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A rare case of chronic progressive external ophthalmoplegia in a patient with high myopia

[1]Dr Sonal Gowaikar, Senior Resident, D Y Patil Medical College, Hospital and Research Institute, Kadamwadi, Kolhapur - 413006

[2]Dr Aishwarya Ambre, Junior Resident, D Y Patil Medical College, Hospital and Research Institute, Kadamwadi, Kolhapur -413006

Corresponding Author: Dr Aishwarya Ambre, Junior Resident, D Y Patil Medical College, Hospital and Research Institute, Kadamwadi, Kolhapur -413006

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Abstract

Background: Chronic progressive external ophthalmoplegia (CPEO) is a frequent clinical characteristic of mitochondrial illnesses. It frequently manifests as ptosis and progresses to gradual involvement of the extraocular muscles, as well as varying degrees of oropharyngeal and limb weakness. The genetic underpinnings of CPEO are varied, as they are in many other clinical mitochondrial disorders. The majority of instances are brought on by extensive mt DNA rearrangements, including deletions, duplications, or both. Case report-a thirty-six years old female presented with inability to move eyeballs for two years and drooping of both upper eyelids for six months of gradual onset. On examination OU severe ptosis, external ophthalmoplegia, high myopia and myopic fundus was present. Ice pack test showed OU 3mm of improvement in ptosis. Conclusion: on the basis of clinical evaluation CPEO and myasthenia gravis were the differentialdiagnosis. Neurophysician opinion was sought, a trial of Tab GRAVITOR 60mg half tablet TID was given for 5 days but the patient had no clinical improvement of ptosis, hence myasthenia gravis was ruled out. The genetic analysis was negative for whole genome mitochondria. Due to its difficulty in diagnosis, mitochondrial illness hasn't been the subject of much research.

Keywords: CPEO, myasthenia gravis, chronic progressive ophthalmoplegia, mitochondrial disease, bilateral ophthalmoplegia, ptosis, mtDNA, drooping of upper eyelids.

Introduction

Among patients with mitochondrial disorders, chronic progressive external ophthalmoplegia (CPEO) is a common manifestation. All extraocular musclesare impacted by this progressive illness, which is

characterised by bilateral, global eye movement restriction (external ophthalmoplegia) and drooping of upper eyelids (ptosis).[1]It shows autosomal dominant or recessive inheritance. Mutations in the genes encoding either nuclear DNA (nDNA) or mitochondrial DNA (mtDNA) can contribute to mitochondrial illnesses, a varied category of disorders characterised by primary abnormalities in mitochondrial function (mtDNA)[2]. However, all of these mtDNA mutations are not just linked to the symptom or condition of CPEO but also have the potential to produce additional distinct mitochondrial disorders.[3]

Case report: A thirty-six years old female presented to ophthalmology OPD with complaints of inability to move eyeballs for two years and drooping of both upper eyelid forsix months of gradual onset. Patient was a known case of migraine and hypertension since1 year on regular medications. Patient had no familial history of genetic condition. Patient had history of spectacle use since childhood and had no history of ocular trauma or surgical intervention in the past.

On examination: On inspection patient had chin elevation without head tilt. Forehead showed wrinkles and both the eyebrows were at higher level than the normal. OU upper eyelid showed severe ptosis with OD MRD1 -0, MRD2- 4mm and Palpebral fissure height-4mm and OS MRD1-1mm, MRD2-4mm, palpebral fissure height – 5mm.On levator function test OU the excursion was <4mm. OU anterior segment was within normal limits. Visual acuity (unaided) OD finger counting 3 meters with pinhole 6/12 with (-7.00) DS;(-2.00) DC at 120^0 VA 6/12, OS finger counting 3 meters with pinhole 6/9 with (-5.00) DS; (-2.00) DC at 120^0VA 6/12 was recorded on Snellen's chart. On extra ocular movement examination revealed absent movements in all the gazes. Ice pack test showed OU 3mm of improvement in ptosis Funduscopic examination revealed myopic disc and no pigmentary changes in the peripheral retina. Routine blood investigations were within normal limits.

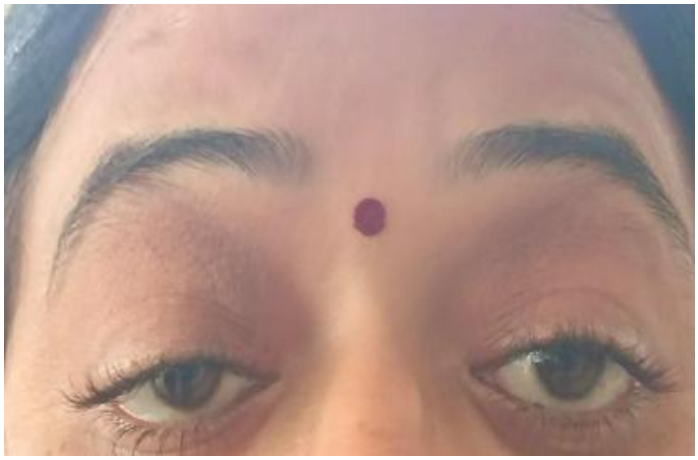
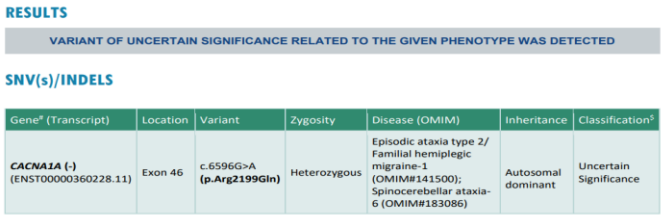


Fig.1: Showing OU severe ptosis and absent movements in all gazes (external ophthalmoplegia).



