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CASE REPORT

Posterior Polymorphous Dystrophy: An Unusual Presentation

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ABSTRAK

'Posterior Polymorphous Dystrophy' (PPD) adalah penyakit yang jarang ditemui, tidak berbahaya, serta tidak bergejala di mana sel endothelium pada kornea memaparkan sifat serupa seperti epitileum. Ia kebiasannya terdapat pada kedua-dua mata, meskipun pada tahap yang berbeza. Kami melaporkan kes seorang kanak-kanak berumur 10 tahun yang mempunyai sejarah penyakit glaukoma dalam keluarganya tampil mengadu kekaburan pada penglihatannya sejak beberapa tahun yang lepas. Beliau turut menyatakan kerap menukar cermin mata kerana penglihatannya yang tidak bertambah baik. Refraksi penglihatan menunjukkan rabun silau yang tinggi serta 'amblyopia' (mata malas) pada mata kanan. Tahap penglihatan terbaik (diperbetulkan) beliau pada kedua mata adalah 6/9. Pemeriksaan 'slit-lamp' mendapati terdapat luka berbentuk kantung pada lapisan endotelium di bahagian tepian kornea kedua-dua mata. Kepadatan sel endotelium juga berkurangan pada mata kanan dibandingkan dengan mata kiri, dan mata kanan juga mengalami silau yang lebih teruk. Selain daripada itu, pemeriksaan mata tidak mendapati apa-apa yang luar biasa. Pesakit diarahkan untuk menampal mata kiri beliau secara berkala. Walaupun kebiasaannya pesakit 'Posterior Polymorphous Dystrophy' (PPD) dikesan pada umur dewasa, usia pada waktu diagnosis adalah berbeza-beza. Penyakit ini jarang berlaku, dan diwarisi melalui corak 'autosomal dominant'. PPD jarang menyebabkan silau, yang mana ia didapati pada pesakit ini disebabkan permukaan kornea yang berbentuk tidak muncung pada mata kanan dan muncung pada mata kiri. Kepadatan sel endotelium adalah rendah pada mata kanan berbanding mata kiri, di mana ia mungkin menjejaskan ketajaman penglihatan beliau lantas memerlukan pembedahan transplan kornea pada masa hadapan. Pesakit ini juga mempunyai sejarah glaukoma dalam keluarganya, justeru

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itu saringan mata pada ahli keluarga bagi melihat penyakit berkait endotelium adalah diperlukan.

Kata kunci: kornea, mata malas, silau

ABSTRACT

Posterior Polymorphous Dystrophy (PPD) is a rare, innocuous and asymptomatic condition in which corneal endothelial cells display characteristics similar to epithelium. It is often bilateral and frequently asymmetric. We report a case of a 10-year-old girl with a family history of glaucoma who presented with right eye blurring of vision since few years. She had frequent spectacle-prescription changing due to unimproved visual acuity. Cycloplegic refraction revealed high astigmatism and moderate amblyopia over the right eye. Her best corrected vision was 6/9 for the right and left eyes. Slit-lamp examination showed a vesicular-like lesion at the periphery of corneal endothelial layers in both eyes. Endothelial cell density was much reduced on the right eye compared to the left, with more severe astigmatism. Other ocular examinations were unremarkable. Patient was instructed to patch her better left eye periodically. Although patients with Posterior Polymorphous Dystrophy (PPD) present at the age of adulthood, the age at diagnosis is highly variable. It is rare and inherited through an autosomal dominant pattern. PPD may rarely lead to astigmatism, by which was present in the patient due to the features of non-keratoconic and keratoconic cornea on the right and left eye, respectively. Endothelial cell counts were reduced more on the right eye compared to the left, which later may worsen her visual acuity thus indicating corneal transplant in the future. Patient also had a strong family history of glaucoma. Hence, screening of associated disease of corneal endothelial dystrophy in the family may be necessary.

Keywords: amblyopia, astigmatism, cornea

INTRODUCTION

Posterior Polymorphous Dystrophy (PPD) is a disease of the cornea endothelium which the clinical spectrum varied greatly, ranging from presence of posterior corneal vesicles to the formation of peripheral anterior synechiae and cornea edema. Patients can present asymptomatically as an incidental finding or with a certain degree of visual impairment (Teekhasaenee et al. 1991).

