Teaching *NeuroImages*: Gelsolin-related amyloidosis

A rare cause of progressive facial diparesis

Ioannis Karakis, MD
H. Royden Jones, MD
Ashish A. Gajjar, MD,
MPH
Dwayne B. Baharozian,
MD
Jayashri Srinivasan,
MBBS, PhD

Correspondence to
Dr. Karakis:
ioannis.karakis@emory.edu

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,¹ is a rare, autosomal dominant cause of progressive facial diparesis associated with cutis laxa and corneal lattice dystrophy.²

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, acquisition of data. 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.

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Figure 1 Prominent facial diparesis and cutis laxa upon eye closure

Note the bilateral lagophthalmos and facial drooping with marked dermatochalasis and superciliary madarosis.

Figure 2 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.

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