

## Retinitis pigmentosa associated with blepharophimosis, blue dot cataract and primary inferior oblique overaction: A new syndrome complex?

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A 15-year-old girl with retinitis pigmentosa, blepharophimosis, blue dot cataract and primary overaction of inferior oblique muscle in both the eyes is being reported. Computerized search using Medline did not reveal any such previously reported association.

**Key words:** Blepharophimosis, blue dot cataract, electroretinography, inferior oblique overaction, retinitis pigmentosa.

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Retinitis pigmentosa (RP) are a group of inherited disorders of the retina that are characterized by progressive dysfunction involving photoreceptors leading to eventual atrophy of several retinal layers. Blepharophimosis is a congenital disorder of ocular tissues characterized by shortening of horizontal palpebral fissure, ptosis, telecanthus, epicanthus inversus and certain other associated features.<sup>1</sup> Blue dot cataracts are bilateral, largely stationary, usually visually insignificant cataracts, with an early onset.<sup>2</sup> We report a patient with RP associated with blepharophimosis, blue dot cataract and primary overaction of inferior oblique muscle in both the eyes. We could not come across any such association of retinitis pigmentosa in previous published reports (computerized search using Medline).

### Case Report

A 15-year-old girl presented to us with complaints of defective vision that was more pronounced at night time. The systemic and family history were noncontributory.

The best corrected visual acuity was 20/40, N<sub>8</sub> in both the eyes with the retinoscopy showing a myopic refraction. Anterior segment examination revealed telecanthus with an inner intercanthal distance of 35 mm and an interpupillary distance of 54 mm. There was mild blepharophimosis. The horizontal fissure width was 26 mm and 28 mm in the right and left eyes respectively. The vertical fissure width was 8 mm and 8.5 mm in the right and left eyes respectively. There

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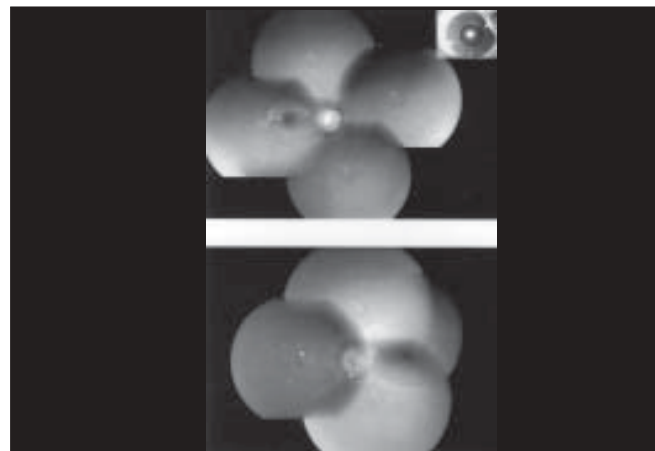
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was mild ptosis, euryblepharon and epicanthus inversus. Based on the above findings, a diagnosis of blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) Type I of sporadic inheritance was made.

Alternate cover test revealed a small exophoria in primary position. Ocular motility testing revealed a primary overaction of inferior oblique with a V pattern [Fig. 1]. A dilated slit-lamp examination revealed the presence of blue dot cataract in both the eyes [Fig. 2A, inset]. Fundus examination revealed disc pallor and attenuated vessels [Figs. 2 A, B]. However, there was no bone corpuscular pigmentation. An electroretinogram (ERG) was done which showed totally attenuated responses for both scotopic and



**Figure 1:** Photographs showing primary inferior oblique overaction in both the eyes. Center photograph shows the blepharophimosis, epicanthus inversus and telecanthus and euryblepharon. Topmost photograph left side: Note the upshooting of the right eye on levelevation, suggestive of right inferior oblique overaction. Topmost photograph right side: upshooting of the left eye on dextrolevation suggestive of left inferior oblique overaction. Center up, center right, down right, center down, down left and center left denote elevation, dextroversion, dextrodepression, depression, levodepression and levoversion respectively



**Figure 2:** (A) Red-free montage of the fundus of the right eye showing markedly attenuated vessels. Inset is the slit-lamp photograph of the anterior segment showing multiple opacities within the substance of the lens suggestive of blue-dot cataract, (B) Red-free montage of the fundus of the left eye showing markedly attenuated vessels

photopic stimuli [Fig. 3]. Visual fields showed a peripheral constriction of the central fields. Based on the above findings, a diagnosis of RP associated with BPES Type I, blue dot cataract and primary inferior oblique overaction in both the eyes was made. Systemic examination failed to reveal any other abnormality. The patient refused to undergo cytogenetic studies and surgical correction for the blepharophimosis. The patient was referred to the visual rehabilitation center for possible visual aids to improve the near vision.

