Case Report

Macular Corneal Dystrophy in an Adolescent Managed with Deep Anterior Lamellar Keratoplasty

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Introduction: Macular corneal dystrophy (MCD) is a rare autosomal recessive stromal dystrophy. The prevalence of inherited diseases varies in different communities. It is not commonly observed in Nepal.

Case: A 14-year male presented with bilateral gradual and painless blurred vision with photophobia of three years duration. Cornea of both eyes showed focal grayish opacities in anterior and mid stroma with intervening stromal haze. There was no history of consanguinity in the family. Anterior segment ocular coherence tomography showed thinning of cornea and deposits in corneal stroma. Macular corneal dystrophy was the clinical diagnosis and the patient underwent deep anterior lamellar keratoplasty in the right eye. Histopathological examination and histochemical staining with Alcian Blue supported the diagnosis.

Conclusion: Significant improvement in vision was observed in the operated eye. MCD may be present in communities without evidence of consanguinity. It can cause visual impairment at an early age. Timely intervention improves vision and quality of life.

Key words: Alcian Blue stain, Deep anterior lamellar keratoplasty, Macular corneal dystrophy.

INTRODUCTION

Corneal dystrophies are rare inherited disorders that are bilateral, non-inflammatory, symmetric, progressive and not related to environmental or systemic factors (Aggarwal et al, 2018; Singh et al 2021). Macular corneal dystrophy (MCD) is an autosomal recessive disease and is common in regions with consanguinity in marriages (Aggarwal et al, 2018; Naz et al 2018; Singh et al 2021). There is an abnormal accumulation of glycosaminoglycan primarily in corneal stroma (Aggarwal et al, 2018). Clinically, it shows multiple grayish-white stromal opacities appearing in the central cornea then spreading in the entire cornea causing progressive visual

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impairment (Aggarwal et al, 2018). In Nepal, stromal dystrophy accounted for 0.06% of total corneal transplants, which were either granular or lattice type (Bajracharya et al, 2013).

We report MCD in an adolescent, who underwent deep anterior lamellar keratoplasty (DALK) with improvement in vision.

CASE

A 14-year male presented to cornea clinic with blurring of vision in both eyes (BE) for 3 years, which was gradual, painless and progressive in nature and associated with photophobia. He was not able to perform well in school. There were no history or past records to suggest any inflammatory or infectious cause for his ocular conditions. There was no consanguinity in the family and such ocular conditions were not known in his family pedigree in three generations. At presentation his unaided visual acuity (VA) was 6/60 in the right eye (RE) and 6/36 in the left eye (LE) without any improvement with correction in either eye. Anterior segment evaluation of BE showed focal grayish white opacities with indistinct borders in anterior and mid corneal stroma with relatively thin cornea in slit beam. The intervening stroma between opacities was hazy (Figure 1). Anterior segment ocular coherence tomography (ASOCT) showed thin cornea (420µm in RE and 440µm in LE) with deposits in bowman's layer and anterior corneal stroma (Figure 2). Clinical diagnosis of MCD was made.

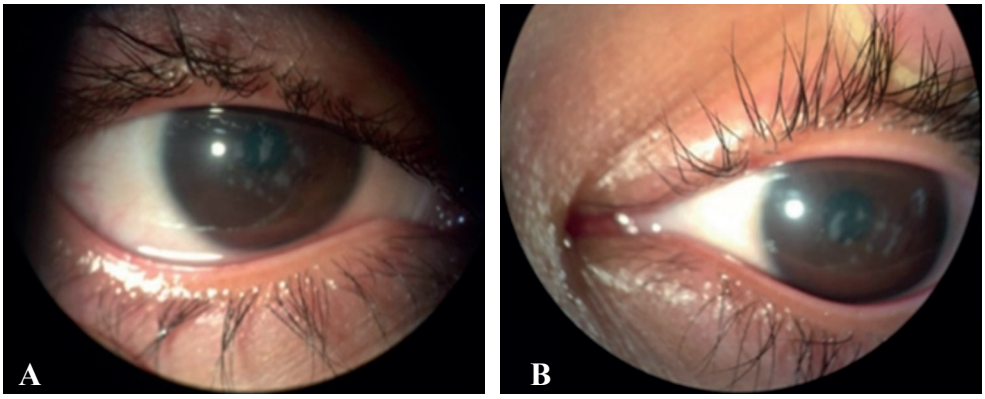

Figure 1: Anterior segment of the right eye (A) and left eye (B) showing whitish opacities in the central and paracentral cornea at the level of anterior and mid stroma.

