GENETICS AND GENODERMATOSES

LATE-ONSET OF NEUROFIBROMATOSIS TYPE I (NF 1)

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BACKGROUND: Neurofibromatosis type I (NF1) is an autosomal dominant genetic disorder characterized by the presence of neurofibromas, plexiform neurofibromas, cafe au lait macules, freckling in the axillae and groin, Lisch nodules and bony defects. Dermatologic examination often helps to establish the diagnosis since cutaneous lesions are often the initial clinical features of the disease.

OBSERVATION: A 57-year old man came to Department of dermatovenereology for evaluation of skin changes on his trunk that developed 3 years ago. A skin exam revealed numerous brown and skin coloured nodules with soft consistency that showed a “buttonhole” sign after being gently pressed. Also more than 6 cafe au lait macules were found on his trunk and extremities (diameter varied from 2 to over 10 centimeters). Patient denied visual, auditory or any other neurological disorders. Asked about family history he reported no history of skin lesions but he mentioned neurological condition his son was diagnosed several years ago that he didn’t know much about. A biopsy of one of the lesions was obtained and histological finding was that of a neurofibroma. Compared to his son’s findings which revealed genetically confirmed neurofibromatosis type I, we came upon correct diagnosis of late-onset NF1 in his case.

KEY MESSAGE: Late onset of NF1 is a rare condition. It should be recognized since patients with NF1 have an increased risk of developing a variety of extracutaneous tumors, including optic gliomas, malignant peripheral nerve sheath tumors, pheochromocytomas and CNS tumors. Therefore they should be followed-up for disease progression and possible complications.