PPD also has been reported to be in association with other anterior segment abnormalities such as Iridocorneal Endothelium (ICE) Syndrome and keratoconus (Blair et al. 1992; Kiel 1999). Genetic mutation as the hypothesized pathogenesis of these association has also been studied, which revelaed similar changes in

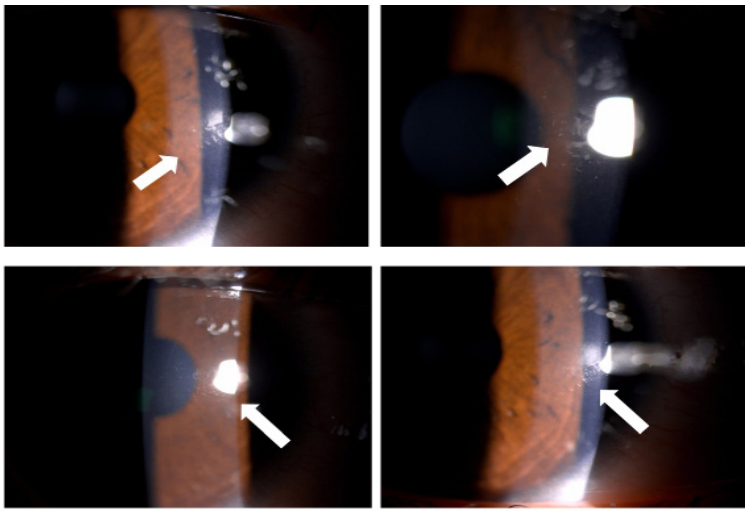

Figure 1: Presence of vesicular-like lesion at periphery of corneal endothelial layers in both eyes (arrow)

their molecular basis and lead to the characteristic corneal dystrophies (Heon et al. 2002).

We report a case of PPD in a child who presented with poor vision secondary to high astigmatism and amblyopia. This case is interesting since the age of presentation and presenting signs and symptoms were rather unusual. The degree of disease severity also did not correlate well with the typical keratoconus.

CASE REPORT

A 10-year-old girl with a family history of glaucoma presented with blurring of vision in the right eye for few years. Her best corrected vision was 6/9 for both eyes. Slit-lamp examination showed vesicular-like lesions at the level of Descemet membrane and endothelial layers of the cornea in both eyes (OU) (Figure 1). Intraocular pressure (IOP) was 12 and 13 mmHg for the right and left eye, respectively. Both optic discs were non-glaucomatous with cup-to-disc ratio (CDR) of 0.4. Other ocular examinations were unremarkable. Her mother was diagnosed to have glaucoma and underwent filtering surgery on the left eye.

Cycloplegic refraction revealed high astigmatism in the right eye (OD) with cylindrical power of -3.00 D, while the left eye (OS) was -0.50 D. Corneal topography showed normal regular astigmatism in OD but irregular astigmatism with paracentral coning suggestive of keratoconus in OS (Figure 2). Endothelial cell density was reduced in the OD (1745 cells/mm^2) compared to the OS (2172 cells/mm^2), with no features of polymegathism or pleomorphism (Figure 3).

Patient was diagnosed to have both eyes PPD with moderate amblyopia in the right eye. She was advised to patch the left eye for 2 hours/day and given follow-up with special attention on worsening of visual acuity, refraction, IOP and endothelial cell counts. Serial

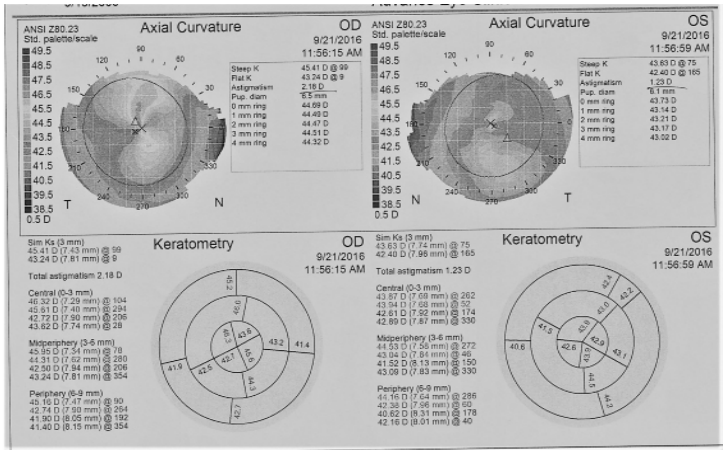

Figure 2: Corneal topography of a patient with PPD showing regular

corneal topography was scheduled to monitor progressive steepening of cornea indicating risk of endothelial dysfunction. Her best corrected vision during follow-up period of 3 months improved to 6/7.5 and 6/6 for right and left eye, respectively