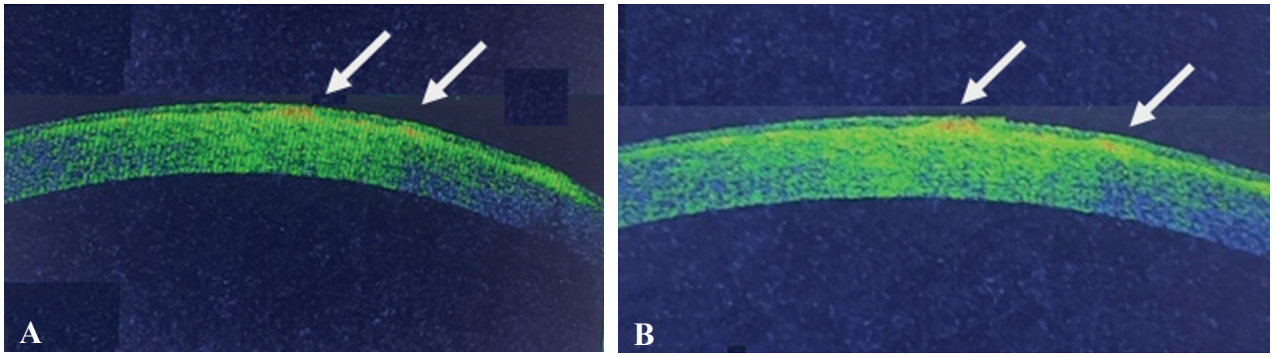

Figure 2: Anterior segment OCT showing deposits (white arrows) in the anterior corneal stroma

