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A CASE STUDY - UNUSUAL FEATURE OF CHRONIC PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA (CPEO)

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Abstract

A sixty-seven year old man presented with bilateral marked ptosis, gross restriction in elevation, reduced lateral gaze and mild restriction in depression. These ocular movements were associated with an unusual feature of wriggling of the ears on attempted laevo and dextro depression.
Extensive investigation of this patient found mitrochondrial DNA deletion which is associated with ophthalmoplegia, thus the diagnosis of CPEO.
The type of innervational problem seen here may be associated with similar innervational characteristics that occur in cases such as Duane's Syndrome, Marcus Gunn Jaw Winking Syndrome, third nerve palsies and in a case of congenital fibrosis syndrome.
Key Words: *chronic progressive external ophthalmoplegia (CPEO), mitrochondrial DNA, ocular myopathy, wriggling ears.*

CASE STUDY

A 67 year old Indian man presented to clinic complaining of reduced visual acuity in his right eye over the previous twelve months and 'droopy' eyelids. His general health was good and he was not on any medication. He stated there was no family history of eye problems, however both sons had bilateral ptosis one more severely affected than the other (this was noted when the sons presented with their father at an appointment).

The diagnosis of right cataract was made and he underwent a right extracapsular cataract extraction with intraocular lens (IOL) insertion later that year. No comments were made concerning his droopy eyelids.

A year later, he presented to clinic concerned about his 'droopy' eyelids. History taking revealed the following:-

- the ptosis had been present for more than 30 years.
- he had seen a specialist in Bombay who had advised him that his condition was probably congenital.
- he had become aware of the bilateral ptosis during the last 4-5 years.
- he stated that the eyelids often felt heavy and had to be held up.
- the ptosis was worse in the evenings and when tired.
- he was not aware of diplopia, but was aware of impairment of gaze in both the vertical

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and horizontal direction, moreso in elevation.

The treatment options for his ptosis problems were discussed with him. These included: - ptosis sling that attach to glasses frame or surgery involving leavator excision with a bilateral frontalis suspension[4]. Given these options he was happy to continue as he was.

Orthoptic assessment revealed the following:-

Visual Acuity - right & left 6/9

Abnormal head posture - chin up, tilt to right, slight turn to left (see figure 1)

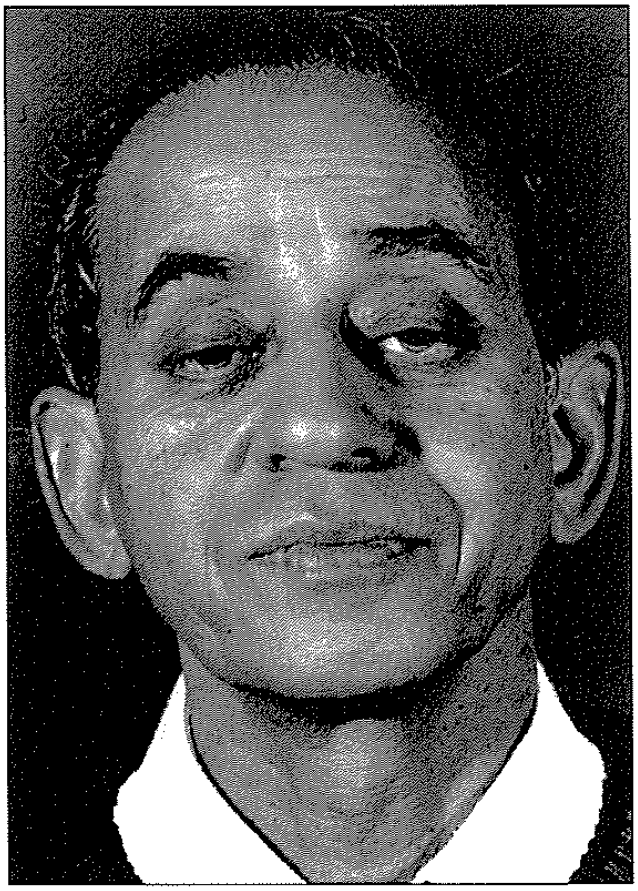

Figure 1.

Bilateral ptosis - poor to fair levator function
R10mm L11mm

Cover test - small left divergent squint with right hypertropia far greater than near

Prism cover test - FR 6-12Δ exo, R/L 4Δ

Visual fields - full to confrontation

Fundi - discs & maculae NAD

Pupillary responses - normal

Ocular movements

- marked restriction in elevation both eyes (no vertical upgaze)

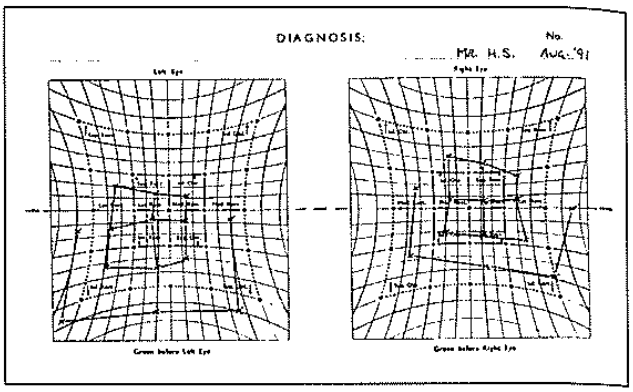

Figure 2.

