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Case Report

Atypical presentation of macular corneal dystrophy

Priyadarshini Parthasarathi[1,*], **Venipriya**[1], **Justin Prashanth**[1], **Hannah Ranjee Prasanth**[1]

[1]*Dept. of Ophthalmology, Pondicherry Institute of Medical Sciences, Puducherry, India*

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ABSTRACT

A 65-year old male patient presented to our ophthalmology OPD for regular check up. On examination visual acuity of the right eye was 6/24 improving to 6/12p with pinhole and left eye was 6/18 improving with pinhole 6/9. On examination of anterior segment both eyes cornea showed multiple white round deposits at deep posterior stroma and Descemet membrane – endothelium complex scattered circumferentially in the peripheral cornea and the central cornea clear and lens showed Immature cataract. Fundus examination was within normal limits. A differential diagnosis of stromal corneal dystrophy or endothelial corneal dystrophy was made. By exclusion, we came to the diagnosis of macular corneal dystrophy.

1. Introduction

Corneal dystrophies (CD) are defined as a group of inherited non-inflammatory disorders of abnormal deposition of substances in the cornea. The term CD was coined in 1890 by Arthur Groenouw and Hugo Biber, and the efforts of Ernst Fuchs, Wilhelm Uhthoff, and Yoshiharu Yoshida solidified the foundation of the understanding of these diseases.[1] Corneal dystrophies can be divided by anatomical classification and IC3D classification. They are again subdivided into many types. Corneal dystrophy is often bilateral and symmetrical. Corneal dystrophy is not an inflammatory disorder and is not related with to environmental or systemic factors. The final treatment for all dystrophies is keratoplasty although recurrence can occur in some dystrophies.

2. Case Report

A 65-year-old male, a resident of Pondicherry, hospital attender by occupation, came to ophthalmology department for regular check-up. C/o defective vision in both eyes for 6 months. No h/o of ocular pain, photophobia, redness or watering of eyes. No other comorbidities. No significant family history. Visual acuity of the right eye was 6/24 improving to 6/12p with pinhole and left eye was 6/18 improving with pinhole 6/9. Slit lamp examination of anterior segment of both eye cornea showed multiple white round deposits at deep posterior stroma and Descemet membrane – endothelium complex scattered circumferentially in the peripheral cornea sparing the limbus with clear central cornea and sensation was normal.

Both eyes showed immature cataract.

AS – OCT: Anterior segment OCT showed deposits in the posterior stroma with normal corneal thickness.

Dilated fundus examination was found to be normal in both eyes. Since central cornea was clear and patient was not willing for cataract surgery, he was advised to come for regular follow up.

* *Corresponding author*.
E-mail address: priyadarsh.ini3696@gmail.com (P. Parthasarathi).

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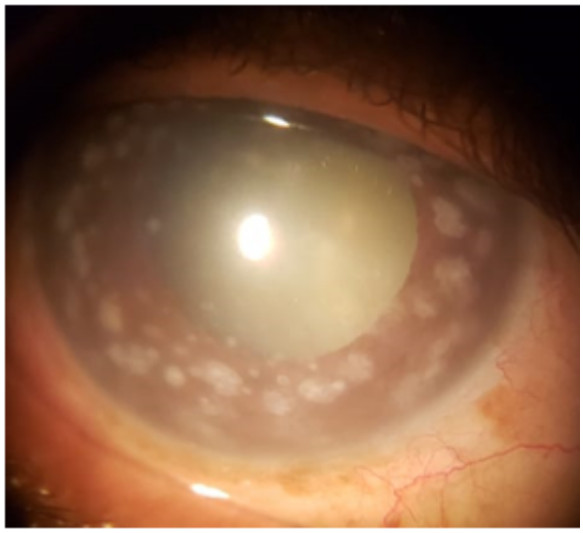

Fig. 1: Right eye showing ring of white stromal and descemet's deposits with clear central cornea