## Discussion

Retinitis pigmentosa has been described with facial<sup>3</sup> and eyelid anomalies including unilateral upper eyelid ptosis and enophthalmos<sup>4</sup> previously but not with BPES. Alternatively, ocular defects associated with congenital blepharophimosis include strabismus, nystagmus, amblyopia, microphthalmus, anophthalmus, ptosis, epicanthus inversus, microcornea and hypermetropia.<sup>5</sup> Our patient had a hitherto unreported association of primary inferior oblique overaction and an associated V pattern.

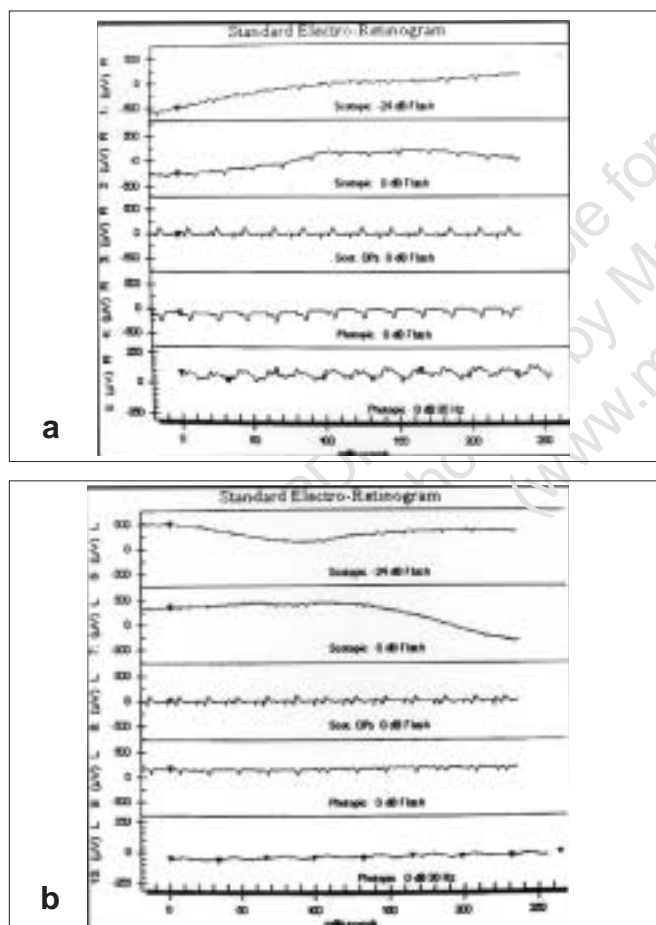
Macular heterotopia has been reported in association with blepharophimosis.<sup>6</sup> The fovea in either eye in our patient was normal in position. The blepharophimosis-ptosis-epicanthus inversus syndrome has been described with congenital cataract with acquired somato-facial dysmorphism.<sup>7</sup> Our patient had blue dot cataracts in both the eyes with no other systemic anomalies.

The blepharophimosis-ptosis-epicanthus inversus syndrome has been localized to Chromosomes 3 and 7.<sup>1</sup> Interestingly, both RP and BPES have been reported in association with abnormalities centered around chromosome 14. Subtelomeric deletion of the long arm of chromosome 14 in a case of blepharophimosis has been reported previously.<sup>8</sup> Retinitis pigmentosa has been reported to occur along with ring chromosome 14 cases, in all probability due to mutations of the neural retina leucine zipper (NRL) gene located at 14q11.1-q11.2.<sup>8</sup> However, occurrence of both these phenotypes at a single genotypic location has not been reported so far. It is quite possible that a chromosomal anomaly probably in the region of chromosome 14 would have been discovered in our patient, had she been willing to undergo the cytogenetic analysis.

We are reporting this case for the rare association of RP with BPES, blue dot cataract and primary inferior oblique overaction. Although this could be an unusual chance association, the possibility of a new syndrome complex cannot be ruled out. Further cytogenetic studies, especially in the region of chromosome 14 are required in cases of RP and BPES.

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**Figure 3:** (a and b) Electroretinograph of the right (top) and left (bottom) eyes of the patient showing extinguished scotopic and photopic responses