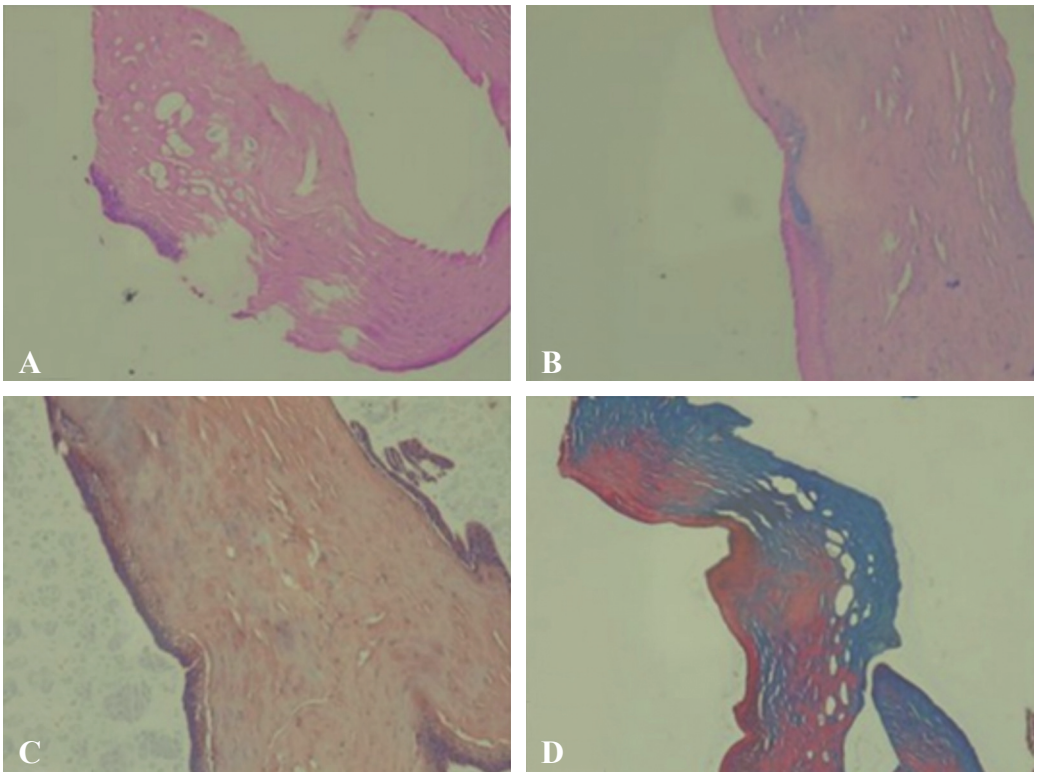

Figure 3: A: Histopathology section of cornea showing glycosaminoglycans deposited in stroma. B: Alcian blue stain showing mucopolysaccharide material deposited in corneal stroma. C: Congo red stain failed to show brick red material. D: Masson Trichome stain showing amorphous material in corneal stroma.

Under general anesthesia, DALK was performed in RE. Donor and recipient sizes were 7.75mm and 7.5 mm respectively. Intraoperatively, a big bubble could not be formed in the plane between stroma and Descemet's Membrane (DM), so stroma was dissected layer by layer to maximum depth. DM stripped from donor cornea, placed over recipient bed and sutured with interrupted 10-0 nylon sutures. Recipient corneal specimen was sent for histopathological examination (HPE) and special stain Alcian Blue. Congo Red and Masson Trichrome staining was also addressed in order to distinguish it from other types of stromal dystrophy. Postoperatively, there was some corneal edema, which was gone in a weeks' time. Patient was given prednisolone acetate eye drop 1% five times/ day, ofloxacin eye drop 0.3% six times/day; eye ointment ciprofloxacin at bedtime initially.

HPE showed glycosaminoglycans deposited in corneal stroma (Figure 3A). Alcian Blue stain showed stain positive mucopolysaccharide deposits in corneal stroma, consistent with macular type of dystrophy (Figure 3B). Congo Red and Masson Trichome staining showed negative for granular and lattice type of dystrophy (Figure 3C and 3D).

His vision started to improve from the second postoperative week. Topical steroids and antibiotic drops were given 4 times a day each

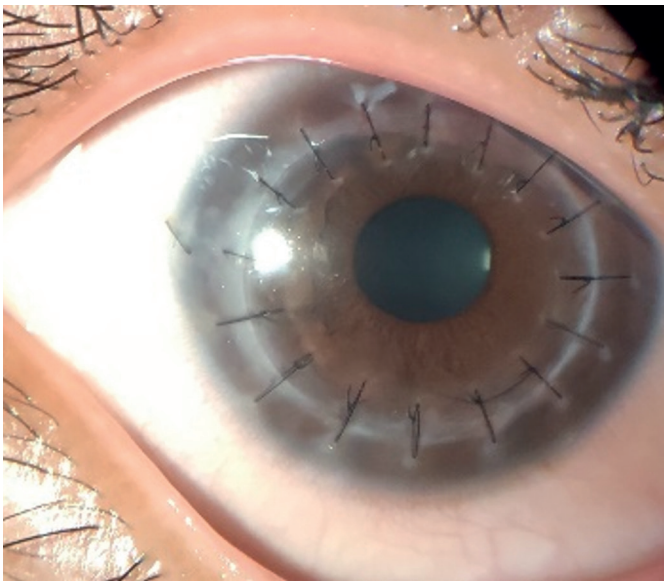

Figure 4: Post-operative DALK surgery at 3 months

and continued for 2 months. Thereafter, topical antibiotics were stopped, topical steroids tapered to 3 times a day and ointment at bedtime was continued. At three months, unaided visual acuity improved from 6/60 to 6/24 and it corrected to 6/12 with a pin hole in the right eye. Graft was clear and the sutures were intact (Figure 4). Steroid drops further tapered to twice a day. Patient was counseled about the need for regular follow up. Genetic counseling was done to the parents regarding the pattern of inheritance and the need to avoid consanguinity in marriage.

