

Chronic Progressive External Ophthalmoplegia a Not Rare Case Report

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Abstract: Chronic progressive external ophthalmoplegia (CPEO) describes an array of hereditary myopathies affecting extraocular muscles (EOMs), commonly manifesting as bilateral ptosis and ophthalmoplegia. It is a chronic, progressive, bilateral, typically symmetric, and external (i.e., spares the pupil) ophthalmoplegia.^[1] It is often the only feature of mitochondrial disease, in which case the term CPEO may be given as the diagnosis. Herein we report a 30 year old lady with progressive ptosis without diplopia and without any similar family history, No history of fatiguability, fluctuations in drooping of eyelids. No H/O consanguinity. Neurological examination showed moderate bilateral ptosis and marked limitation of conjugate gaze in all direction. Serum acetylcholine receptor antibody was negative. Neostigmine test was negative. MRI brain normal. Repetitive nerve stimulation test showed no decremental response both at rest and post exercise. Muscle biopsy was not done, referred to higher Centre for the same. Patient was diagnosed as having a mitochondrial myopathy and given supportive therapy.

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I. Introduction

CPEO is a slowly progressive myopathy primarily involving and often limited to extra ocular muscles. CPEO was described in 1868 by von graefe. men and women are equally affected. Ptosis beginning in childhood and sometimes in adolescence is followed by ophthalmoparesis. Ciliary and iris muscles are not involved. Pattern of inheritance is mainly autosomal dominant, rare recessive or uncertain. DNA mutations like ANT1, POLG2, and PEO1 have been reported.^[2] It is a progressive disorder in which all extra ocular muscles are affected, characterized by bilateral, generalized restriction of eye movements (ophthalmoplegia) and drooping of the upper eyelids.^[3]

II. Case Report

A 30 year old female presented with history of drooping of both eyelids started 12 years back . No history of fatiguability, fluctuations in drooping of eyelids. No H/O diplopia, Drooping of eyelids are slowly progressive. No motor weakness. No H/O consanguinity. No h/o any weakness, breathlessness, weight loss, no family H/O similar complaints, no h/o night blindness and no h/o snakebite. On examination, her vitals were normal, Neurological examination showed moderate bilateral ptosis and marked limitation of conjugate gaze in all direction (FIG 1). Her visual acuity was normal and visual fields were normal. Neither retinal pigmentary degeneration nor optic atrophy was found. Other neurological examination was unremarkable. On ENT examination, there was no evidence of sensori-neural hearing loss. Nystagmus was absent. Pupils were round and regular, equal in size, and promptly reacted to light. On the basis of history and examination, we had kept a differential diagnosis of chronic progressive external ophthalmoplegia, thyroid associated ophthalmopathy and ocular myasthenia gravis. On investigations, her CBC, ESR, RBS and ECG were normal. ECG showed the presence of non-significant Q wave changes. ECHO and MRI were normal. Muscle biopsy was not done, referred to higher centre for the same.



FIGURE 1: Bilateral ptosis with restricted ocular movements in all the positions of gaze

III. Discussion

CPEO is a slowly progressing disease. It may begin at any age and progresses over a period of 5–15 years. The first presenting symptom of ptosis is often unnoticed by the patient until the lids droop to the point of producing a visual field defect. Ophthalmoplegia is usually symmetrical. As such, double vision is sometimes a complaint of these patients. In fact, the progressive ophthalmoplegia is often unnoticed till decreased ocular motility limits peripheral vision. Myopathies are ubiquitous in mitochondrial disorders, and even asymptomatic patients will typically have significant muscle pathology on biopsy. Muscle biopsy is diagnostic. The accumulation of enlarged mitochondria produce a dark red staining of muscle fibres stained with Gomori trichrome stain, called as the “ragged red fibres”^[4]. The levels of cytochrome C oxidase (COX) deficiency in chronic progressive external ophthalmoplegia patients are high.^[4] Mitochondrial DNA, which is transmitted from the mother, encodes proteins that are critical to the respiratory chain required to produce adenosine triphosphate (ATP). Deletions or mutations to segments of mtDNA lead to defective function of oxidative phosphorylation. This may be made evident in highly oxidative tissues like skeletal muscle and heart tissue. However, extra ocular muscles contain a volume of mitochondria that is several times greater than any other muscle group. As such, this results in the preferential ocular symptoms of CPEO. This classical manifestation of mitochondrial diseases either can develop in isolation (or) in association with other neurological features referred to as CPEO-plus. This includes additional signs and symptoms hearing loss, neuropathy, ataxia, Parkinsonism and depression. In most cases CPEO occurs due to a sporadic deletion or duplication within the mitochondrial DNA.^[5]

IV. Conclusion

To conclude, CPEO is a rare mitochondrial myopathy and all the patients with slowly progressive bilateral ptosis and external ophthalmoplegia should be investigated in terms of CPEO. Muscle biopsy would have been conclusive in our patient but was not possible in our set up. Genetic testing is not feasible in our country at present. However, awareness about classical clinical presentation will lead to more data about true prevalence and evolution of therapeutic strategies in our country.

References

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