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Case report
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Is this really juvenile glaucoma?

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Abstract

Background: Pigmentary glaucoma (PG) is a clinical diagnosis characterised by liberation of pigments from the iris pigment epithelium into the anterior segment, with the evidence of glaucomatous optic neuropathy.
Case presentation: A 34-year-old man presented with bilateral visual acuity of 6/9, normal anterior segment, intraocular pressure of 17 mmHg on 3 antiglaucoma eyedrops and cup-to-disc ratio of 0.8. Gonioscopy showed open angles in both eyes. Initial diagnosis was bilateral juvenile open-angle glaucoma. During subsequent follow-up, he was noticed to have bilateral concave iris and dense hyperpigmentation in both angles. Anterior segment optical coherence tomography (AS-OCT) showed bilateral posterior bowing of the mid-iris. Therefore, diagnosis was revised to PG. The reverse pupillary block was successfully treated with laser peripheral iridotomy.
Conclusion: The use of AS-OCT provides an objective visualisation of iris configuration which aids in the diagnosis of PG.

Keywords: anterior segment optical coherence tomography, laser peripheral iridotomy, pigmentary glaucoma

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Adakah ini benar-benar glaucoma juvana?

Abstrak

Latar belakang: Glaukoma pigmen (PG) merupakan satu diagnosis klinikal yang dicirikan oleh penyebaran pigmen dari epitelium pigmen iris ke dalam segmen anterior mata, dengan bukti neuropati optik glaukoma.
Pembentangan kes: Seorang lelaki berumur 34 tahun hadir dengan ketajaman penglihatan dua hala 6/9, segmen anterior yang normal, tekanan intraokular 17 mmHg dengan penggunaan tiga jenis titisan mata antiglaukoma, dan nisbah cup-disc sebanyak 0.8. Gonioskopi menunjukkan sudut terbuka di kedua-dua mata. Diagnosis awal adalah glaukoma sudut terbuka juvana dua hala. Semasa susulan seterusnya, didapati pesakit mempunyai iris cekung dua hala dan hiperpigmentasi yang ketara pada kedua-dua sudut. Tomografi koherens optik segmen anterior (AS OCT) menunjukkan kelengkungan posterior pada bahagian tengah iris secara dua hala. Oleh itu, diagnosis disemak semula kepada PG. Blok pupillari terbalik telah berjaya dirawat dengan laser periferal iridotomi.
Kesimpulan: Penggunaan AS OCT memberikan visualisasi objektif terhadap konfigurasi iris yang mambantu dalam diagnosis PG.

Kata kunci: tomografi koheren optik segmen anterior, laser periferal iridotomi, glaukoma pigmen

Introduction

Pigment dispersion syndrome (PDS) is a clinical diagnosis characterised by liberation of pigments from the iris pigment epithelium into the anterior segment. The prevalence of pigment dispersion syndrome is thought to be relatively uncommon. Ritch *et al.* had reported that in 1993, 2.5% of the screened population in New York City had at least one slit lamp finding consistent with PDS,[1] while approximately 1 in 25 (4.4%) of screened glaucoma patients were found to have features of PDS or pigmentary glaucoma (PG).[2] However, true incidences of PDS and PG are difficult to ascertain and may be higher due to the likelihood of underdiagnosis in view of the subtle clinical signs that can be easily missed.

PDS is characterized by the liberation of pigment from the iris pigment epithelium, which may be seen throughout the anterior segment, for instance, on the corneal endothelium, lens capsule, and in the angle structures. It results from the underlying pathology of a concave iris contour.[3] Over time, PDS can progress to PG, which is a form of secondary open-angle glaucoma where individuals present with glaucomatous optic nerve damage or visual field loss. PG is caused by chronic elevation of IOP as the liberated pigment causes irreversible changes

to the denuded trabecular beam and obstructs aqueous outflow. However, this condition may be underdiagnosed and misdiagnosed, as the ocular symptoms frequently overlap with other clinical entities and the classical clinical signs may be subtle or absent in certain patients or ethnicities. It has been reported that Black and Chinese patients with PDS rarely demonstrate the typical midperipheral radial iris transillumination defects.[4,5]

Hence, anterior segment optical coherence tomography (AS-OCT) is an important imaging modality for visualizing the relation of the peripheral iris to the angle structures, which in turn leads to correct diagnosis and management. The aim of this case report is to highlight the usage of imaging modalities to improve diagnostic accuracy as timely management may prevent vision loss due to misdiagnosis.

