GENETICS AND GENODERMATOSES

CIRCUMSCRIBED NODULES IN THE TRUNK: THINK ABOUT SEGMENTAL NEUROFIBROMATOSIS

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Background: Segmental neurofibromatosis (SN), first described in 1931 by Gammel, is a localized form of neurofibromatosis type 1 (NF1). It may be due to somatic mosaïcism resulting from a postzygotic NF1 gene mutation. Despite the fact that NF1 is very common genetic disease, SN is a very rare variant. Herein we report a new case.

Observation: A 78-year-old male, with a history of tremor of extremities, presented with multiple nodules in the abdomen. Physical examination revealed skin colored, fleshy, soft to firm nodules, painless in the pressure, ranging from 1cm to 6cm in size, limited in a circumscribed area in the left flank. They have been growing for several years and he had no similar cases in the family. He didn't have café-au-lait macules. Axillary and groin freckling as well as plexiform neurofibroma was also absent in our patient. Hence, the diagnosis of SN was suspected. A skin biopsy of a nodule was performed revealing a non-encapsulated nodular infiltration of fusiform cells. Tumor cells showed positive staining with S-100 protein. Thus the diagnosis of neurofibromatosis was confirmed. There were neither neurological nor ophthalmic symptoms.

Key messages: SN is an uncommon genodermatosis. The typical clinical feature is the presence of neurofibromas following the lines of Blaschko and less frequently café-au-lait spots. The most affected area is the trunk followed by the face. It can be unilateral or bilateral. In front of circumscribed nodules on the trunk, segmental neurofibromatosis should be suspected. The final diagnosis relies mainly on histopathological examination. The risk of systemic involvement and development of malignant tumors are not well known that's why a regular follow up is recommended.