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Case Report

Clinical Findings and Review of Posterior Polymorphous Corneal Dystrophy

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Abstract: The purpose of present study is to discuss the clinical findings and management options of posterior polymorphous corneal dystrophy and to review the current literature on this rare entity. We report a case of asymptomatic posterior polymorphous corneal dystrophy. Posterior polymorphous corneal dystrophy is a rare, familial clinical entity with subtle findings that are important to recognize, as this disease can have serious visual consequences.
Keywords: posterior polymorphous.

INTRODUCTION

Posterior polymorphous corneal dystrophy (PPMD) is an uncommon and often overlooked condition of the corneal endothelium that can range from asymptomatic findings to devastating ocular disease. Diagnosis often involves recognition of subtle clinical signs, but becomes essential when PPMD presents as a vision threatening condition. We present a case of posterior polymorphous corneal dystrophy and review current diagnostic and management options for this condition.

CASE REPORT

A case report was presented. A review of the literature in Science Direct and PubMed was performed. An otherwise healthy 18 year old male presented for a routine eye exam with no visual complaints. On examination, his refraction was measured as -4.00 + 0.75 x 90 in the right eye and -4.25 + 0.75 x 90 in the left eye, which allowed him 20/20 vision bilaterally. His pupils showed no afferent pupillary defect. Intraocular pressure was 14 mmHg in each eye by Goldmann applanation. Motility and confrontational visual fields were full in each eye.

On slit lamp exam, his right eye showed multiple vesicular lesions and snail tracking on the corneal endothelium (Figure 1, Figure 2) but was otherwise unremarkable. Examination of the left eye was unremarkable. His dilated fundus exam showed healthy optic nerves with a cup to disc ratio of 0.3 in each eye. The macula, vessels, and periphery were also noted to be healthy in both eyes.

Specular microscopy was performed and showed pleomorphism and polymegathism of corneal endothelial cells in the right eye (Figure 3). The corneal endothelium of the left eye was noted to be normal. These findings of snail tracking and vesicles on the corneal endothelium are consistent with posterior polymorphous corneal dystrophy.

DISCUSSION

PPMD is a rare, slowly progressive dystrophy of the cornea that presents early in life [1]. The exact prevalence of this entity is unknown. It was first described by Keoppe in 1916 [2]. Inheritance is most commonly autosomal dominant but can less commonly be autosomal recessive or sporadic [3]. Genetic mapping has traced the responsible gene locus to 20q11 [4].While genetic testing can differentiate PPMD into different subtypes genotypically, these subtypes have no difference in terms of clinical findings or disease course [5].

Pathology:

The most distinctive findings of PPMD are abnormal, multilayered endothelial cells that display many clinical characteristics of epithelial cells or fibroblasts [6].The endothelial cells grow rapidly and easily in cell culture. In addition, they show microvilli, stain positive for keratin, have intercellular desmosomes, and display proliferative properties [7].

Clinical Features

PPMD usually manifests between the 3rd and 5th decade and affects females more often than males[8]. Commonly noted symptoms include diminished visual acuity and pupillary distortion. Pain is a less frequent complaint. While symptoms may manifest unilaterally, the uninvolved eye frequently shows abnormal endothelial pleomorphism indicating asymmetric bilateral involvement [9].

Exam Findings

On clinical examination, patients may have small, discreet round lesions that resemble vesicles, often surrounded by a diffuse gray halo [10]. These may appear anywhere in the posterior cornea and may be progressive or remain relatively stable, although very few studies have followed the long-term course of this disease. Broad bands of thickened Descemet's membrane may also be visualized along with large discrete, geographic gray lesions in the posterior cornea [11].Diagnosis is typically made on the basis of these clinical findings. However, corneal imaging studies such as specular microscopy and topography are helpful in evaluating severity and monitoring progression [12]. PPMD may lead to corectopia, broad iridocorneal adhesions and peripheral anterior synechiae[13].Severe cases may also present with stromal and epithelial edema [14].Closed and open-angle glaucoma are both frequent sequelae[15].

It may be difficult to distinguish PPMD from iridocorneal endothelial syndrome (ICE), which is also characterized by endothelial cell abnormalities and subsequent endothelial overgrowth. Both give way to similar appearances of the posterior cornea and can lead to distortion of the iris, peripheral anterior synechiae and glaucoma. However, iridocorneal endothelial syndrome is typically sporadic and unilateral while PPMD often shows autosomal inheritance and presents asymmetrically bilateral [16].In nearly all cases, there are no systemic associations with PPMD[17].However, a recent article by Jang et al suggests that it may be associated with agenesis and hypoplasia of the corpus callosum and a series by Teekhasaenee et al suggests that PPMD may be associated with Alport Syndrome [18,19]. In addition, several reports in the literature discuss an association between PPMD and keratoconus [20].

