Teaching NeuroImages: Gelsolin-related amyloidosis
A rare cause of progressive facial diparesis

A 71-year-old diabetic man of German-Polish heritage, with no relevant family history, presented with 4 years of slowly progressive bilateral facial weakness. Clinical examination demonstrated bilateral, asymmetric facial paresis and skin laxity (figure 1). Screening ophthalmologic examination for diabetic complications revealed bilateral corneal lattice dystrophy (figure 2). EMG showed bilateral facial neuropathies, carpal tunnel syndrome, and mild axonal polyneuropathy. Genetic testing for gelsolin amyloidosis identified heterozygous status for Gelsolin-Asn187 (G654A mutation). Familial gelsolin-related amyloidosis of Finnish type, also known as Meretoja syndrome, is a rare, autosomal dominant cause of progressive facial diparesis associated with cutis laxa and corneal lattice dystrophy.

AUTHOR CONTRIBUTIONS
I. Karakis and H.R. Jones: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, acquisition of data, study supervision. A.A. Gajjar: drafting/revising the manuscript, contribution of vital reagents/tools/patients. D.B. Baharozian: drafting/revising the manuscript, contribution of vital reagents/tools/patients. J. Srinivasan: drafting/revising the manuscript, study concept or design, analysis or interpretation of data.

STUDY FUNDING
No targeted funding reported.

DISCLOSURE
The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

REFERENCES

Corneal lattice dystrophy (arrow) on slit-lamp examination of the right eye due to the lace-like deposition of amyloid within the cornea stroma.

From the Department of Neurology (I.K.), Emory University School of Medicine, Atlanta, GA; Department of Neurology (H.R.J., A.A.G., J.S.), Lahey Clinic, Burlington, MA; and The Family Eye Care Center (D.B.B.), Westford, MA.

© 2013 American Academy of Neurology.
Teaching NeuroImages: Gelsolin-related amyloidosis: A rare cause of progressive facial diparesis
Ioannis Karakis, H. Royden Jones, Ashish A. Gajjar, et al.
Neurology 2013;80;e94
DOI 10.1212/WNL.0b013e318281cc5c

This information is current as of February 25, 2013