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Case Report

Optic Disc Drusen & a Constellation of Other Features of Retinitis Pigmentosa: A Case Report

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Abstract: *Aim:* We describe a case of optic disc drusen in retinitis pigmentosa with a myriad of other ocular associations of retinitis pigmentosa coexisting in our patient. To the best of our knowledge, this is the first case reported in a Nigerian. *Method:* This is a case report of a 16 year old who presented top our eye clinic with a 2 year history of poor night vision with an associated history of difficulty seeing far since childhood. The best corrected visual acuity was 6/18 and 6/12 respectively in the right and left eye. Slit lamp biomicroscopy revealed bilateral grade 1 posterior subcapsular cataracts. Binocular indirect ophthalmoscopy of both eyes revealed vitreous opacities, pale, cupped discs with nasalization of the vessels and yellowish, crystalline deposits at the inferior and superior disc margins. The retina was pale with attenuated vessels, bone spicule pigmentation, atrophic maculopathy and epiretinal membranes bilaterally. *Results:* An assessment of optic disc drusen, glaucoma, atrophic maculopathy with epiretinal membrane in retinitis pigmentosa was made. The diagnosis of optic disc drusen was confirmed by characteristic ocular B-scan findings. He was commenced on guttae betaxolol BD in both eyes and counselled on the nature of the eye pathology. *Conclusion:* Optic disc drusen co-exists with retinitis pigmentosa. Though uncommon, optic disc drusen may occur in Africans as seen in our patient. Other ocular associations including myopia, glaucoma and macula lesions like epiretinal membrane and atrophic maculopathy may also be present. The central visual field changes in a patient with retinitis pigmentosa, optic disc drusen and glaucoma may be attributable to all three ocular entities and not just the glaucoma. Prompt diagnosis, follow –up and adequate patient counselling is essential in the management of these patients.

Keywords: Optic Disc Drusen, Retinitis Pigmentosa, Glaucoma, Myopia, Atrophic Maculopathy, Epiretinal Membrane

1. Introduction

Optic disc drusen (ODD) are calcified deposits located both intracellularly and extracellularly around the optic nerve head which was first described in 1858. [1] These calcified deposits occur in association with various ocular and systemic disorders ranging from acquired myelinated nerve fibres, idiopathic parafoveal telangiectasia to glaucoma, *Down* syndrome, Sturge Weber syndrome and schizophrenia amongst others. [2]

Retinitis pigmentosa *(RP)* is the commonest inherited retinal dystrophy affecting the photoreceptors, the rods initially then the cones subsequently leading to nyctalopia and visual field loss. [3] The classical triad of clinical features in RP are bone-spicule pigmentation, waxy pallor of *the* disc and attenuated vessels. (3) The association between optic disc drusen and retinitis pi gmentosa (RP) has long been established as the very first diagnosis of optic disc drusen was made in a patient with RP. (1, 2) This case report highlights the various ocular associations of retinitis pigmentosa co-existing in our index patient especially the presence of optic disc drusen. To the best of our knowledge this is the first reported case of optic disc drusen in a Nigerian.

2. Case Report

A 16 year old Nigerian male presented to the eye clinic with a 2 year history of poor night vision. There was no antecedent

history of ocular trauma, flashes of light nor floaters. There *was* a positive history of bumping into objects especially at night. He had been wearing spectacles for the past 5 years and the present prescription was two and a half years old. There was no history of redness nor loss of peripheral field vision. He was not a known hypertensive, diabetic, asthmatic nor peptic ulcer disease patient and his genotype *was* AA. There *was* no known history of allergies nor prior surgeries. Family history was not significant for blinding eye disease nor night blindness.

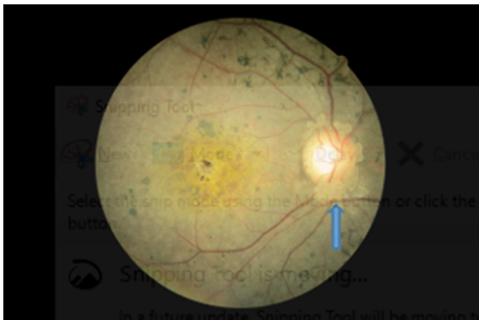
The best corrected visual acuity was 6/18 and 6/12 with refractive correction of – 5.00DS -0.75DC axis 15 and -5.00DS- 1.25 DC axis 15 in the right and left eye respectively. Ocular examination revealed normal anterior segment with early posterior subcapsular cataract (grade 1) in both lenses. The intraocular pressures were 13mmHg in both eyes by Goldmanns' applanation tonometry.

Gonioscopy revealed open angles with all angle structures visible up to the ciliary body corresponding to grade 4 of Schaffers' classification.