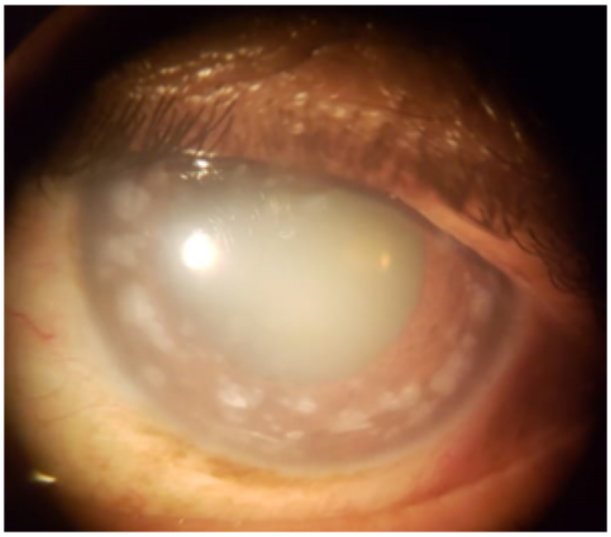

Fig. 2: Left eye showing ring of white stromal and descemet's deposits with clear central cornea

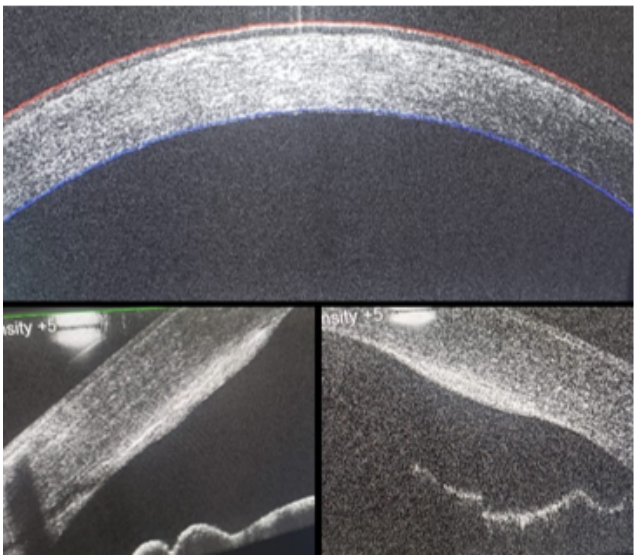

Fig. 3: Right eye AS OCT showing hyper reflective deposits seen in pre descemet's membrane in the peripheral cornea

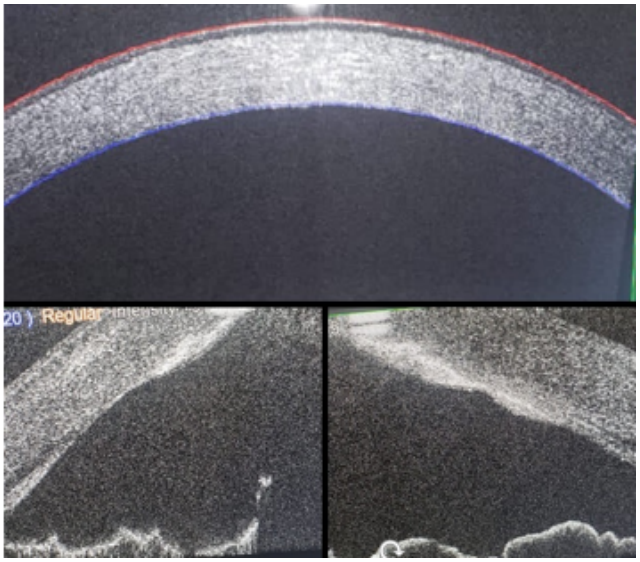

Fig. 4: Left eye AS OCT showing hyperreflective deposits seen in pre descemet's membrane in the peripheral cornea

3. Discussion

Corneal dystrophies are defined as a group of inherited disorders that affect any layer of the cornea and are usually bilateral, progressive, symmetrical, non-inflammatory and not related to any environmental and systemic factors.[2]

Depending on the anatomical sites, CDs can be classified into 3 subtypes: Anterior, stromal and posterior corneal dystrophy.

