GENETICS AND GENODERMATOSES

SEGMENTAL HYPERPIGMENTED PATCH WITHIN SCATTERED CAFE´ AU LAIT MACULES, AN EARLY SIGN FOR SEGMENTAL NEUROFIBROMATOSIS TYPE 1 OR JUST SOLITARY BENIGN PIGMENTATION?

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Background: The presence of café-au-lait macules (CALMs) in a child is a common reason for referral to dermatologists because of its high prevalence. One of the form called segmental CALMs, which usually following the lines of Blaschko, and was limited to a specific body segment. Segmental neurofibromatosis type 1 (NF1; OMIM 162200) is characterized by the segmental distribution of CALMs and (or) neurofibromas. However, pediatric patients only with segmental CALMs were usually underdiagnosed. Segmental CALMs may be solitary benign findings or may indicate the presence of mosaic NF1 with its associated complications. Thus, a correct diagnosis is important for clinical counseling, prenatal diagnosis and precision medicine in the future.

Observation: A 3-years-old girl presented to our outpatient clinics for a large hyperpigmented patch within scattered cafe´ au lait macules over the right subaxillary region since birth. In this study, we performed somatic mutation analysis in 3 cell cultures (melanocytes, keratinocytes and fibroblasts) of skin biopsies from a CALM and a clinically normal skin to investigate the presence of mosaicism, and we found biallelic mutations only in melanocytes, including a total deletion of NF1 gene and a nonsense mutation c.C910T (p.R304X), which confirmed a diagnosis of segmental NF1.

Key message: This study showed that comprehensive mosaic mutation analysis such as identify mosaicism with low frequency mutation in different tissues by deep sequencing can help to make a definite diagnosis and estimate prognosis. And we provide information that segmental hyperpigmented patch within scattered CALMs in pediatric patient since birth is probablely an early sign for segmental NF1.