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A rare case of linear and whorled nevoid hypermelanosis with global developmental delay, scoliosis, and retinal degeneration with dermoscopic features

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Only 40 cases of the rare sporadic Linear and whorled nevoid hypermelanosis (LWNH) are described in medical literature worldwide. It is characterized by hyperpigmented, reticulated, streaky and whorled patches along Blaschko lines, without atrophy or preceding inflammation. It reflects an underlying mosaicism and is occasionally associated with systemic abnormalities.

A 5 year-old female presented in our institution with multiple uniformly hyperpigmented patches, some linear over right upper extremity, both lower extremities and trunk with midline demarcation. Lesions were unchanged and asymptomatic since birth. Dermoscopy showed linear brown streaks with alignment along Blaschko's lines over right upper extremity, reticular over right thigh, and brown structureless zones interrupted by dotted perifollicular hypopigmentation over right posterior trunk and anterior thigh. Histopathology revealed basal layer hyperpigmentation, sparse superficial lymphocytic dermal

infiltrates, melanocytic hypermelanosis and flat-topped papillomatosis. Hematological and biochemical tests revealed no abnormalities. The patient has decompensated thoracic scoliosis with 2-cm divergence from plumb line. Developmental Pediatrics referral revealed assessment of global developmental delay and examination under anesthesia showed peripheral retinal degeneration on both eyes. While this rare occurrence presents a challenging situation and is one reason why data is lacking, affected individuals should be evaluated because of disease associations, including a search for developmental and growth delays, skeletal abnormalities, and other systemic abnormalities. Despite few associations from previous case reports, no promise of chromosomal abnormalities can be found using next generation or whole exome sequencing. Together with the fact that treatments have been tried without much success, makes it a necessity for reporting.

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