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CASE REPORT

Congenital Idiopathic Familial Ptosis

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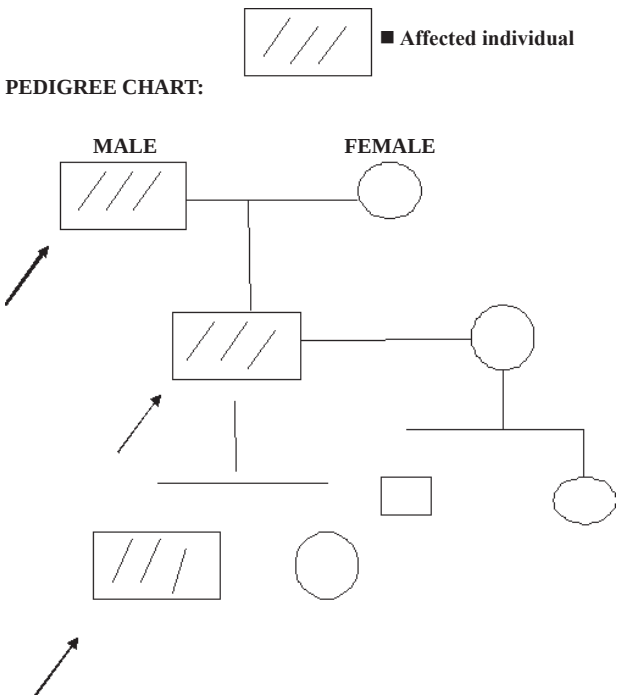
ABSTRACT

Familial congenital ptosis is a kind of simple congenital ptosis, which is seen in the families. It occurs through autosomal dominant inheritance. Common familial occurrences suggest that genetic or chromosomal defects are likely. We have seen such a familial occurrences in our institute.

KEY WORDS: Congenital Ptosis, Familial Ptosis, Dysgenic Levator Palpebrae.

Case Report

A 57 year old male patient came to the surgical opd with an ulcer on the foot and incidentally noticed with unilateral drooping of left eyelid. On taking detailed history, he stated that was present from his birth. It is non progressive and did not affect his vision. His family history was quite interesting, on further history taking revealed that his father also had drooping of both eyelids, and so did the patient's son. No muscular weakness no other neurological symptoms. Neither history of snake bite nor botulinum toxin injected [1] .On examination no other systemic and ocular findings are present. No features of myasthenia gravis [2]. No known etiology can be found except the familial in nature for the three generations.



There was no deformity seen externally with orbit or the surrounding anatomical structures [3]. Pupil is reactive to light and patient does not have divergent squint so that

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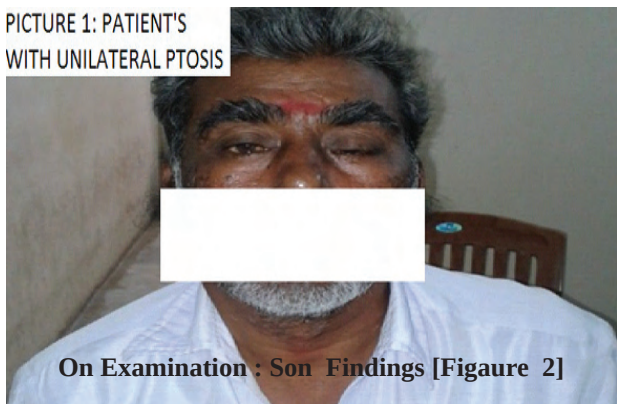
3rd nerve palsy is ruled out, Patient doesn't have epicanthus inversus n telecanthus [4], anhidrosis, enopthalmos nor myosis, lid elevation does not occur with mastication or with movement of jaw to the opposite side. No thickening of eyelids. Patient does not have polychromatic cataract, gonadal atrophy and premature thinning of hair and no progressive muscle weakness.

On Examination :Father Findings [Figaure 1]

FEATURES	RIGHT EYE	LEFT EYE
Forehead	Normal	Normal
Eye brow	Normal	Normal
Creases	+	-
Palpebral fissure	6 mm	2 mm
Vision	Normal	Normal



On Examination : Son Findings [Figaure 2]

FEATURES	RIGHT EYE	LEFT EYE
Forehead	Normal	Normal
Eye brow	Normal	Normal
Creases	+	-
Palpebral fissure	8 mm	4 mm
Vision	Normal	Normal

Differential Diagnosis

Blepharophimosis syndrome,Marcus Gun-Jaw-Winking syndrome,3rd cranial nerve palsy, Horners syndrome5, Myasthenia gravis,Myotonis dystrophy, Blepharochalasis

Discussion

Most cases seen are isolated eyelid malposition. No other ocular & other system associated with this. It can be unilateral or bilateral and noticed after birth which is a non progressive condition of the eye with persistent ptosis, (i.e.) not altered with eye movement or innervations of other cranial nerves. Some times familial element is present, but a gene responsible for congenital ptosis has yet to be identified.

Clinical feature of isolated congenital ptosis is poor levator function. Etiology of this dysfunction is not clear. Studies are done to find out whether it is due to primary muscular problem or primary neurogenic problem.

Failure to properly innervate the developing levator palpebrae leads to poor development of muscle. Outcome of this developmental insult is the creation of dysgenic levator palpebrae muscle in which the normal striated muscle fibers are replaced with fatty and fibrous tissue, especially in the anterior portion of the muscle.

This dysgenic muscle does not contract and relax normally. The clinical consequence is poor eyelid elevation on up gaze & eyelid lag on down gaze. All can be corrected surgically and results are seen in 75 % of patients [6].

The differential diagnoses of congenital ptosis are ruled out by the following: Patient does not have epicanthus inversus n telecanthus so it is not blepharophimosis. pupil is reactive to light and patient does not have divergent squint so that 3rd nerve palsy is ruled out. anhidrosis, enopthalmos, myosis is not present exclude the Horner's syndrome. lid elevation does not

occur with mastication or with movement of jaw to the opposite side so that marcus gunn jaw winking syndrome is ruled out. no thickening of eyelids so that blepharochalasis is ruled out. patient does not have polychromatic cataract, gonadal atrophy and premature thinning of hair and no progressive muscle weakness is seen so that myotonic dystrophy not seen in our patient.

Conclusion

This case report highlights the presence of the familial pattern with congenital ptosis. Which needs further evaluation with the genetic study to find out the genetic anomaly present in these patients. The association still is not isolated hence further study is required to find the chromosome responsible for the occurrence and hence can it be prevented before its manifestation. These patients are planned for a genetic study whose reports are yet to be available which will add more clue to the diagnosis the cause and effect of the disease and to improve the management of the patient. Is it limited to the ocular system or any other part is involved in the human system which is not manifested along the ptosis need more research.

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