- reduced lateral gaze R & L
- mild restriction in depression

A Hess chart was plotted (Figure 2.) This showed a restriction in elevation, an exo-deviation and a small right over left deviation.

The above ocular movements were associated with an unusual feature that involved wriggling of the right and left ears on movement of the eyes in dextro-depression and laevo-depression. The ears moved forward and backwards.

It was following these findings that he was referred to the medical clinic for further investigation.

Neurological testing was unremarkable. He was found to be alert and well oriented. All modalities of sensation, motor system and coordination testing were normal. Reflexes and ECG were also normal. An impression of ocular myopathy was the diagnosis made.

Several months later he returned to clinic complaining of horizontal and vertical diplopia. Orthoptic assessment was as the previous examination. The diplopia was overcome by incorporating a 2Δ base in prism in the left lens and a 3Δ base down prism in the right lens of his current glasses.

Following this visit, Mr. H.S. had been symptom-free for up to twelve months. Shortly after this time he presented to the Neurology clinic complaining of 'no energy', tiring very quickly and continual eye muscle weakness. Further extensive investigations were undertaken. Using Polymerase Chain Reaction (PCR) techniques, a molecular defect in mitrochondrial DNA was found and the diagnosis of Chronic Progressive External Ophthalmoplegia (CPEO) was made.

He was commenced on co-enzyme tablets to help his energy problem, but on last follow up was having side effects.

DISCUSSION

CPEO is a slowly progressive muscle disease with onset early in life and characterised by ocular myopathy. It is characterised initially by ptosis and is followed by progressive paralysis of all extraocular muscles. There is no involvement of the intraocular muscles[1].

In the case of Mr. H. S., not only were all these features present, but the additional unusual feature of wriggling ears on eye movements added interest. This unusual feature may be consistent with the innervational type characteristics seen in other ocular syndromes such as Duane's Syndrome[2,3,6,8]. Marcus Gunn Jaw Winking Syndrome[2,4,5,6,9]. Third nerve palsies[2,6,9], and Congenital Fibrosis Syndrome[4,6].

The most common aetiological factors responsible for many of these conditions is anomalous innervation as a result of any of or a combination of the following:

- miswiring of nerve fibres, at the level of the brainstem, during embryonic development[5,7].
- anatomical abnormalities which are either congenital, that is absent nuclei or reduced innervation[2,3,6] or acquired following lesions, injuries or other pathology that cause regeneration of nerve fibres responsible for peripheral anomalous innervation[2,5,6,9].
- normal nerves have dual innervation[2,4,6].

Many of these findings have been confirmed by autopsy examination[3], or by electromyographical studies[2,4,6].

The most likely explanation for the wriggling ears is neural misdirection as a result of abnormal neural sprouting secondary to the neuropathology of the CPEO. This may affect the pathways controlling the eye movements and ears. With the progressive nature of this disease the paralysis of the muscles means a possible loss of innervation allowing for peripheral aberrant innervation to take place. Another possible explanation is that the muscle controlling movement of the ears is the auricularis posterior muscle controlled by the facial (VIIth)

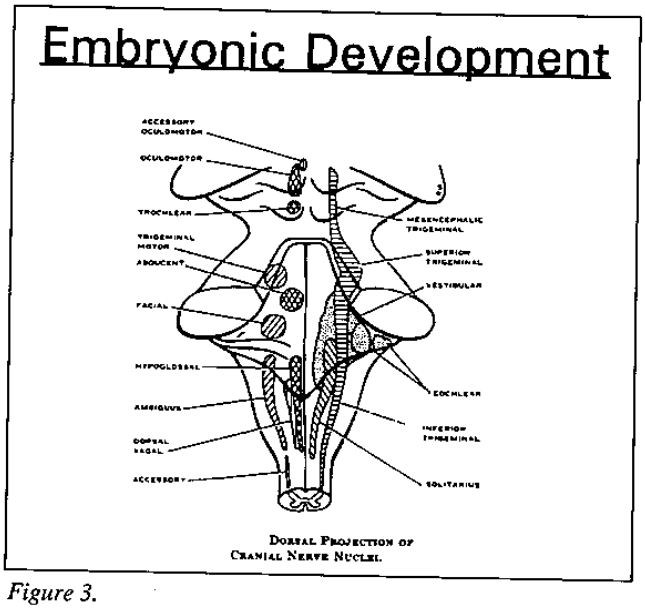

Figure 3.

nerve. During embryonic development neurones making up the VIIth nucleus and VIth necleus develop together and are in close proximity to each other (Figure 3). It is therefore possible that connections at this level persist and would at least explain the activity of the auricularis posterior muscle and activity of the lateral rectus muscle occurring simultaneously[7]. Lateral gaze and depression were least affected. Absence of innervation in elevation may have lead to nerves regenerating and activating other nerves, for example the ear.

CONCLUSION

The recent testing procedure of PCR testing to screen blood samples has been very advantageous in the diagnosis of many diseases. Early diagnosis of CPEO is most important prior to commencement of any form of treatment because of the progressive nature of the disease.

In this patient the wriggling ears was probably an acquired feature secondary to CPEO.

We, as clinicians, when confronted with neuromuscular diseases like this case should in future, not only observe ocular movements but possibly ear movements as well.

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