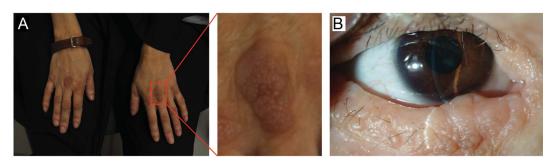
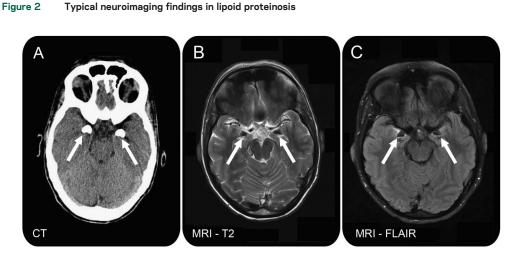
## Lipoid proteinosis with bilateral amygdalae calcifications, headache, and cognitive impairments

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).



(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<sup>1</sup> and is occasionally associated with cognitive impairment, headache, and temporal lobe epilepsy.<sup>2</sup> 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.

David Arkadir, MD, PhD, Israela Lerer, PhD, Laurent Klapholz, MD, Michael Halpert, MD, J.P. Newman, PhD, J. Moshe Gomori, MD, Alexander Lossos, MD

From Hebrew University–Hadassah Medical Center (D.A., I.L., L.K., M.H., J.P.N., J.M.G., A.L.), Jerusalem, Israel; and Columbia University Medical Center (D.A.), New York, NY.

Author contributions: David Arkadir: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, acquisition of data. Israela Lerer: drafting/revising the manuscript, analysis or interpretation of data, acquisition of data. Laurent

Klapholz: drafting/revising the manuscript, study concept or design, analysis or interpretation of data. Michael Halpert: study concept or design. J.P. Newman: drafting/revising the manuscript, analysis or interpretation of data. J. Moshe Gomori: analysis or interpretation of data. Alexander Lossos: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, contribution of vital reagents/tools/patients, acquisition of data, study supervision.

*Study funding*: David Arkadir is supported by a fellowship grant in movement disorders from the Parkinson's Disease Foundation. *Disclosure*: The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures. *Correspondence to Dr. Arkadir: arkadir@gmail.com* 

- 1. Hamada T, Wessagowit V, South AP, et al. Extracellular matrix protein 1 gene (ECM1) mutations in lipoid proteinosis and genotype-phenotype correlation. J Invest Dermatol 2003;120:345–350.
- 2. Goncalves FG, de Melo MB, de L Matos V, Barra FR, Figueroa RE. Amygdalae and striatum calcification in lipoid proteinosis. AJNR Am J Neuroradiol 2010;31:88–90.

304



## Lipoid proteinosis with bilateral amygdalae calcifications, headache, and cognitive impairments David Arkadir, Israela Lerer, Laurent Klapholz, et al. *Neurology* 2013;81;303-304 DOI 10.1212/WNL.0b013e31829bfe1c

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/81/3/303.full
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/81/3/303.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Clinical Neurology http://n.neurology.org/cgi/collection/all_clinical_neurology All Genetics http://n.neurology.org/cgi/collection/all_genetics CT http://n.neurology.org/cgi/collection/ct MRI http://n.neurology.org/cgi/collection/mri
Permissions & Licensing	Information about reproducing this article in parts (figures,tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

## This information is current as of July 15, 2013

*Neurology* ® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2013 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

