

## Case Report

# Unilateral Retinitis Pigmentosa in One Eye and Tilted Hypoplastic Disc in the Other Eye (Two in One Disease)

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*Pak J Ophthalmol 2007, Vol. 23 No .2*

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Received for publication  
November' 2006  
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We report a case of 32 year old woman who presented with night blindness in her right eye and defective vision in her both eyes. On basis of history, clinical examination, VEP and visual fields, she was diagnosed as a case of advanced unilateral retinitis pigmentosa in her right eye and hypoplasia of optic nerve in left eye with tilted disc and situs inverses.

We describe this unique combination of retinal dystrophy in one eye and congenital anomaly of optic disc in other eye as two in one disease. To our knowledge this rare combination of two retinal pathology has never been reported before.

Since the initial description of this disease by Pedraglia in 1865<sup>1</sup> the so called unilateral pigmentary degeneration of the retina has so far been observed in about 100 cases. However this clinical entity still represents an enigma and its existence and relationship to the bilateral form are debated.

Tilted disc is related to incomplete closure of the optic fissure. This results due to oblique insertion of the optic nerve into the globe. This is a bilateral condition in 80% of patients and associated with moderate to severe hypoplasia or dysplasia of optic nerve. There is thinning of the retina, choroids and sclera along with infero-nasal peripapillary crescent (Fuchs coloboma) and situs inversus of the vessels emerging from the disc. The condition is also associated high amount of myopic oblique astigmatism with refractive and meridional amblyopia resulting in veriable impairment of visual acuity.

To our knowledge this unique combination of retinal dystrophy in one eye and tilted disc with congenital hypoplasia in the other eye of the same patient has never been reported in the literature. We describe it as two in one disease and it was a casual finding in our clinic.

## CASE REPORT

A 32 year old Omani female patient presented in our eye clinic in June' 2001, for complete blindness in her right eye at night. She had history of migraine for which she was already under pharmacological treatment. Her past and family history was unremarkable.

Ophthalmic examination revealed her best corrected visual acuity in the right eye 6/12 P with - 0.5 DSP/-1 DCL @ 160, and in the left eye 6/24 with - 5 DCY @ 30, normal IOP and normal anterior segment.

Her fundoscopic examination revealed a pale disc in her right eye, markedly attenuated retinal arterioles and clumps of bone spicules pigments scattered in the mid periphery in all quadrants (Fig. 1). The macula appeared normal in this eye.

The fundoscopic examination of left eye revealed a hypoplastic small tilted disc, generalized retinal atrophy, anisometric oblique myopic astigmatism with inferonasal peripapillary atrophic crescent. The arterioles showed a bright and normal reflex and macula was normal. No pigmentary disturbance was found anywhere in this retina (Fig. 2).

Visual field examination by a Goldmann perimeter demonstrated a severe loss of peripheral visual field in right eye constricted to 30 degree resulting in tubular vision, where as central vision was spared. Visual field in left eye revealed minimal temporal visual field defects (Fig. 3-a and 3-b).

VEP Using mono-ocular pattern stimulus reversing at a rate of 2 Hz, showed a delay in the P 100 potential on the left side compared to the right, indicating optic nerve pathology in left eye (Fig. 4-a and 4-b). The results of syphilis serology were negative.

ERG Maximum Response (Rod+Cone) was markedly subnormal in right eye and normal in left eye (Fig. 5-a). 30 Hz flicker response (Cone response) was markedly reduced in right eye and normal in left eye (Fig. 5-b).

Patient has been under observation at our clinic since last five years. Uptill 5 years of follow up her ocular findings are unchanged and visual acuity is stable in both eyes.

## DISCUSSION

The etiology of unilateral retinitis pigmentosa is unknown and there has been no proof that it is inherited. Fundus photography, visual field testing, ERG and dark adaptation are necessary to confirm the diagnosis of unilateral retinitis pigmentosa. Many cases of unilateral retinitis pigmentosa have been reported since Pedraglia's initial account<sup>1</sup>, but as reports began to accumulate, the concept that this is a true retinal heredo-degeneration was questioned. In 1952, Francois and Verriest<sup>2</sup> on the basis of a review of 56 patients reported in the literature concluded that only half of them were authentic cases. They then proposed following four criteria which a presumptive case of unilateral retinitis pigmentosa must satisfy:

1. Functional changes and an ophthalmoscopic appearance typical for a primary pigmentary degeneration must be present in the affected eye.

2. Symptoms of a tapeto-retinal dystrophy must be absent in the fellow eye with a normal ERG.
3. An inflammatory cause in the affected eye must be excluded.
4. The period of observation must be long enough to rule out a delayed onset in the fellow eye (over 5 years).