Case presentation

A 34-year-old man with no known medical illness presented with progressive worsening painless blurring of vision in his left eye over the course of a month, which he further described as a wider central scotoma. He had previously received treatment at a private eye clinic for 7 years and his condition was stable. The patient denied any other symptoms such as ocular pain, redness, floaters, or metamorphopsia. He was known to have high bilateral myopia of -7.0 dioptres since age of 10. There were no significant history of ocular trauma or steroid usage. His family history was unremarkable.

During the initial examination, the patient's bilateral visual acuity was measured at 6/9. A negative relative afferent pupillary defect was noted, along with a normal anterior segment. The patient's intraocular pressure (IOP) was 17 mmHg on 3 antiglaucoma eyedrops used bilaterally, namely timolol 0.5%, latanoprost 0.005%, and dorzolamide 2%, which had been started by the private eye clinic. IOP prior to treatment was unknown. Fundus examination revealed a cup-to-disc ratio of 0.8, with no signs of retinal tear or break. Gonioscopic findings showed Shaffer grade III open angles bilaterally. Visual field testing showed a glaucomatous paracentral scotoma in the left eye with a mean deviation of -10.66 dB, but in the right eye, visual field test showed poor reliability indices. On the basis of 27 as the age of diagnosis, bilateral cupping of optic nerve head, and open angles in both eyes, he was initially diagnosed as bilateral juvenile open-angle glaucoma.

During subsequent follow-up, the patient's IOP remained uncontrolled despite good compliancy to treatment. His IOP was 23 mmHg bilaterally on 3 similar antiglaucoma eyedrops which was started by private eye clinic. A thorough clinical examination revealed subtle bilateral concave iris with iris pigment on the anterior lens capsule and corneal endothelium. Repeated gonioscopy showed dense hyperpigmentation of the trabecular meshwork in both angles involving 6 clock hours

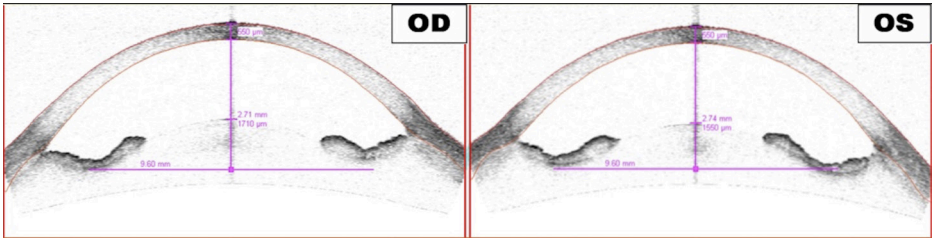

Fig. 1. AS-OCT before LPI.

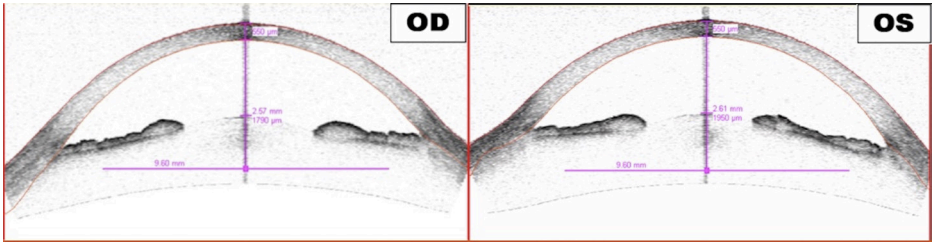

Fig. 2. AS-OCT after LPI.

in both eyes. In suspicion of an abnormal iris contour due to consistent findings of pigment granules in the anterior segment, AS-OCT was performed, which showed posterior bowing of the mid-iris in both eyes (Fig. 1). The diagnosis was revised to PG. In the same setting, the reverse pupillary block was successfully treated with laser peripheral iridotomy (LPI), as evidenced by repeated AS-OCT (Fig. 2). One month post LPI, his IOP was still in the range of 26–28 mmHg with good compliance to medication, hence his antiglaucoma eyedrops were escalated to brinzolamide/brimonidine (10 mg/ml + 2 mg/ml) and travoprost/timolol (40 mcg/ml + 5mg/ml).

Thereafter, his IOP fell within the range of 12–16 mmHg in both eyes while on brinzolamide/brimonidine and travoprost/timolol. Subsequently, the patient defaulted follow-up for a year. A Humphrey 24-2 visual field test 2 years later showed severely depressed fields in both eyes with mean deviation of -24.02 dB in the right eye and -26.13 dB in the left eye. Refractive assessment showed the patient has good unaided visual acuity of 6/7.5 bilaterally. In the latest ocular examination, the anterior segment was unremarkable with a patent peripheral iridotomy, but fundus examination showed a palish optic disc with cup-to-disc ratio of 0.9. Bilateral IOP ranged from 18–20 mmHg as the patient was not compliant to the prescribed medication.

