Cutaneous and Superficial Soft Tissue Lesions Associated With Albright Hereditary Osteodystrophy: Clinicopathological and Molecular Genetic Study of 4 Cases, Including a Novel Mutation of the GNAS Gene

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Abstract: Albright hereditary osteodystrophy is a rare syndrome, in which cutaneous and superficial soft tissue lesions traditionally include osteomas and calcifications. We report 4 patients from 2 families affected with Albright hereditary osteodystrophy and demonstrate that the spectrum of these cutaneous and soft tissue lesions is broader than is usually defined in the literature. In addition to osteomas in the dermis and subcutis, including so-called plaque-like osteoma, we identified the following lesions: calcifying aponeurotic fibroma-like lesion, calcinosis circumscripta-like lesion, and unusual nevi with osteoid and/or peculiar intranuclear pseudoinclusions. One osteoma and the calcifying aponeurotic fibroma-like lesion were analyzed by HUMARA and proved to be clonal. In a family, a novelmutation in the GNAS genewas also identified.

Key Words: Albright hereditary osteodystrophy, multiple osteoma cutis, calcification, osteonevus, GNAS gene
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