Treatment/Management

Most cases of posterior polymorphous dystrophy are asymptomatic and thus can be monitored with annual exams and with photos and specular microscopy. Mild epithelial edema may be reduced with the use of hypertonic ointments [21]. More severe disease may require corneal transplantation. Endothelial keratoplasty may also be considered in mild cases that lack significant stromal findings [22]. While there are no large studies evaluating the safety of refractive surgery in patients with PPMD, several small studies suggest that both PRK and LASIK can safely be performed in mild cases. Moshifar et al reported a series of 4 eyes with PPMD in which LASIK showed good outcomes at 1 year [23].Bower et al reported a series of 14 eyes in which PRK was safely performed on patients with PPMD [24].Larger studies with longer follow up after refractive procedures have not been reported.

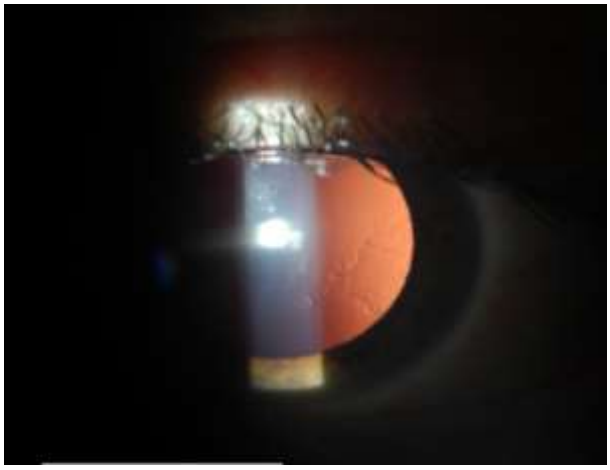

Fig-1: Retroillumination view of the cornea showing posterior vesicular lesions

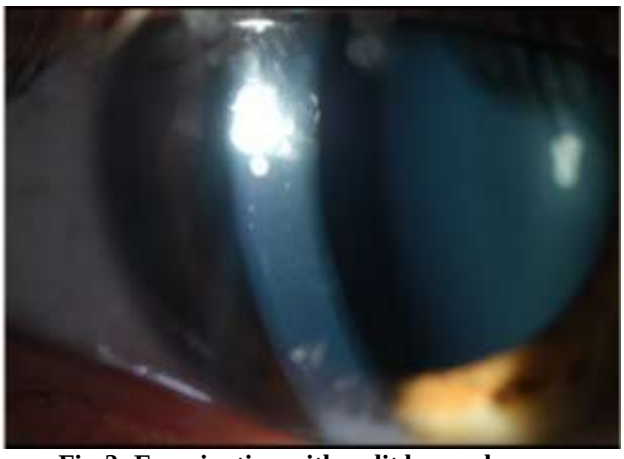

Fig-2: Examination with a slit beam shows characteristic snail tracking of the posterior cornea

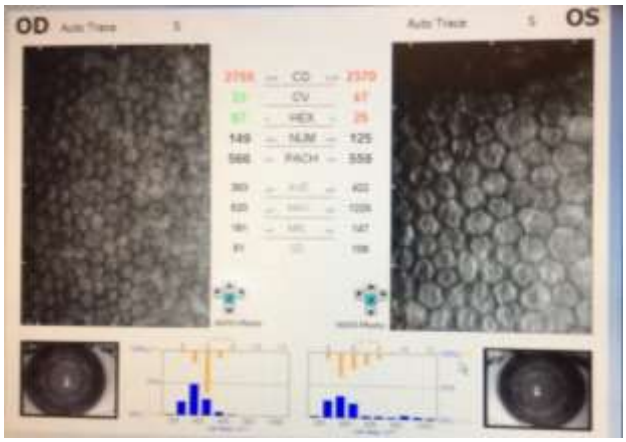

Fig-3: Specular microscopy showing pleomorphism, polymegathism, and a decreased endothelial cell count in the left eye.

CONCLUSION

Posterior polymorphous corneal dystrophy is a rare and perhaps overlooked dystrophy of the posterior cornea with oftentimes subtle clinical findings. However, its more severe forms may be vision threatening. Diagnosis is made on clinical grounds. Management depends on the severity of the clinical

manifestations and can range from simple monitoring to intensive glaucoma therapy or even corneal transplantation. LASIK and PRK have been shown to be safely performed in patients with PPMD.

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