Lipoid proteinosis with bilateral amygdalae calcifications, headache, and cognitive impairments

Lipoid proteinosis (LP) is a systemic autosomal recessive disorder caused by mutations in the ECM1 (extracellular matrix protein 1) gene and is occasionally associated with cognitive impairment, headache, and temporal lobe epilepsy. A 37-year-old woman with characteristic cutaneous lesions (figure 1), bilateral cataracts, and lens subluxation was evaluated for diffuse cognitive impairment and headache. Neuroimaging revealed bilateral amygdaloid calcifications typical for LP (figure 2). Sequencing the ECM1 gene identified homozygosity for a splice-site mutation, c.195+1G>C in intron 1. This case illustrates the various clinical manifestations of LP (none suggestive of amygdala involvement), which should be considered in the differential diagnosis of cerebral calcifications.

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