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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Teaching NeuroImages: Lipoid proteinosis (Urbach-Wiethe disease): Typical findings in this rare genodermatosis
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