



Blepharophimosis Syndrome with Retinitis Pigmentosa (RP) A Rare Syndrome Complex

Authors

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Abstract

Blepharophimosis is a congenital condition where the patient has severe bilateral, symmetric ptosis, telecanthus (an abnormally wide intracanthal distance with normal interpupillary distance), epicanthus inversus (skin fold arising from the lower eyelid that covers the medial canthus) and blepharophimosis (profound narrowing of the palpebral fissure).

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. We report a patient with RP associated with blepharophimosis. We could find only one case of such features reported earlier.

Case Report

Reporting a case of 5 yr old boy of blepharophimosis syndrome who had undergone lid reconstructive surgery (fig-1). There was no significant family history.

The patient complained of diminished vision which was more in night time. His BCVA was 6/18 in right eye and 6/24 in left eye and n8 in both eyes. The patient had myopic astigmatism in both eyes. Anterior segment examination revealed telecanthus-intercanthal distance 36mm.

Horizontal fissure length-24mm and 22mm in right and left eyes, Vertical fissure length—7.5mm and 7mm in right and left eyes respectively mild ptosis with epicanthus inversus. Based on the above findings, a diagnosis of blepharophimosis-ptosis-epicanthus inversus syndrome (BPES type 1) was made. Fundus examination revealed disc pallor and attenuated

vessels (fig- 2.) However, there was no bone corpuscular pigmentation. An electroretinogram (ERG) was done which showed totally attenuated responses for both scotopic and photopic stimuli. Systemic examination failed to reveal any other abnormality. Patient refused cytogenetic studies due to financial reasons.

Discussion

Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome (BPES) first described by Komoto in 1921, is a dominantly inherited disorder characterized by four features that are present at birth severe bilateral, symmetric ptosis, telecanthus (an abnormally wide intracanthal distance with normal interpupillary distance) epicanthus inversus (skin fold arising from the lower eyelid that covers the medial canthus), and blepharophimosis (profound narrowing of the palpebral fissure)

BPES is classified in two types:

Type I - eyelid findings and premature ovarian failure and infertility

Type II - eyelid findings without ovarian failure

Other ocular association with BPES include euryblepharon, strabismus, microphthalmos, lacrimal drainage abnormalities and optic disc coloboma. Extra ocular manifestations include a broad, flat nasal bridge, arched palate, and cup-shaped ears

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

Retinitis pigmentosa has been described with facial and eyelid anomalies including unilateral

upper eyelid ptosis and enophthalmos⁽⁴⁾ but not BPES. The blepharophimosis-ptosis-epicanthus inversus syndrome has been localized to Chromosomes 3 and 7.^[1] Interestingly, both RP and BPES have been reported in association with abnormalities centered around chromosome 14. Subtelomeric deletion of the long arm of the blepharophimosis-ptosis-epicanthus inversus syndrome has been localized to Chromosomes 3 and 7.^[1]

Till now only one case has been reported having BPES with RP changes, blue dot cataract and primary inferior oblique over action^[2,6] We are reporting this case for the rare association of RP with BPES. Although this could be an unusual chance association, the possibility of a new syndrome complex cannot be ruled out.



Figure .1. Patient with features of Blepharophimosis syndrome.

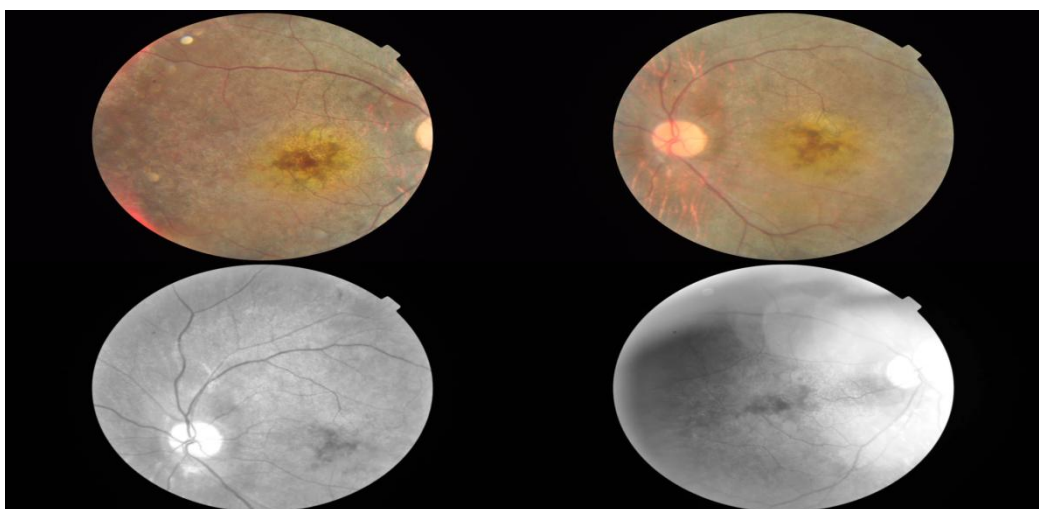


Figure 2. Fundus picture showing RP features.

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