Teaching NeuroImages:

Lipoid proteinosis (Urbach-Wiethe disease)

Typical findings in this rare genodermatosis

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Figure Lipoid proteinosis in a 21-year-old woman with epilepsy and amnesic deficit

(A–C) Symmetrical and bilateral selective horn-shaped uncus calcifications are depicted on multiplanar noncontrast CT and T2/fluid-attenuated inversion recovery images. (D, E) Cutaneous abnormalities were characterized by moniliform blepharosis (beaded eyelid papules) and hyperkeratosis with verrucous lesions on the extensor surfaces of the elbows.

Lipoid proteinosis (LiP) results from a multisystem intracellular deposition of noncollagenous proteins and glycoproteins. This deposition appears to be related to the synthesis and metabolism of collagen and is a result of an autosomal recessive mutation with loss-of-function and reduced or absent expression of the extracellular matrix protein 1 gene (*ECM1*) on chromosome 1q21.¹

Striking mucocutaneous and imaging findings might support diagnosis (figure) in a nonspecific scenario of hoarseness, short stature, migraine, memory deficits, seizures, and psychiatric manifestations.² LiP typically follows a benign, slowly progressive course with a normal lifespan. No effective treatment is currently accepted.^{1,2}

AUTHOR CONTRIBUTIONS

A.J. da Rocha was responsible for interpretation of data and critical revision of the manuscript. M.B. Quirici was responsible for writing the manuscript and interpretation of data.

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