Slowly progressive descending facial nerve paralysis indicates hereditary gelsolin amyloidosis

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A 63-year-old woman attended the oculoplastic clinic with a 10-year history of progressive weakness of the muscles of her forehead, and drooping eyebrows and eyelids that were affecting her field of vision (figure). Notably both her mother and brother had similar symptoms. The patient had no other significant medical history.

On examination, she was generally well; she had bilateral brow ptosis, bilateral dermatochalasis, and cutis laxa. The patient had marked weakness of the muscles supplied by the temporal branches of the facial nerve, and to a lesser extent, weakness of those supplied by the zygomatic branches (video). The remaining branches of the facial nerve were relatively spared. Sensation supplied by the ophthalmic branch of the trigeminal nerve was reduced, with sparing of the maxillary and mandibular branches (video). She had no upper eyelid weakness, no anisocoria, and normal extraocular movements. Mild bilateral type 2 corneal lattice dystrophy was seen on slit lamp biomicroscopy (figure), and corneal sensation was reduced. The patient had good corrected visual acuities of 6/9 in the right, and 6/6 in the left eye. We noted no lagophahalmos but a moderate Bell's phenomenon. The patient's clinical signs and family history were consistent with hereditary gelsolin amyloidosis (AGel), and previous genetic testing had confirmed a known amyloidogenic GSN gene mutation: c.640G>A; p.(Asp214Asn) in the patient and her mother.

Conservative bilateral direct brow lift and upper eyelid blepharoplasty was performed, with a marked improvement in the patient's symptoms. At one year follow-up the patient remained happy with her surgical outcome.

Amyloidoses are a group of diseases characterised by deposition of insoluble proteins which aggregate into distinctive fibrillar forms. Hereditary amyloid polyneuropathies are autosomal dominantly inherited and are defined according to the amyloid precursor protein (transthyretin, apolipoprotein A-1, or gelsolin). AGel was first described in Finland, where there are approximately 1000 carriers. The most common manifestations are ophthalmological, with lattice dystrophy, dermatological, with cutis laxa, and neurological, with cranial and peripheral neuropathies; renal amyloid deposits are also common and there may occasionally be cardiac amyloid infiltration. Facial nerve involvement has a unique presentation—it is progressive, bilateral, and starts with the temporal branches, descending to the other branches of the facial nerves over time. Severity ranges from mild involutional changes to complete bilateral facial paralysis. Patients should be counselled appropriately regarding oculoplastic facial surgery. Improved visual function must be balanced against progressive facial and trigeminal neuropathy, and ultimately corneal protection in the long term.

Contributors
All authors provided care for the patient. MK conceptualised the manuscript and wrote the original draft. MK, JG, and TG reviewed, and edited the manuscript. Written consent for publication was obtained from the patient.

Declaration of interests
We declare no competing interests.

Figure: **Slowly progressive descending facial nerve paralysis indicates hereditary gelsolin amyloidosis**
(A) Photograph showing weakness of the muscles of the forehead, and drooping eyebrows and eyelids.
(B) Slit lamp photograph of mild bilateral type 2 corneal lattice dystrophy (arrows)

Multiple-choice question
A 63-year-old woman attended our clinic with a 10-year history of progressive weakness of the muscles of her forehead, and drooping eyebrows and eyelids that were affecting her field of vision. Notably both her mother and brother had similar symptoms. The clinical presentation was consistent with hereditary gelsolin amyloidosis. Which of the following manifestations are not usually present?
A) Corneal lattice dystrophy
B) Cardiac and renal complications
C) Cutis laxa
D) Cognitive and psychiatric disorders

Answer D) using images (A) and (B)