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# Frustrations of a fixed gaze: An unusual case

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**Abstract**
We take our vision for granted. The smooth coordination of both the eyeballs that gives us a crystal clear vision is a boon. When this mechanism fails, life gets severely disrupted. We present a case here who's been suffering from bilateral drooping of eyelids, diplopia, falls because of visual incoordination and severe depression resulting from above.

**Keywords:** Frustrations, fixed gaze, eyelids, Female housewife

## Introduction
A 41 years old Female housewife, known case of hypothyroidism since 2 years (on Tab Thyronorm 50 mcg) presented with complaints of drooping of both eyelids, diminished vision, sometimes double vision, inability to adduct, abduct, elevate and depress eyeballs since past 4 to 5 years. She also complained of difficulty in moving around and going outside with history of falls due to visual disturbances. Besides above mentioned complaints she had dyspnoea on exertion, generalized weakness, headache, giddiness, loss of appetite, constipation, piles with bleeding PR. Out of frustrations from all these symptoms and no relief in sight, she was plagued with negative thoughts and believed there's no sense in continuing to live. She had seriously considered ending her life.

## On Examination
She was afebrile, vitally stable with BP-110/70, HR-72, SpO2-99% on room air, RBS-122. Pallor was evident. Skin folds on forehead were prominent as a result of chronic contraction of frontalis muscles. She had bilateral drooping of eyelids. She compensated for this by elevating her chin. Cranial nerve examination revealed normal finger counting and normal visual acuity. There was severe restriction of adduction, abduction, elevation and depression of eyeball bilaterally. Occasional diplopia was reported during attempted movements of eyeballs. There was no loss of sensation of smell, taste and hearing, No loss of sensation over face, no difficulty in mastication, no facial asymmetry or deviation of angle of mouth, no nasal intonation, no dysphagia, no dysarthria, no deviation or wasting of tongue. Motor function evaluation revealed no atrophy/hypertrophy. Tone, power and coordination was normal in all limbs and trunk muscles. Sensory function evaluation revealed no loss of superficial deep and cortical sensations. Her superficial deep and visceral reflexes were normal. Cerebellar and autonomic function evaluation revealed no abnormality. Ice pack test-showed no improvement in ptosis after applying ice for 3 minutes. Her single breath count test was normal.
So, clinically, her diagnosis was "Fixed external Opthalmoplegia".

## MRI Brain
No focal abnormality. ***Near symmetric atrophy of bilateral rectus muscles and minimal myositis.***

## NST
There was no evidence of demyelinating sensory neuropathy in both upper and lower limbs and no significant decremental response was noted in the examined muscles.

## Discussion
Chronic progressive external Opthalmoplegia (CPEO) is characterized by slowly progressive paralysis of the extraocular muscles. Patients usually complain of bilateral drooping of eye lids, which is a very irritating experience. There is a constant urge to contract frontalis muscle to counter the ptosis. This is usually followed by inability to move the eyeballs, leaving the patient with a fixed gaze, making day to day life very bothersome. Visual acuity per se isn't affected.

CPEO is the most frequent manifestation of mitochondrial myopathies [1, 2]. there may or may not be a demonstrable skeletal muscle weakness. Moreover, a wide array of permutations and combinations can occur, giving rise to different clinical manifestations.

For instance, these disorders can affect unique anatomic structures such as

1. Eye (Leber hereditary optic neuropathy)
2. Multiple systems, resulting in sensory, ataxia, neuropathy, dysarthria, and ophthalmoplegia [SANDO].
3. Kearns-Sayre syndrome (KSS) is the most dreaded, with poly system involvement. KSS is marked by CPEO with early onset (before age 20) and pigmentary retinopathy. KSS may also have cardiac conduction defects, cerebellar manifestations, mental retardation, Babinski sign, hearing loss [3], seizures, short stature, delayed puberty, and endocrine disorders such as diabetes mellitus, hyperparathyroidism [4].

**Table 1:** Investigations: (abnormalities in bold and italics).

| **CBC** | ***Hb-9.9*** |
|---|---|
| WBC- | 9600 |
| Platelets- | 347000 |
| Creatinine | 0.3 |
| Electrolytes | Na-142, K-4.3, Bun-5.1 |
| Sr Albumin | 4.2 |
| Transferrin saturation | 6.7 |
| ***CPK*** | ***995*** |
| Alkaline Posphatse | 73 |
| T3/T4/TSH | 2.87/1.24/1.88 |
| Acetylcholine receptor Ab | 0.11(Negative) |
| Serum lactate | 1.4 |
| Antimusk Ab: | Normal. |
| ***Serum Iron*** | ***27*** |
| ***Ferritin*** | ***6.94*** |
| Serum Lactate | 1.4 |

**Differential Diagnosis**

1. Neuromuscular junction disorders (myasthenia gravis).
2. Multiple muscle paralysis, restrictive disorders (congenital muscle fibrosis).
3. Thyroid ophthalmopathy.
4. Progressive supranuclear palsy.

The eye movements of CPEO do not show the fluctuation/ variability as that in myasthenia gravis. Tensilon test may be positive in some patients, throwing the clinician off track. It should be noted that the slow saccadic eye movements are an early sign in CPEO. MRI shows marked atrophy of rectus muscles.

Mitochondrial DNA (mt DNA) encodes for essential components of the respiratory chain. Deletions of various lengths of mt DNA, nuclear DNA (n DNA) [5, 6], mt-t RNA mutations [7] and mutations in 12S r RNA and 16S r RNA [8] result in defective mitochondrial function.

This dysfunction is particularly problematic in highly oxidative tissues (Eg, muscle, brain, heart). Extraocular muscles are particularly affected severely because of the rich content of mitochondria in ocular muscles [9, 10].

The protein synthesis is impaired. When stained with Gomori trichrome stain, affected Muscle fibres show an abnormal accumulation of enlarged mitochondria beneath the sarcolemma. (Ragged red fibres).

When there is a gross deletion of mitochondrial DNA, there is a progressive replacement of muscle by fat cells [11].

The distribution and degree of affection of underlying tissues (with deleted mt DNA) will ultimately go on to determine whether the tissue and the resultant organ / system will be affected clinically [12].

**Diagnosis**

Chronic progressive external opthalmoplegia.

**Treatment Plan**

A. (A)Patient education is the most important aspect of management. This lady was counselled in details. She was gently made to accept that this is, unfortunately, a progressive disorder and that there is no known sure fire remedy right now at least. A psychiatric reference might be needed in future.
B. (B)Her Iron, Vit B12 and D3 deficiencies were corrected. This alone would probably go a long way in restoring her sense of wellbeing.
C. (C)She has been recommended "Crutch glasses" for both eyes-a device to reduce her ptosis. Lubricant/ moisturising eye drops have been initiated, which will reduce the soreness/griping sensation in her eyes.

**Conclusion**

This is a case of CPEO presented with bilateral ptosis and severe restriction of extraocular muscle movements resulting in inability to adduct, abduct, elevate or depress both eyeballs following a genetic mitochondrial defect.

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