A clue for familial mitochondrial disorder

Discovering a vintage painting

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Figure A vintage painting of the patient's maternal great-grandfather

While there are many examples of such strong family history, the painter highlighted astonishing details of severe ptosis, disconjugate gaze, and facial weakness, similar to the patient's presentation. Reprinted with permission of the family.

A 73-year-old man presented with decades of slowly progressive bilateral ptosis and hearing loss. He also had peripheral polyneuropathy, intermittent binocular diplopia, and mild dysphagia and dysarthria. His mother and maternal grandmother had similar symptoms. He later discovered a vintage painting of his maternal great-grandfather (figure), which showed marked ptosis, disconjugate gaze, and facial weakness. Autosomal and maternal mitochondrial genome sequencing from blood revealed likely pathogenic heterozygous mutations at C10ORF2: c1366C > G (p.Leu456Val). The patient was diagnosed with autosomal dominant progressive external ophthalmoplegia, which can be associated with missense mutation of the *C10orf2* gene encoding mitochondrial DNA helicase.¹

Author contributions

Janice C. Wong: interpretation of data, writing of manuscript, editing of manuscript for critical content. Laura A. Foster: interpretation of data, editing of manuscript for critical content. Reza

Sadjadi: conception of study, interpretation of data, writing of manuscript, editing of manuscript for critical content, patient follow-up.

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Reference

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