The case presented satisfies the mentioned criteria in all respects. There are a few retinal conditions which mimics the picture of true retinitis pigmentosa. Pseudo retinitis pigmentosa can be caused by trauma, chorioretinitis, drugs, retinal detachments etc. Regarding the first possibility, the patient history was negative for ocular injury, so that this cause could be excluded. Francois and Verriest<sup>2</sup> have mentioned a list of fourteen possible exogenous agents, mostly infectious, which could produce the similar retinal condition. In particular, syphilitic chorioretinitis may resemble retinitis pigmentosa and can be unilateral<sup>3</sup>. However, the results of serological studies of our patient were all negative. Various drugs can cause a pigmentary retinopathy, among which are thioridazine<sup>4</sup> chloroquine<sup>5</sup> and hydroxychloroquine. In this case, the patient had not taken any of these drugs. Retinal detachment may show profound retinal disturbances on settling. The fundus appearance and the patient's history, however, did not show any evidence in support of this possibility. A reasonably long follow up did not reveal delayed bilateral involvement. Therefore we describe it unilateral retinitis pigmentosa. There are reports of association of unilateral retinitis pigmentosa with glaucoma<sup>6</sup>, exfoliation syndrome<sup>7</sup>, and amblyopia. Till today this finding of unilateral retinitis pigmentosa in one eye and tilted hypo plastic disc in other eye has probably never been described, therefore the association of unilateral retinitis pigmentosa and hypoplastic tilted disc in other eye could be regarded as exceptional. Partial amblyopia in left eye is most probably due either to anisometropia or astigmatism.

We are the first to report this exceptional and unique association and we describe it as "two in one disease".

On the basis of the present study it is not possible to state if there is a significant association between the two conditions. Nevertheless, the possibility of relationship in the pathogenesis of both unilateral retinitis pigmentosa and hypoplastic tilted disc in other eye cannot be excluded. Further research is

required to find the nature and pathogenesis of this

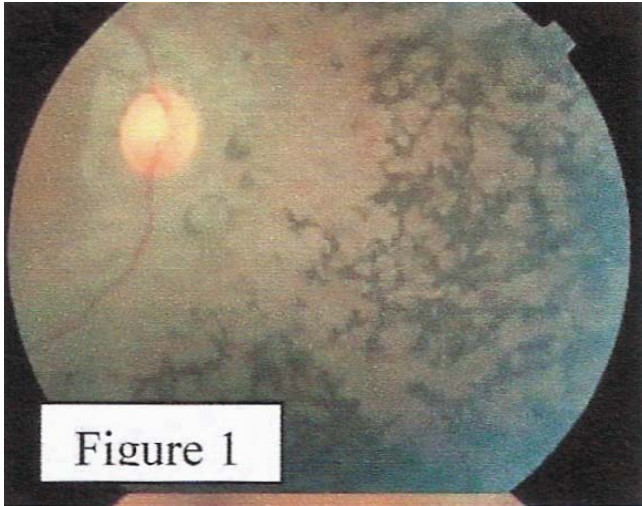


Fig. 1:

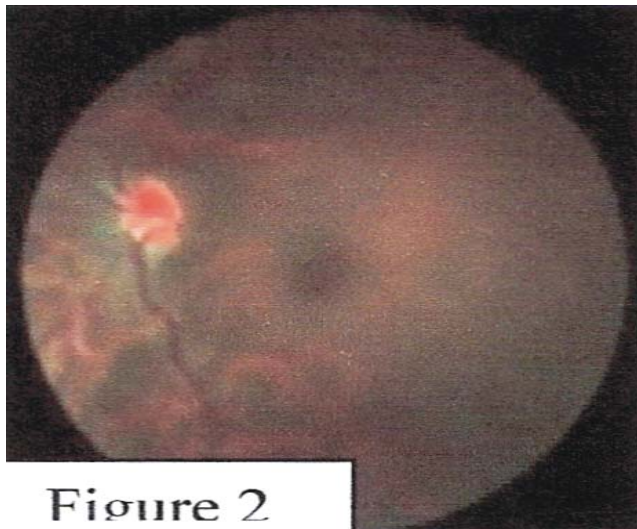


Fig. 2:

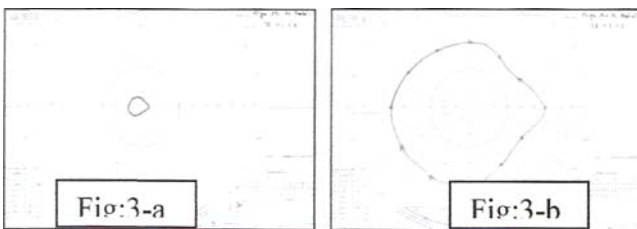


Fig. 3a-3b:



association.

Fig. 4a-4b:

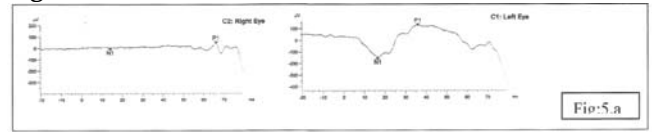


Fig. 5a:

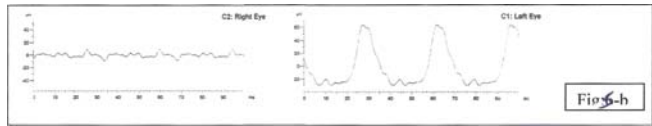


Fig. 5b:

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