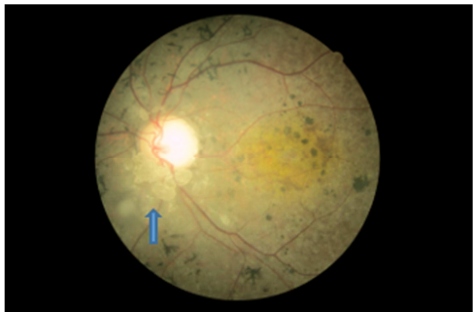
Bilateral vitreous opacities was observed in both eyes. Binocular indirect ophthalmoscopy of the right eye revealed posterior vitreous detachment, a pale, cupped disc with cup disc ratio of 0.6 by 0.6 and nasalization of the vessels with light yellowish, raised, glistening, peripapillary hyaline bodies arising below the rim extending across *six* clock hours from 11.30 to 6.30 o clock. The retina was pale with bone spicule pigmentation along the equator and posterior pole and foveal atrophy with an epiretinal membrane. *(figure 1)*

The disc on the left eye was pale and cupped with a cup disc ratio of 0.9 by 0.7 with nasalization of the vessels and yellowish, glistening, raised hyaline bodies extending across three clock hours from 5.30 to 8.30 o' clock. A posterior vitreous detachment was present while the retina was *pale* with bone spicule pigmentation extending from the posterior pole to the equator with pigmentary changes and foveal atrophy *(figure 2)*. There were no peripheral retinal lesions present bilaterally. A and B ultrasonography scans revealed hyperreflective lesions with high spikes on A –scan corresponding with the location of the drusens at the optic nerve head which are characteristic findings typical of optic *disc* drusen. *(figures 3 & 4)* Optical coherence tomography confirmed the clinical diagnosis of foveal atrophy with central foveal thickness of 61 *u*m and 76 *u*m respectively in the right and left eye and epiretinal membranes on both macula with some distortion of the inner retinal layers as seen in figures *5 & 6*.

An assessment of retinitis pigmentosa with background myopia, atrophic maculopathy and suspicious discs for glaucoma was made. He was reviewed by the glaucoma unit. The central visual fields 24-2 which was documented in the clinical notes showed poor gaze tracking in the patient but revealed bilateral double arcuate scotoma. Optical coherence tomography findings of the optic nerve head was in keeping with glaucoma. Phasing of intraocular pressures which was done over 18 hours did not reveal any diurnal variations. He was subsequently commenced on guttae betaxolol BD both eyes and scheduled for regular follow up by the glaucoma unit.



Figures 1. T*he fundus picture of the right eye showing waxy, disc pallor, bone spicule pigmentation and atrophic maculopathy bilaterally. The optic disc drusen is highlighted by the blue arrow and are seen as yellow, hyaline- like excrescences arising from the optic nerve head margins.*



Figures 2. *The fundus picture of the left eye showing waxy, disc pallor, bone spicule pigmentation and atrophic maculopathy bilaterally. The optic disc drusen as highlighted by the blue arrow is more prominent in the inferior peripapillary area and seen as yellow, hyaline- like excrescences arising from the optic nerve head margins.*

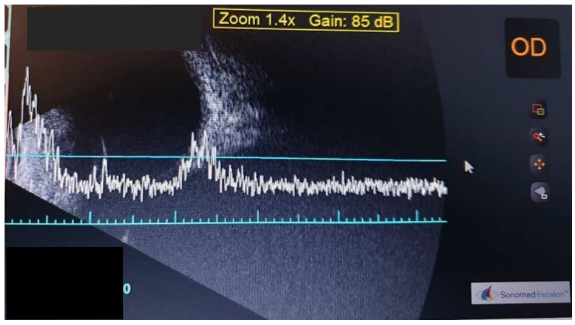


Figure 3. *The B-scan ultrasonography of the right eye with a hyperreflective area on the optic disc corresponding with spikes on A-scan.*

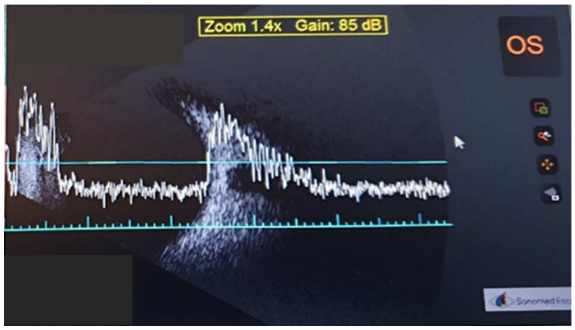


Figure 4. T*he B-scan ultrasonography of the left eye with hyperreflective areas on the optic disc corresponding with spikes on A-scan.*