DISCUSSION

MCD was first observed and described by German ophthalmologist Oskar Fehr in 1904 (Naz et al, 2018). Autosomal recessive inheritance was first proposed by Bücklers in 1938 due to a defect of the carbohydrate sulfotransferase 6 (CHST6) gene located on chromosome 16 (16q22) (Naz et al, 2018). Because of this, there is alteration in the synthesis of keratan sulphate which is the major glycosaminoglycans in the corneal stroma and is vital in collagen fibril organization and in corneal transparency (Al-Swailem et al, 2005; Naz et al, 2018). As a result of abnormal keratin sulphation, keratan cannot be metabolized and they precipitate in extracellular matrix, also there is decrease in interfibrillar spacing causing loss of corneal transparency (Aggarwal et al, 2018).

According to the registry made at Klinworth in the United States, prevalence of MCD is 0.3 individuals per 2,50,000 inhabitants (Aggarwal et al, 2018; Singh et al, 2021). MCD, a rare entity, has been recognized throughout the world and is most common in regions where there is consanguinity in marriage (Aggarwal et al, 2018; Singh et al 2021). Saudi Arabia and Iceland report more cases of MCD (Aggarwal

et al, 2018; Singh et al 2021). In fact, one third of all corneal transplants were done for MCD in Iceland (Jonasson et al, 1989).

The abnormal deposits in MCD are initially observed in superficial cornea as fleck-like opacities, which later spread to reach the limbus, deeper stroma and the endothelium (Singh et al, 2021). Lesions of MCD start in the first decade of life and the disease often progresses to develop severe vision loss by the third decade of life which is comparatively severe than in other types of stromal dystrophies (Al-Swailem et al, 2005; Weiss et al, 2015). A study done in Pakistan by Naz et al (2018) mentioned 40% of MCD cases presented in the age group 21-30 year and 24% in the age group of 20 years or below. In their study, MCD was seen more in male than females (M: F = 4:1) (Naz et al, 2018).

Phototherapeutic keratectomy (PTK) can improve VA in MCD in early stages, when opacities are limited to superficial stroma, but recurrence rate is high as 90% at average follow-up of 3.4 years (Hafner et al, 2005). PTK can delay keratoplasty, but cannot obviate it. In our patient, clinical evaluation showed opacities up-to mid-stroma and cornea was so thin, therefore PTK was not opted. Penetrating Keratoplasty (PK) was the procedure of choice in the past but currently it is replaced by DALK due to less chance of endothelial rejection (Sogutlu Sari et al, 2013). There had been a rare case of MCD, reported by Chaurasia et al (2019) in which deposits were predominantly at DM and endothelial layer, which was treated with Descemet's stripping endothelial keratoplasty. In our case the deposits were mostly in the anterior stroma, and deeper layers were not involved as shown in ASOCT. At the end of three months of DALK, VA was improved in RE, which enhanced the child's wellbeing with positive impact in his learning and education, which is crucial for his age. He is planned to undergo DALK in left eye as well in the near future. He needs long term follow up, as recurrence of MCD is well known in the literature and had been a challenge (Sogutlu Sari et al, 2013). In a randomized control trial by Sogutlu Sari et al (2013), evidence of recurrent MCD occurred in 4.8% and 5.7% after PK and DALK respectively. Aggarwal et al (2018) mentioned that time from keratoplasty to MCD recurrence ranged from 20 months to 30 years. So it is important to counsel the patient about the possibility of recurrence of the disease. In our patient, in future, if recurrence occurs with significant decrease in vision, a repeat keratoplasty may be needed and the type of surgery (DALK or PK) will depend on the depth of opacities in the stroma.

CONCLUSION

MCD, though being rare, can adversely affect the vision in patients at an early age, and with the advent of surgeries such as DALK, can help regain the vision and wellbeing of the patients. Follow up care is important for any recurrence of the disease.

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