DISCUSSION

PPD is a rare autosomal dominant disorder where there are multi-layered endothelium appearing as vesicles, bands-, or diffuse opacities. Although majority of patients present during adulthood, the presentation age is highly variable due to the broad spectrum of disease severity (Raber et al. 2011). It is usually bilateral, asymptomatic, and non-progressive, with a variable degree of symmetry (Raber et al. 2011). In this case, the patient presented at the age of 10 years with asymmetrical degree of astigmatism, together with incidental findings of PPD which involve both corneas.

While PPD was described in association with keratoconus (Cremona et al. 2009), cases of patients with PPD with non-keratoconic corneas have been reported (Aldave et al. 2013). In these cases, steep corneal curvatures were seen but, no clinical or topographical evidence of keratoconus (Ahn et al. 2017). Interestingly, our patient had both features; keratoconic cornea on the left eye and paracentral vesicular lesions of PPD.

In children, abnormal development of corneal endothelium might affect corneal shape and refractive power, thus decreases their visual acuity (Derespinis et al. 1996). Asymmetric or unilateral involvement of the cornea might risk them to have anisometropic amblyopia associated with greater astigmatism (Derespinis et al. 1996). This is consistent with the findings in our patient where uncorrected asymmetrical astigmatism was detected on both eyes, thus treatment for amblyopia was prescribed which resulted in improvement of visual acuity.

In the present case, the patient was

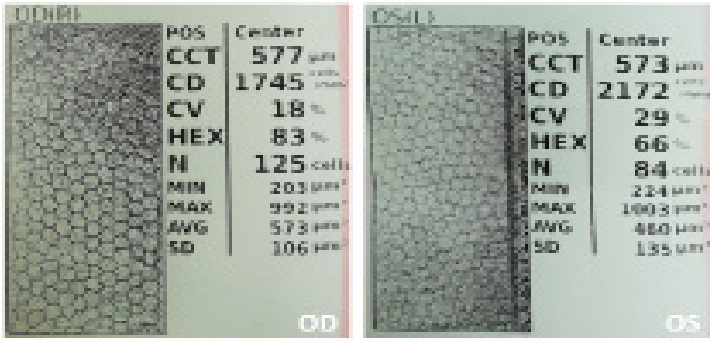

Figure 3: Endothelial specular microscopy for right (OD) and left eye (OS)

prescribed with patching regimen of her better eye for 2 hours/day and best corrected visual acuity was assessed to monitor for improvement. Both strabismic or anisometropic amblyopia would benefit from occlusion of the sound eye for as few as 2 hours/day, with an average of improvement reported as 0.9 lines compared to patients prescribed with optical corrective glasses (Wallace et al. 2006).

Although 25% of patients with PPD were reported to have peripheral iridocorneal adhesions, only 13% of them developed glaucoma, which explains the slowly progressive or non-progressive pathogenesis of PPD (Rodrigues et al. 1980). Gonioscopy and evaluation of optic disc and visual field should be performed to detect glaucoma, as well as screening of family members for glaucoma (Cibis et al. 1977). In the present case, the patient did not have glaucoma at this stage as she was still young but needs to be followed-up closely. A strong family history of glaucoma on the maternal side put her at risk of glaucoma.

Visual prognosis for patients with unilateral PPD may be related to the position of the lesions, age at diagnosis, severity of astigmatism, and early-childhood corrective spectacle use (Jeon & Hyon 2017). Therefore, practitioners should closely monitor patients with PPD for development of anisometropia and amblyopia. Majority of PPD patients may retain normal vision and remain symptom-free (Cibis et al. 1977), avoiding the need for corneal transplant.

Corneal transplant may be required at a later stage especially in eyes with keratometric value greater than 48.0D, as the degree of endothelial dysfunction is greater in eyes with pronounced corneal steepening (Aldave et al. 2013). This explains the importance of monitoring serial corneal topography in PPD cases. Keratoplasty in simultaneous corneal ectasia and PPD may be unavoidable, but clinician must first decide whether the indication is keratoconus or PPD or both. If the PPD lesions are away from visual axis and endothelial cell count is well over 2000, deep anterior lamellar keratoplasty may be the choice of treatment. Likewise, if the endothelial count is poor, performing penetrating keratoplasty may be the better option in such cases.

