Teaching Video NeuroImages: *MT-TL1* mutation presenting as chronic progressive external ophthalmoplegia

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A 19-year-old woman presented with bilateral ptosis and progressive ophthalmoparesis with onset at least 7 years prior. Family history was pertinent for a great aunt with bilateral ptosis. Neurologic examination was positive for bilateral ptosis with near complete ophthalmoplegia (video), otherwise unremarkable. Retinal examination showed no pigmentary changes. Cardiac evaluation and EMG were normal. Orbicularis oculi biopsy confirmed mitochondrial hyperplasia and ragged-red fibers. Blood testing was positive for a pathogenic variant in *MT-TL1* (m.3243A>G), most commonly associated with mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes.[1] Chronic progressive external ophthalmoplegia is a rare though not novel presentation of this mutation.[2]

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Author contributions

A.M. Parsons: drafting the original manuscript and literature review. S.H. Mehta: critical revision of manuscript for content. M.D. Acierno: critical revision of manuscript for content. R. Dhamija: critical revision of manuscript for content and study supervision.

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Disclosure

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