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Mitochondrial Myopathy with Chronic Progressive External Ophthalmoplegia: A Case Report

Case Report

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Abstract

A 44-year-old male patient presented with progressive ptosis. Mitochondrial Myopathy with Chronic Progressive External Ophthalmoplegia was diagnosed. Case findings along with a discussion of existing literature are presented.

Introduction

Chronic Progressive External Ophthalmoplegia (CPEO) is a slowly progressive disorder affecting the extraocular muscles, which was first described by Von Graefe in 1868. Initially, it was believed to be caused by neuronal degeneration, but later studies have confirmed that it has a myopathic origin. CPEO usually presents with ptosis in childhood or adolescence, followed by ophthalmoparesis, and it does not involve the ciliary and iris muscles. Both males and females are equally affected, and the pattern of inheritance is usually autosomal dominant, although rare recessive or uncertain cases also exist. Some nuclear genes mutation such as POLG1, Twinkle, and ANT1 have been implicated in CPEO. Although some cases of CPEO transmitted in a Mendelian manner are not of mitochondrial origin. Imaging studies usually show thin and symmetrical extraocular muscles. Muscle biopsy remains the definitive test for diagnosis, although Polymerase Chain Reaction (PCR) can also be used to confirm the diagnosis [1].

Case Presentation

A gentleman aged 44 years presented with progressive bilateral symmetrical ptosis since adolescence (Figure 1). The disease was progressive for 15 years and static for the last 5 years. He did not complain of fluctuating symptoms, diplopia, blurring of vision, gait disturbance, seizures or any other muscle group weakness. The patient had never suffered any head/facial trauma and no family member had any similar complaints

Examination revealed bilateral incomplete ptosis. Pupils were

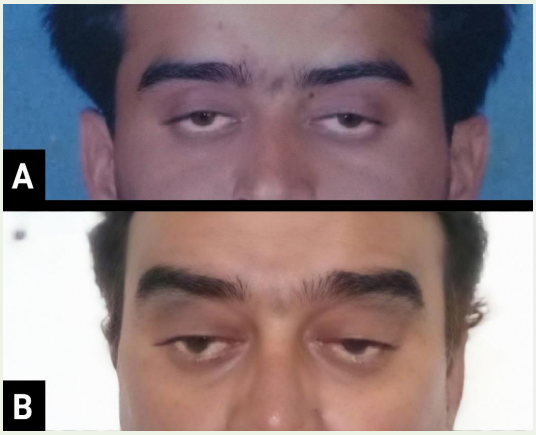

Figure 1

round, equal and reactive to light. Extraocular movements and facial symmetry were preserved. Fundus examination and axial and skeletal neuromuscular examination was unremarkable.

A plain CT scan of the brain was normal. The patient had undergone an MRI brain and bilateral orbits which also did not show any abnormality. Thyroid Function Tests were within normal limits and Anti-Acetylcholine receptor (AChR) antibodies were negative. Nerve conduction studies (NCS) were normal and repeated nerve stimulation (RNS) did not show any decremental response. Serum creatine kinase levels were elevated (682 IU/L). Right deltoid muscle biopsy revealed cytochrome oxidase (COX) deficient fibres and ragged blue fibres on succinate dehydrogenase (SDH) stain. The patient was advised to wear spectacles with eyelid supports.

Discussion

Neuromuscular causes of ptosis were ruled out with the absence of fluctuation of symptoms, negative Anti-AChR antibodies and normal response on RNS. Local causes were ruled on with an MRI brain and orbit. No clinical evidence of the presence of other syndromes like Tolosa-Hunt, Kearns-Sayre or congenital muscular dystrophies (oculopharyngeal or myotonic dystrophy) was found.

Causes of non-progressive ophthalmoparesis are agenesis of extraocular muscles, congenital fibrosis syndrome, and congenital myopathies (centronuclear myopathy, central core myopathy, and multicore myopathy) [1].

The patient presented with ptosis without extraocular muscle weakness, which is an uncommon finding. Most patients present with some amount of gaze abnormalities [2].

Investigations in CPEO reveal elevated creatine kinase levels and minimal extraocular muscle volume loss despite marked weakness [3]. Muscle biopsy shows COX deficient fibres, ragged red fibres on Gomori Trichrome stain and ragged blue fibres on SDH and NADH-TR stain [4]. Electron microscopy of mitochondria shows multiple abnormal mitochondria in the subsarcolemmal region, with paracrystalline inclusion bodies [5]. Electron microscopy and genetic testing could not be performed on our patient.

Mitochondrial myopathies may present as isolated CPEO, CPEO-plus (CPEO with hearing loss, neuropathy, ataxia, parkinsonism, or depression), or Kearns-Sayre syndrome. These presentations may reflect a clinical spectrum rather than discrete illnesses [6].

Treatment includes symptomatic management with eyelid crutches or taping. Anecdotal success without proven benefit has been seen with coenzyme Q10, riboflavin supplementation and ketogenic diet. Candidates for surgery are patients with preserved bell's phenomenon. However, surgery still carries a high risk of exposure keratopathy even in carefully selected patients [4].

Conclusion

Mitochondrial myopathy and CPEO may be suspected in patients who present with ophthalmoparesis with or without other muscle group weaknesses. Progressive external ophthalmoplegia is a striking but nonspecific clinical sign that occurs in a variety of disease states such as myasthenia gravis, thyrotoxicosis, Guillain-Barre syndrome and Refsum's disease. Genetic testing has a large role in establishing diagnosis, prognostication and genetic counselling. Although current treatment options for CPEO are limited, continued investigation is promising.

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