1. Anterior CD: Anterior basement membrane dystrophy (ABMD) and Meesman's epithelial dystrophy.
2. Stromal CD: Reis-Buckler's dystrophy, honeycomb dystrophy, lattice dystrophy, granular dystrophy, Avellino dystrophy, macular dystrophy, Schnyder crystalline dystrophy, Fleck dystrophy and congenital hereditary stromal dystrophy.
3. Posterior CD: Fuch's dystrophy, congenital hereditary endothelial dystrophy, and posterior polymorphous dystrophy.[3]

Depending on the genetic, clinical and pathological information, IC3D classification classifies corneal dystrophy as

1. Epithelial dystrophies

2. Bowman's layer dystrophies
3. Stromal dystrophies
4. Endothelial dystrophies.[4]

Macular corneal dystrophy (MCD) is an autosomal recessive corneal stromal dystrophy characterized by bilateral diffuse stromal haze and scattered focal stromal opacities that predominantly involve the anterior stroma in the centre of the cornea and the posterior stroma in the peripheral cornea. Mutations in the carbohydrate sulfotransferase 6 gene (CHST6) are causative of MCD. CHST6 protein in human cornea is responsible for the enzymatic sulfation of keratan sulfate (KS), a key process in collagen matrix assembly and homeostasis[5] MCD is classified into two subtypes, type I and type II, defined by the respective absence and presence of sulphated keratan sulphate in the patient serum, although both types have clinically indistinguishable phenotypes.[6]

Macular corneal dystrophy is least common than of all the corneal stromal dystrophies. The onset of corneal haze is variable. It can be seen in infancy but usually apparent in the second or later decades of life. Visual impairment can be severe, especially by mid-life. Corneal thickness is reduced, presumably due to abnormally dense packing of collagen fibrils in the stroma. The epithelium does not seem to be involved. The stroma, Descemet membrane, and endothelium are involved as keratocytes and endothelial cells accumulate intracytoplasmic vacuoles of glycosaminoglycans. They stain with alcian blue.[7] Usually central cornea will be involved but in our case it is unique as the deposits characteristic of macular corneal dystrophy were scattered more toward the peripheral cornea.

Only one or two reports mentioned about isolated peripheral corneal involvement sparing the central cornea.[5]

Recurrent corneal erosions are treated with a bandage contact lens and antibiotics. After early healing of the corneal erosion, preventive treatment consists of sodium chloride 5% drops and artificial tear lubricants during the day and sodium chloride 5% ointment at night. The hypertonic salt medicine helps to secure the bond between the epithelium of the cornea and the underlying basement membrane layers to prevent recurrence of the recurrent corneal erosion.[8] Substantial corneal erosions and a slight reduction in visual acuity can be treated with Phototherapeutic keratectomy (PTK) using an excimer laser. PTK usually only works well for very superficial opacities.[8] Penetrating keratoplasty is the treatment of choice in case of severe reduction in visual acuity or large and deep opacities.[9] Recurrence can occur many decades after PKP.[10]

4. Conclusion

Screening of the CHST6 gene and measurement of serum keratan sulfate are useful means to confirm or exclude the diagnosis of MCD in cases with an atypical presentation. Only very few case reports have mentioned about this type of macular dystrophy with peripheral corneal involvement.

5. Source of Funding

None.

6. Conflict of Interest

None.

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Author biography

Priyadarshini Parthasarathi, Resident https://orcid.org/0000-0001-8386-7424

Venipriya, Associate Professor

Justin Prashanth, Senior Resident

Hannah Ranjee Prasanth, Professor

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