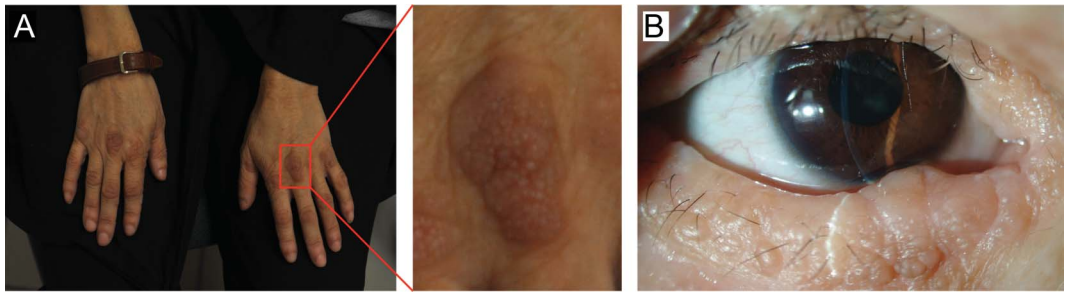
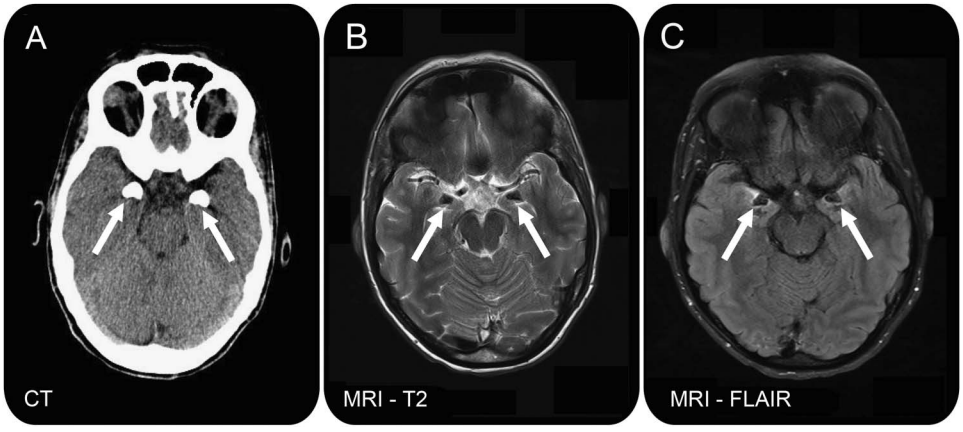
Lipoid proteinosis with bilateral amygdalae calcifications, headache, and cognitive impairments

Figure 1 Dermal manifestations of lipoid proteinosis are caused by intracellular deposition of hyaline material



(A) Warty plaques over the dorsal aspect of the metacarpo-phalangeal joints. (B) Thickening and beading of the eyelid margins (moniliform blepharosis).

Figure 2 Typical neuroimaging findings in lipoid proteinosis



(A) Bilateral bean-shaped calcifications of the amygdalae bordering temporal horns demonstrated by CT. (B, C) Corresponding symmetric and circumscribed T2 hypointensities on MRI. FLAIR = fluid-attenuated inversion recovery.

Lipoid proteinosis (LP) is a systemic autosomal recessive disorder caused by mutations in the *ECM1* (extracellular matrix protein 1) gene[1] and is occasionally associated with cognitive impairment, headache, and temporal lobe epilepsy.[2] 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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