of a patient who has not been adequately managed despite the presence of large cutaneous neurofibromas. This would allow the International Case Reports Journal readers to identify images of advanced stages of neurofibromatosis type 1, so it can capture the visual experience of health professionals to enrich their experience or for educational purposes.

Keywords: Neoplasms; Neurofibromatoses; Neurofibromatosis 1; Von Recklinghausen disease

CLINICAL IMAGE

A 56-year-old male patient diagnosed with hypertension and "cutaneous fibromas" presented with hematemesis, managed properly as acute gastritis, but his underlying diagnosis raised further considerations. On examination, he had multiple nodules (0.5–8 cm) occupying more than 90% of the body surface area. These lesions were judged to be cutaneous neurofibromas, with a pinkish tinge, localized tenderness to pressure, and pedunculated (white arrows, **Figure 1A**). A whorled hyperpigmented macule in the chest appeared as a confluence of "café-au-lait" macules (black arrows). Plexiform neurofibromas were found on his right forearm and hypothenar eminence (black arrows, **Figure 1B**) and skin-fold freckles (Crowe sign) involving both groin areas. The presence of iris hamartoma (Lisch nodules) was not confirmed, but these features support the diagnosis of neurofibromatosis type 1 (Von Recklinghausen disease). Even though disfiguring facial plexiform neurofibromas were absent in this case, the patient claimed these lesions had compromised his quality of life.



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None.

CONFLICTS OF INTEREST

The author has no conflicts of interest to declare.

ETHICAL STATEMENTS

The patient consent form for publication was obtained, and his anonymity was preserved. The author is accountable for all aspects of the work in ensuring its accuracy or integrity.