GENETICS AND GENODERMATOSES

BROOKE–SPIEGLER SYNDROME: A CASE REPORT

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Background: Brooke–Spiegler syndrome (BSS) is a rare genodermatosis, with an autossomal dominant pattern of inheritance, caused by mutations in the CYLD gene. It is characterized by the development of multiple skin appendage tumors, namely spiradenomas, cylindromas and trichoepitheliomas. Although they are typically benign, malignant transformation occurs in 5 to 10% of cases.

Observation: A 43-year-old female patient referred to our department due to multiple skin colored papules and nodules, with a smooth surface, firm and painless, located on the face and scalp, with 12 years of evolution. The patient reported that the mother had similar skin lesions. An excisional biopsy of a skin nodule of her scalp was performed and the histological examination revealed a spiradenoma. Clinical and histological findings combined with family history suggested BSS and a genetic test was conducted, confirming the diagnose. No suspicious lesions were identified and the patient is under follow-up.

Key message: This case highlights the importance of skin lesions as a diagnostic clue for systemic diseases. A prompt diagnosis enables the genetic counseling of the patient and his relatives, through a multidisciplinary approach, allowing the early detection of the underlying malignancies, namely malignant transformation of cutaneous lesions and salivary glands tumors.