

Multiple Arm and Trunk Papules

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A 50-year-old female presents with multiple soft papules on her trunk and arms that have been present her entire life. They are generally asymptomatic, though occasionally tender.

What is your diagnosis?

- a. McCune-Albright syndrome
- b. Neurofibromatosis
- c. Tuberous sclerosis
- d. Noonan syndrome
- e. Gardner syndrome

Answer

Neurofibromatosis (**answer b**) is an autosomal dominant syndrome characterized by multiple neurofibromas, café-au-lait macules, and axillary and/or inguinal freckling. Less commonly, plexiform neurofibromas, skin hyperpigmentation, sacral hypertrichosis, and giant, pigmented, hairy nevi are observed.

Lisch nodules (iris hamartomas) are a specific sign of this syndrome. Various bone and endocrine abnormalities have been reported, as well as neurologic sequelae such as mental retardations, epilepsy, and intracranial malignancies.



The diagnosis of neurofibromatosis is based on a constellation of findings. These diagnostic criteria are readily available. Treatment of neurofibromas is primarily by excision. Deaths have been reported from intracranial meningiomas and gliomas, peripheral nerve sarcomas, and other associated malignancies.

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