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SHORT COMMUNICATION

Primary open angle glaucoma in a case of mitochondrial encephalomyopathy (Kearns-Sayre syndrome)

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PURPOSE. *Kearns-Sayre syndrome is characterized by chronic progressive external ophthalmoplegia, tapetoretinal degeneration and severe generalized myopathy.*
METHODS AND RESULTS. *We report on a 82-year-old male patient with Keams-Sayre syndrome with open angle glaucoma.*
DISCUSSION. *Reports of primary open angle glaucoma with Keams-Sayre syndrome are very rare, but it is difficult to believe that this association is merely coincidental. (Eur J Ophthalmol 2005; 15: 809-10)*

KEY WORDS. *Kearns-Sayre syndrome, Mitochondrial myopathy, Primary open angle glaucoma*

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INTRODUCTION

Kearns-Sayre syndrome (KSS) is a well-defined clinical entity characterized by chronic progressive external ophthalmoplegia, tapetoretinal-like degeneration, and cardiac conduction defects (1). The syndrome is caused by deletions in mitochondrial DNA (mtDNA). We present a case of definite KSS with a rare coincidence of primary open angle glaucoma (POAG). Reports of POAG in patients with this disease have been previously published only two times.

Case report

In February 2004, an 82-year-old man sought care because of decreased vision in his right eye over the last 6 weeks. His birth and early development were said to be normal and the family history was negative. When he was 18 he noticed reduction of visual acuity of the left eye and he was admitted to the University of Siena Eye Clinic for investigation.

Visual acuity at that time was recorded as 20/20 in the right eye (RE) and counting fingers at 1 foot in the left eye (LE). Ophthalmoscopic examination showed pigmentary changes that involved peripheral retina in both eyes and macular region in the left eye. The patient was treated with streptomycin and then he was discharged home with a diagnosis of disseminated choroiditis.

On presentation, ophthalmologic examination showed visual acuity of 20/20 in RE and counting fingers at 1 foot in the LE. A widely asymmetric ptosis was diagnosed with a complete ptosis in the left eye (Fig. 1). He showed exotropia of 20°. Range of eye movement was restricted in all directions of gaze without abduction. Anterior segment examination was unremarkable. Applanation pressure was 25 mmHg in RE and 26 mmHg in LE. In the RE, G1x-Octopus visual field demonstrated a large arcuate scotoma in the upper hemifield. The perimetric defect type and stage evaluation was performed using the Brusini glaucoma staging system (2) and the grading was Mixed 4. In the LE it was impossible to perform visual

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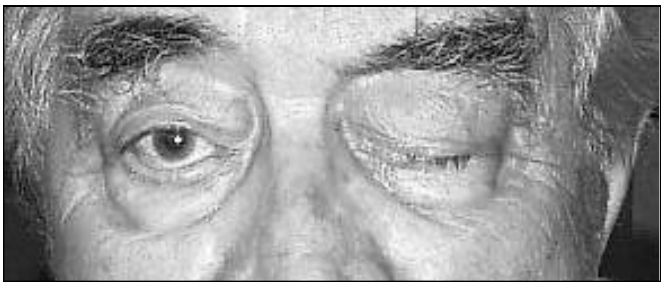

Fig. 1 - *Widely asymmetric ptosis, with a complete ptosis on the left eye.*

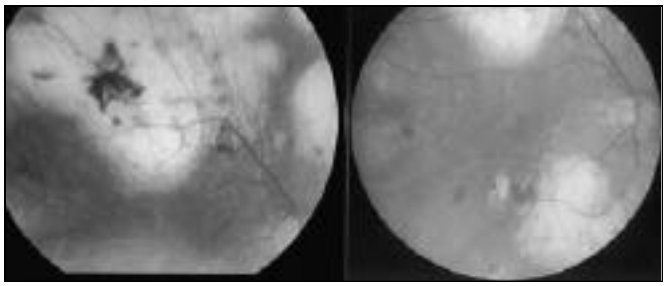

Fig. 2 - *Peripheral fundus shows mottled degeneration of retinal pigment epithelium with minimal pigment clumping.*

field examination because poor visual acuity of this eye. In both eyes the optic discs vertical C/D ratio was 0.9. A regimen of latanoprost in both eyes was begun and IOP decreased to 18 mmHg in both eyes.

Macular region in the LE and peripheral retina in both eyes showed atrophic areas and moderate pigment clumping (Fig. 2).

Electroretinographic findings showed absence of scotopic b wave amplitude. Neurologic examination revealed no abnormalities.

Electromyography of deltoid, bicipitis brachii, rectofemoral, tibialis-anterior, and gluteus maximum muscles showed ubiquitari signs of muscular damage. Muscle biopsy of quadriceps femoris showed not typical ragged red fibers after Gomori trichrome stain and numerous COX negative fibers, suggesting a mitochondrial myopathy.

Light microscopy of quadriceps femoris biopsy stained with Gomori trichrome showed abnormal increase of subsarcolemma.

Electron microscopic examination revealed subsarcolemmal accumulation of mitochondria. Molecular analysis of the mtDNA performed by Southern blot revealed a significant single heteroplasmic deletion. Electrocardiography showed signs of ataxia cordis and bundle right branch block.

DISCUSSION

The most common ocular manifestations of mitochondrial ophthalmopathies reported in the literature include external ophthalmoplegia, pigmentary degeneration of the retina, and corneal decompensation. POAG has been observed rarely. To our knowledge, this is the third report (3, 4) of bilateral POAG associated with KSS. In spite of the rarity of the finding, it is difficult to believe that this association is merely coincidental and the common denominator is presumably the mitochondrial abnormality with a large DNA deletion. Further investigations will be needed to confirm the presence of POAG in patients with mitochondrial encephalomyopathy.

The authors have no proprietary interest in any aspect of the article.

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REFERENCES

1. Kearns TP, Sayre GP. Retinitis pigmentosa, external ophthalmoplegia, and complete heart block. Arch Ophthalmol 1958; 60: 280-9.
2. Brusini P. Clinical use of a new method for visual field damage classification in glaucoma. Eur J Ophthalmol. 1996; 6: 402-7.
3. Bastiaensen LAK, Frenken CWGM, TerLaak HJ, et al. Kearns syndrome: a heterogeneous group of disorders with CPEO, or a nosological entity? Doc Ophthalmol 1982; 52: 207-25.
4. Kalenak JW, Kolker AE. Kearns-Sayre syndrome and primary open angle glaucoma. Am J Ophthalmol 1989; 108: 35-6.