Essentially, PPD can present in children with high astigmatism and amblyopia. Careful examination of the corneal endothelium together with the refractive status should be performed in a child complaining of blurring of vision. Corneal topography is a useful adjunct in diagnosis of concurrent keratocnonus.

CONCLUSION

PPD can present at an early age and may be associated with high astigmatism which subsequently leads to amblyopia. Corneal topography helps to identify any early sign of keratoconus. Endothelial cell count should be evaluated during follow-up to anticipate cornea decompensation. Proper corneal evaluation aids in prognostication of the need for future corneal transplant and the type of surgery needed.

REFERENCES

Ahn, Y.J., Choi, S.I., Yum, H.R., Shin, S.Y., Park, S.H. 2017. Clinical features in children with posterior polymorphous corneal dystrophy. *Optom Vis Sci* **94**(4): 476-81.

Aldave, A.J., Ann, L.B., Frausto, R.F., Nguyen, C.K., Yu, F., Raber, I.M. 2013. Classification of posterior polymorphous corneal dystrophy as a corneal ectatic disorder following confirmation of associated significant corneal steepening. *JAMA Ophthalmol* **131**(12): 1583-90.

Blair, S.D., Seabrooks, D., Shields, W.J., Pillai, S., Cavanagh, H.D. 1992. Bilateral progressive essential iris atrophy and keratoconus with coincident features of posterior polymorphous dystrophy: a case report and proposed pathogenesis. *Cornea* **11**(3): 255-61.

Cibis, G.W., Krachmer, J.A., Phelps, C.D., Weingeist, T.A. 1977. The clinical spectrum of posterior polymorphous dystrophy. *Arch Ophthalmol* **95**(9): 1529-37.

Cremona, F.A., Ghosheh, F.R., Rapuano, C.J., Eagle, R.C., Hammersmith, K.M., Laibson, P.R., Ayres, B.D., Cohen, E.J. 2009. Keratoconus associated with other corneal dystrophies. *Cornea* **28**(2): 127-35.

DeRespinis, P.A., Norden, R.A., Rispoli, L.C. 1996. Posterior polymorphous dystrophy associated with astigmatism and amblyopia in children. *J Refract Surg* **12**(6): 709-14.

Heon, E., Greenberg, A., Kopp, K.K., Rootman, D., Vincent, A.L., Billingsley, G., Priston, M., Dorval, K.M., Chow, R.L., McInnes, R.R., Heathcote, G., Westall, C., Sutpin, J.E., Semina, E., Bremner, R., Stone, E.M. 2002. VSX1: a gene for posterior polymorphous dystrophy and keratoconus. *Hum Mol Genet* **11**(9): 1029-36.

Jeon, H.S., Hyon, J.Y. 2017. Unilateral posterior polymorphous corneal dystrophy presented as anisometropic astigmatism: 3 case reports. *Case Rep Ophthalmol* **8**(1): 250-8.

Kiel, J. 1999. Posterior polymorphous dystrophy. *Clin Exp Optom* **82**(1): 20-2.

Raber, I.M., Fintelmann, R., Chhabra, S., Ribeiro, M.P.F., Eagle Jr, R.C., Orlin, S.E. 2011. Posterior polymorphous dystrophy associated with nonkeratoconic steep corneal curvatures. *Cornea* **30**(10): 1120-4.

Rodrigues, M.M., Phelps, C.D., Krachmer, J.H., Cibis, G.W., Weingeist, T.A. 1980. Glaucoma due to endothelialization of the anterior chamber angle: a comparison of posterior polymorphous dystrophy of the cornea and Chandler's syndrome. *Arch Ophthalmol* **98**(4): 688-96.

Teekhasaenee, C., Nimmanit, S., Wijtthiphan, S., Vareesangthip, K., Laohapand, T., Malasitr, P., Ritch, R. 1991. Posterior polymorphous dystrophy and Alport syndrome. *Ophthalmology* **98**(8): 1207-15.

Wallace, D.K., Pediatric Eye Disease Investigator Group. 2006. A randomized trial to evaluate 2 hours of daily patching for strabismic and anisometropic amblyopia in children. *Ophthalmology* **113**(6): 904-12.

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