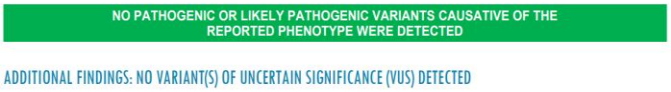
RESULTS

VARIANT OF UNCERTAIN SIGNIFICANCE RELATED TO THE GIVEN PHENOTYPE WAS DETECTED

SNV(s)/INDELS

Gene[4] (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification[5]
***CACNA1A* (-)** (ENST00000360228.11)	Exon 46	c.6596G>A **(p.Arg2199Gln)**	Heterozygous	Episodic ataxia type 2/ Familial hemiplegic migraine-1 (OMIM#141500); Spinocerebellar ataxia-6 (OMIM#183086)	Autosomal dominant	Uncertain Significance

Fig. 2: Result of clinical exome analysis



NO PATHOGENIC OR LIKELY PATHOGENIC VARIANTS CAUSATIVE OF THE REPORTED PHENOTYPE WERE DETECTED

ADDITIONAL FINDINGS: NO VARIANT(S) OF UNCERTAIN SIGNIFICANCE (VUS) DETECTED

Fig. 3: report of whole mitochondrial genome sequencing.

Neurophysician opinion was sought as the differential diagnosis based on clinical examination were CPEO and Myasthenia gravis for which a trial of Tab GRAVITOR 60mg half tablet TID was given for 5 days but the patient had no clinical improvement of ptosis. Genetic analysis of clinical exome tested positive for CACNA1A (-) gene on exon 46, c.6596G>A (p.Arg2199Gln) variant, heterozygous with autosomal dominant of

uncertain significance. Whole mitochondrial genome sequencing was negative.

MRI Brain Plain: Bilateral symmetrical atrophy with thinning of extraocular muscles showing normal signal intensities- Suggest possibility of External Ophthalmoplegia.

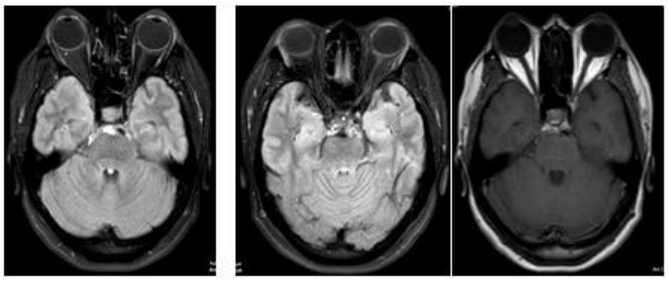


Image 1

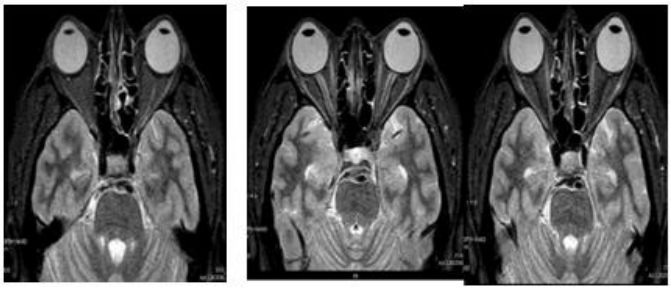


Image 2

Discussion

It is difficult to diagnose and treat mitochondrial illnesses because of the wide variation in their clinical characteristics There have been several accounts of peripheral neuropathy, muscle weakness or atrophy, hypotonia or hypertonia, cerebellar ataxia, and leukodystrophy as presenting symptoms. primary ophthalmological and neuromuscular symptoms such as strabismus, ptosis, external ophthalmoplegia, optic atrophy with progressive vision loss, nystagmus, and retinitis pigmentosa are seen. The following mitochondrial conditions have external ophthalmoplegia: Pearson syndrome, CPEO, and Kearns-Sayre syndrome (KSS). The neuromuscular system is where mitochondrial dysfunction is most frequently first noticed. We report a case of CPEO with high myopia with onset of symptoms at the age of 34 years where as the study shows mean age of onset being 12.8 years[4].

Conclusion

Chronic progressive external ophthalmoplegia as a primary symptom of mitochondrial illnesses may have a better prognosis than anticipated and reported. To generalise the homogeneous aspect of mitochondrial illness, which is heterogeneous, improved classification and additional research are required. There haven't been many researches on mitochondrial disease because it's challenging to diagnose.

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