

Crystalline Subtype of Pre-Descemetic Corneal Dystrophy

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Purpose: To report corneal findings in a familial case of the crystalline subtype of pre-Descemetic corneal dystrophy.

Case Report: A 19-year-old girl and her 44-year-old mother were found to have asymptomatic, bilateral, punctiform and multi-colored crystalline opacities across the whole posterior layer of the corneas. Endothelial specular microscopy revealed the presence of white round flecks located at different levels anterior to the endothelium. No systemic abnormalities or medications could be related to account for these findings.

Conclusion: To the best of our knowledge, this is the third familial report of this rare corneal disorder. Differential diagnosis may include Schnyder corneal dystrophy, cystinosis, Bietti's dystrophy and monoclonal gammopathy.

Keywords: Crystalline Subtype Pre-Descemetic Corneal Dystrophy; Crystalline Corneal Deposition Specular Microscopy

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INTRODUCTION

Crystalline corneal deposits appear in a variety of conditions frequently associated with lipid deposition. This finding requires a systemic work-up in order to rule out any systemic disease (hypercholesterolemia, diabetes, uric acid keratopathy, cystinosis, ichthyosis or paraproteinemias);¹ and a specialized corneal examination looking for the presence of posterior crystalline dystrophy, pre-Descemetic corneal dystrophy (PDCD), or punctiform polychromatic pre-Descemetic dystrophy.²

The crystalline subtype of PDCD, so called punctiform polychromatic pre-Descemetic dystrophy, was first reported in Argentina by Fernandez-Sasso et al³ and is included in the pre-Descemetic corneal dystrophies by the

IC3D classification.² More recently posterior crystalline dystrophy has been also described.⁴ Both disorders are characterized by the presence of diffuse bilateral polychromatic crystalline deposits within the deep corneal stroma at the pre-Descemetic level. The posterior crystalline dystrophy is also associated with a white annular peripheral ring.²⁻⁴

PDCD is usually asymptomatic, with mild and non-progressive loss of visual acuity (VA).² Histopathologic studies by light microscopy show enlarged keratocytes in the posterior stroma with vacuoles and intracytoplasmic inclusions containing lipid-like material in an otherwise normal cornea;² transmission electron microscopy shows membrane-bound intracellular vacuoles containing electron-dense material suggestive of secondary lysosomes and inclusions consistent

with lipofuscin-like lipoproteins suggesting a degenerative process.² Although familial cases over 2 to 4 generations have been described, it has remained unclear whether pre-Descemetic dystrophy is a hereditary or a degenerative disorder, as a definite pattern of inheritance or a genetic link has not been established.²⁻⁴

CASE REPORT

A 19-year-old girl complaining of blurred vision in her left eye was referred to our department. VA was 20/20 in her right eye and 20/30 in her left eye. The patient had no remarkable history of ocular or systemic diseases. Slit lamp examination showed paralimbal corneal infiltrates related to contact lens use. The presence of multiple polychromatic crystalline deposits was also noted in both eyes. Treatment with topical moxifloxacin effectively controlled the corneal infiltration and VA rapidly recovered to 20/20.

The corneal deposits were homogeneous in size, polychromatic and diffuse throughout

the entire corneal surface. Specular microscopy (SM) images revealed the crystals as white round flecks located at the pre-Descemetic level, immediately over the endothelial cells. Anterior segment optical coherence tomography (OCT) scans were unable to show the deposits, probably due to their small size (Figure 1). A complete systemic examination was performed without any relevant finding. Renal and hepatic function, blood counts, glucose, cholesterol, uric acid, gamma-globulins, total proteins and proteinogram, and amino acid profile did not show any abnormality.

Her first-degree family members were examined (grandparents, parents and sisters) and only her mother presented with the same crystalline deposits. She was a 44-year-old healthy woman with VA of 20/20 in both eyes. Slit lamp examination revealed bilateral diffuse deep crystalline punctiform corneal deposits (Figure 2). SM images revealed widespread and multiple round white deposits at the pre-Descemetic level, with an irregular distribution. These deposits were also not visible with

