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Chronic Progressive External Ophthalmoplegia (CPEO)

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Abstract : INTRODUCTION: Chronic Progressive External Ophthalmoplegia (CPEO), also known as Progressive External Ophthalmoplegia (PEO), is a type of eye disorder characterized by a loss of the muscle functions involved in eye and eyelid movement. CPEO can be caused by mutations in mitochondrial DNA. It typically manifests in young adults with bilateral and progressive ptosis as the most common presentation but can also present with difficulty swallowing (dysphagia) and general weakness of the skeletal muscles (myopathy), particularly in the neck, arms, or legs. CASE PRESENTATION: This is a case discussion of 3 cousins who presented to our clinic. A 23-year-old male with past surgical history (PSH) of ptosis repair 2 years ago presented with a chief complaint of nasal intonation for 1.5 years associated with difficulty swallowing. The patient also complained of nasal regurgitation of liquids. He denied any headaches, fever, seizures, weakness of arms or legs, urinary complaints or changes in bowel habits. Physical Examination was positive for facial muscle weakness, including an inability to lift eyebrows (Frontalis), inability to close eyes tightly (Orbicularis Oculi), corneal reflex absent bilaterally, difficulty clenching jaw (Masseter muscle), difficulty smiling (Zygomaticus major), inability to elevate upper lip (Zygomaticus minor). Another cousin of the first patient, a 25-year-old male with no past medical history, presented with complaints of nasal intonation for 2 years associated with difficulty swallowing. He denied a history of nasal regurgitation, headaches, fever, seizures, weakness, urinary complaints or changes in bowel habits. Physical Examination showed facial muscle weakness of the Frontalis muscle, Orbicularis Oculi muscle, Masseter Muscle, Zygomaticus Major, Zygomaticus Minor and absent corneal reflexes. A 28-year-old male, a cousin of the first two patients, presented with chief complaints of ptosis and nasal intonation for the last 8 years. He also complained of difficulty swallowing and nasal regurgitation of liquids. His physical examination showed facial muscle weakness, including frontalis muscle (inability to lift eyebrows), Orbicularis Oculi (inability to close eyes tightly), absent corneal reflexes bilaterally, Zygomaticus Major (difficulty smiling), and Zygomaticus Minor (inability to elevate upper lip). MRI brain and visual field of all the patients were normal. Differential diagnoses, including Grave’s disease, Myasthenia Gravis and Glioma, were ruled out. Due to financial reasons, muscle biopsy could not be pursued. Pedigree analysis revealed only males were affected, likely due to maternal inheritance, so the clinical diagnosis of CPEO was made. The patients underwent symptomatic management, including ptosis surgical correction for the third patient. CONCLUSION: Chronic Progressive External Ophthalmoplegia (CPEO), a rare case entity, occurs in young adults as a manifestation of mitochondrial myopathy. There are three modes of transmission- maternal transmission associated with mitochondrial point mutations, autosomal recessive, and autosomal dominant. CPEO can sometimes be difficult to diagnose, especially in asymmetric presentation. Therefore, it is crucial to keep it in differential diagnosis to avoid delay in diagnosis.

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