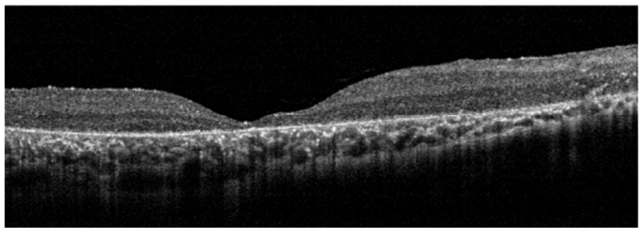


Figure 5. *The optical coherence tomography scan of the macula in the right eye showing marked foveal atrophy with epiretinal membranes, loss of the ellipsoid zone, distortion of the inner retinal layers and chorioretinal atrophy..*

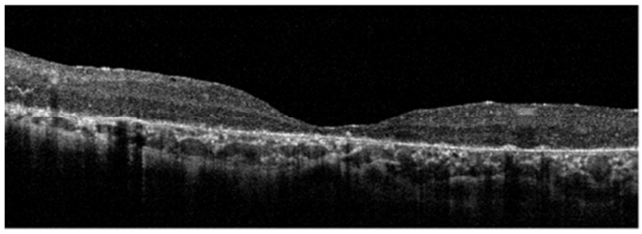


Figure 6. *The optical coherence tomography scan of the macula in the left eye showing marked foveal atrophy with epiretinal membranes, loss of the ellipsoid zone,, distortion of the inner retinal layers and chorioretinal atrophy.*

3. Discussion

Retinitis pigmentosa (RP) is the most common inherited retinal dystrophy affecting about 1:3000 to 4000 individuals worldwide. [3, 4] Various ocular associations have been documented with RP which range from refractive errors, cataract, glaucoma, posterior vitreous detachment and optic nerve drusen to mention a few. [2, 5, 6]

Different macular pathology, more commonly cystoid macula oedema, epiretinal membrane, foveal thinning, vitreomacular traction and macula holes have been described in RP. [7]

Our index patient had a constellation of features including optic disc drusen, glaucoma, posterior vitreous detachment, early posterior subcapsular catara*cts* and myopia coexisting with retinitis pigmentosa; epiretinal membrane and marked foveal atrophy typified by foveal thinning with loss of the ellipsoid zone was present on optical coherence tomography of both maculae.

Optic disc drusens *(ODD)* are known to cause field defects, hence the field changes seen in both eyes of our patient may be due to the combination of both the ODD and glaucoma in both eyes and not just the glaucoma. [8, 10]

ODD are said to be quite rare in blacks and African-Americans possibly due to a larger cup disc ratio and absence of related predisposing genetic factors in caucasians. [10] The gene for optic disc drusen is yet to be identified but it has been postulated from previous studies that the inheritance is in an irregular dominant pattern with incomplete penetrance. [11] To the best of our knowledge, this is the first report of ODD in a Nigerian patient. A similar case of bilateral optic disc drusen and glaucoma in a 17 year old caucasian man with history of same clinical features in his father has been documented in literature though unlike as seen in our patient, there were no features of retinitis pigmentosa present. [12]

Ushers syndrome has also been linked with a higher occurrence of ODD in comparison to all other RP syndromes. [8, 9] Our patient is a healthy young man, a university *undergraduate* with no evidence of mental retardation nor hearing deficit who is doing well academically. Systemic examination was essentially normal with absence of systemic features of syndromic RP.

A and B-scan ultrasonography *as well as* optical coherence tomography were useful in confirming the diagnosis of ODD in our patient. Other investigative modalities like fundus autofluoresence also play an important role in diagnosis of optic disc drusen. In particular, green light fundus autofluoresence has been found to have a high sensitivity for the imaging of optic disc drusen. [8, 13]

Ocular complications like central retinal vein occlusion and choroidal neovascularization occurring as a sequela of optic disc drusen have been reported in literature but these were not present in our patient. [14, 15]

The best corrected visual acuity with spectacles for the patient was 6/18 and 6/12 respectively. This suboptimal vision in our patient with retinitis pigmentosa may be attributed to many factors including foveal thinning with atrophy and loss of the photoreceptors and epiretinal membrane present on both maculae.

Regular follow-up is recommended and essential in monitoring the clinical course and progression of the ocular pathology in our patient. Our patient was interested in knowing if his offspring could be affected in the future and if he would eventually lose his vision. The patient and his parents have been adequately counselled on his diagnosis and the need for regular follow-up emphasized. He has also been counselled on the possibility of needing to learn braille and lifestyle adaptations in the future.

4. Conclusion

Optic disc drusen may co-exist with retinitis pigmentosa. Other ocular associations including myopia, glaucoma and macula lesions like epiretinal membrane and atrophic maculopathy may also be present as seen in our index patient. The central visual field changes in a patient with retinitis pigmentosa, optic disc drusen and glaucoma may be attributable to all three ocular entities and not just the glaucoma. A detailed clinical history and examination combined with available imaging modalities are essential in the diagnosis of optic disc drusen. It is important to differentiate ODD from other causes of pseudopapilloedema like myelinated nerve fibre and tilted discs. A high index of suspicion is also essential for prompt identification of ocular and systemic associations of ODD when present.

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