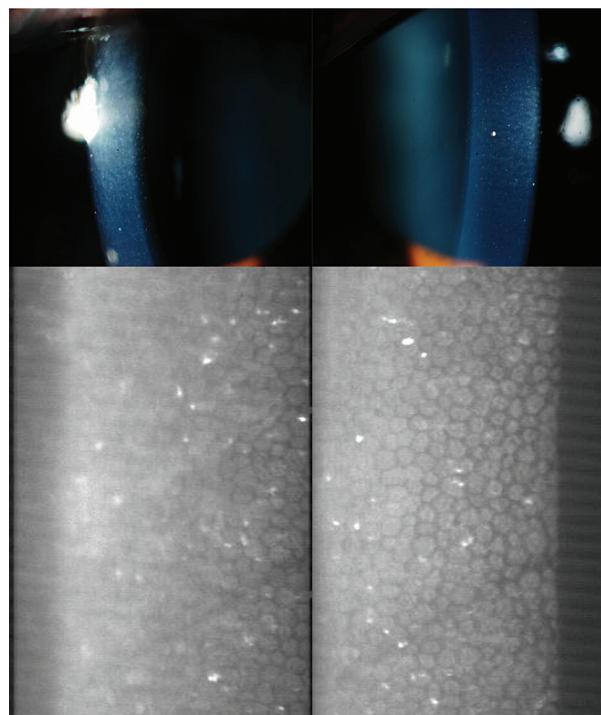


Figure 1. Slit lamp photograph of the 19-year-old female patient showing diffuse crystalline deposits in both eyes; specular microscopy images (lower series) confirm the presence of deep pre-Descemetic deposits.

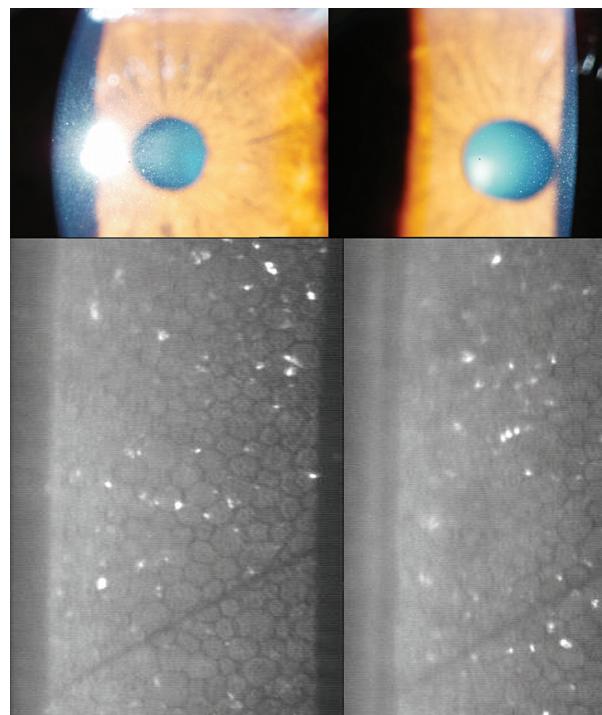


Figure 2. Slit lamp photograph of the 44-year-old woman (first patient's mother) showing diffuse crystalline deposits that are better visualized in specular microscopy images (lower series).

anterior segment OCT. A thorough systemic work-up was performed. Renal and hepatic function, blood count, glucose, cholesterol, uric acid, gamma-globulins, total blood proteins and proteinogram, and blood amino acid profile were unremarkable.

DISCUSSION

The presence of corneal crystalline deposits is frequently associated with a variety of systemic conditions, some of which are life-threatening. Thus, the relevance of this corneal finding is not to provide an ocular treatment but to rule out concomitant and potentially severe systemic pathology.³

The differential diagnosis of corneal crystalline deposits includes several systemic diseases including cystinosis, oxalosis, tyrosinemia, ichthyosis, hypercholesterolemia, diabetes, hyperuricemia, lecithin-cholesterol acyltransferase deficiency, fish eye disease, Tangier disease, dysproteinemias, multiple myeloma, and monoclonal gammopathy (immunotactoid keratopathy).⁵⁻⁶ Furthermore, corneal deposits may be induced by a number of drugs such as gold (chrysiasis), indomethacin, tamoxifen, chlorpromazine, chloroquine, and clofazimine.⁵⁻⁷

Corneal crystalline deposits may also appear in association with different corneal dystrophies. The Schnyder corneal dystrophy is characterized by central comma-shaped crystalline corneal opacities. In Bietti's crystalline corneo-retinal dystrophy, crystals are deposited in the corneal periphery. Infectious crystalline keratopathy is typically related to corneal grafts or long-term corticosteroid use. Dieffenbachia sap related keratitis could also appear with transient pin-shaped crystalline corneal deposits. In posterior crystalline dystrophy and PDCD, there are diffuse polychromatic crystals in the deep corneal stroma with irregular bilateral distribution, the former also has annular peripheral corneal clouding.^{2-4,8}

In the two cases reported herein, no systemic association was detected after a complete systemic work-up. According to slit lamp and SM appearance, both subjects were diagnosed as having the crystalline subtype of PDCD. To our knowledge, this is the third familial occurrence of this rare form of punctiform posterior crystals. The presence of two cases in the same family supports the hypothesis of an inherited occurrence. Further studies should be performed to clarify the inheritance pattern of this rare corneal disorder.

Conflicts of Interest

None.

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