Discussion

PG is a disease with the same underlying pathology as PDS, which is the presence of a concave iris contour that results in rubbing of the posterior pigment layer of iris against the zonules, which leads to the liberation of pigment granules from the iris pigment epithelium and their deposition throughout the anterior segment.[3] Over time, PDS can progress to PG, a form of secondary open-angle glaucoma when there is presence of glaucomatous optic nerve damage and corresponding visual field defects in addition to the classical findings of PDS. The risk factors for conversion of PDS to PG are reported as male gender, myopia, concave iris, and an IOP greater than 21 mmHg at initial examination.[6] Some of which were consistent with our patient.

The prevalence of PG in the general population is not clearly reported. However, it has been reported that the progression of PDS to PG can be as common as 10% at 5 years and 15% at 10 years.[6] In European populations, the prevalence of PG is estimated at 2.45%, assuming 15% of PDS patients develop PG.[7]

The symptoms of PDS include halo, blurry vision resulting from intermittent IOP elevation, increased incidence of non-senile nuclear cataract formation, and visual field loss in advanced cases. PDS patients also tend to have higher risk of retinal lattice degeneration and peripheral retinoschisis, with an incidence of 33.3% and 12.5%, respectively, both of which predispose to retinal detachment. It may be due to the same embryological derivation of pigment epithelium of the iris and retinal pigment epithelium.[8]

The classic findings of PDS are the triad of Krukenberg spindle, spoke-like midperipheral iris transillumination defects, and homogenous heavy trabecular meshwork pigmentation, with some exceptions.[3] It has been found that Black and Chinese patients rarely demonstrate the typical midperipheral radial iris transillumination defects.[4,5] Qing *et al.* reported that out of 1,632 of screened patients, none of the Chinese patients with PDS was found to have radial iris transillumination defects.[5] It was proposed that the reason behind the lack of iris transillumination defects is that dark irides have a greater amount of pigment granules in melanocytes and iris stroma than light irides.[4] The incomplete and at times subtle clinical signs in PDS may delay detection of the disease. Six of the PDS patients in the Qing *et al.* study had been misdiagnosed with primary open-angle glaucoma before referral.[5]

PDS is diagnosed clinically by a thorough slit lamp examination of the anterior chamber focusing on the classic findings of PDS, IOP measurement, fundus assessment, and gonioscopy. To complete the ocular examination and to establish the diagnosis and stages of glaucoma, OCT of the retinal nerve fibre layer and visual field testing can be performed. While gonioscopy is important in the clinical diagnosis of PDS and PG, it is not able to document the findings objectively. AS-OCT and ultrasound biomicroscopy can be used to document iris concavity, insertion site of iris, and lens thickness. Even though PDS is a clinical diagnosis, the use of AS-OCT is of great help in confirming the diagnosis when clinical findings are suspicious.

Trastman-Caruso *et al.* showed that PDS eyes do not have thicker irides or a larger bigger cross-sectional area than controls by using ultrasound biometry.[9] Aptel *et al.* further showed that anterior chamber volume and iridolenticular contact in PDS before LPI were significantly greater and that the ratio of iris volume to length were smaller than in controls.[10] After LPI, the anterior chamber volume and iridolenticular contact area decreased significantly, while the ratio of iris volume to length increased and became similar to those of controls.[10]

As reported by Qing *et al.*, 83.3% of patients had PG and 94.4% had increased IOP at their initial diagnosis of PDS.[5] No formal diagnostic criteria have been established yet. In the future, it would be beneficial to include AS-OCT findings as one of the diagnostic criteria for PDS, as early detection of PDS can improve prognosis by early reversal of the underlying mechanism before the trabecular meshwork architecture changes permanently.

Conclusion

The diagnosis of PDS is commonly missed due to its subtle clinical signs. Hence, it is important to have a high index of suspicion even when the classical signs of PDS are not clearly present, especially in populations with dark irides. Our case demonstrated the usefulness of AS-OCT to provide an objective visualisation of iris configuration and the importance of repeating gonioscopy examination during follow-up, which led to the diagnosis of PG. Timely management may prevent further vision loss due to delayed in diagnosis or misdiagnosis.

Declarations

Informed consent for publication
The patient provided informed consent for the publication of this case report.

Competing interests
None to declare.

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None to declare.

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None to declare.

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