RESEARCH
www.commprac.com
ISSN 1462 2815

FUCHS ENDOTELIAL CORNEAL DYSTROPHY (FECD) WITH CATARACT – A CASE REPORT

Raudatul Janah [1]* and Elfa Ali Idrus [2]

[1] Department of Anatomic Pathology, Cicendo Eye Hospital, Bandung, Indonesia. *Corresponding Author
[2] Department of Oftalmology, Cicendo Eye Hospital, Bandung, Indonesia.

DOI: 10.5281/zenodo.13364497

Abstract

Fuchs Endothelial Corneal Dystrophy (FECD) is a hereditary disease characterized by progressive endothelial cell loss, guttae formation, and worsening stromal edema. It typically presents in the fourth decade of life or later. Patients with FECD are at increased risk of developing cataracts as the corneal disease progresses. Simultaneous optical penetrating keratoplasty, lens extraction, and intraocular lens implantation is an effective treatment options for FECD patients presenting with cataract.

Keywords: Fuchs Endothelial Corneal Dystrophy, FECD, Cataract, Case Report.

INTRODUCTION

Fuchs Endothelial Corneal Dystrophy (FECD) is one of the most common indications for corneal transplantation worldwide. This disorder causes vision loss, with ophthalmological examination the results of corneal edema, endothelial cell density decreased, shape and size, the descement membrane thickened, and guttae were found due to the deposition of extracellular matrix. The defect that occurs in this disorder is progressive endothelial damage which results in endothelial cell mitosis inability. Existing endothelial cells must enlarge and spread to cover the existing defects to maintain a tight layer and function properly as a defense and pump so that it can maintain the clarity of the cornea. The clinical picture of FEDC varies depending on the severity. The cornea guttae appears in the central part and then spreads to the peripheral part. Descemet's membrane thickens and forms folds due to stromal edema. The etiology of FECD is multifactorial, one of which is the presence of oxidative stress and one that relates to a dominant autosomal genetic disorder, namely a mutation in α2 collagen VIII (COL8A2). The incidence of FECD is rare, causing difficulty in assessing its prevalence, often this FECD incidence is obtained from histopathological examinations of patients who will undergo corneal transplantation by an eye specialist. This disorder is more common in women than men, and it usually occurs in both eyes and usually slows down. FECD is divided into early onset and advanced onset, where early onset occurs in the 3rd decade while the advanced onset occurs in the 5-6th decade. In the United Kingdom, it is recommended to treat corneal specimens that have FECD, namely by histopathological examination. The current treatment is keratoplasty.[1-5] This report discusses the diagnosis and histopathology of FECD with cataract so that it will obtain appropriate treatment.

CASE DESCRIPTION

A 78-year-old man was referred to the Infectious and Immunological Clinic of the Eye Hospital Cicendo with a complaint of blurred vision in both eyes. The left eye has been blurred and red for three months, while the right eye has had it for 14 years after being exposed to diesel smoke. The patient has a history of wearing glasses and has

ISSN 1462 2815

received treatment at Al Ihsan Hospital for left cataract, left dry eye syndrome, and right corneal staphyloma.

Ophthalmological examinations showed sharp vision of the right eye light perception and the left eye 0.125. An examination of the anterior segment (figure 1) of right eye showed keratopathy, while the left eyes showed folded Descemet, bullae, guttae, and edema. Right eye ultrasound (figure 2) reveals fibrosis-related vitreous clivage, and left eye OCT (figure 3) reveals central cornea thickening. Biometric measurements of the left eye resulted in a size of +19.50 dioptries for the IOL. The patient was diagnosed with Fuchs Endothelial Corneal Dystrophy (FECD) and cataract, and planned to undergo triple surgery: optical penetrating keratoplasty, lens extraction, and IOL installation.

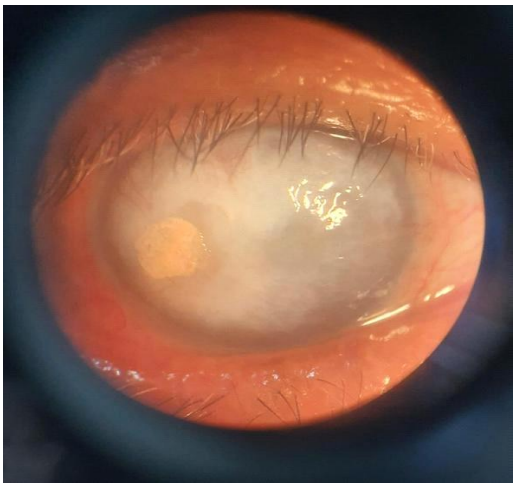

Figure 1: The anterior segment of right eye with corneal opacification in all quadrant

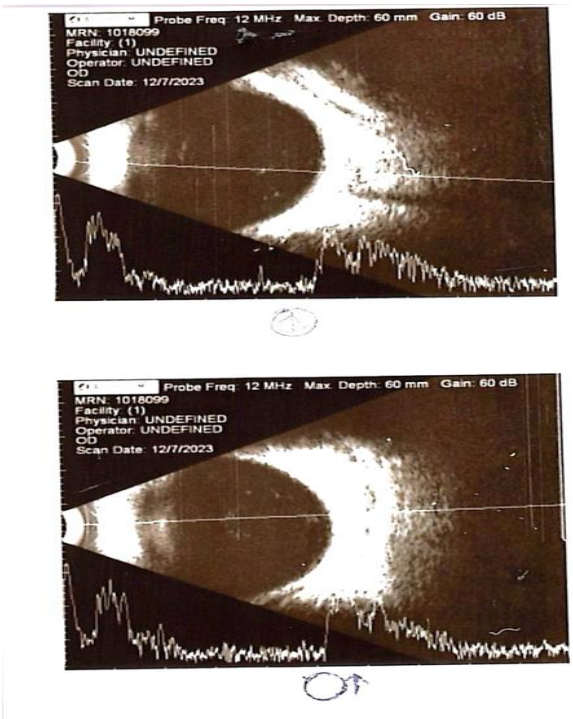

Figure 2: Ultrasonography: On the ultrasound examination of the right eye appears the fibrous vitreous stiffness caused by the fibrosis vitreous

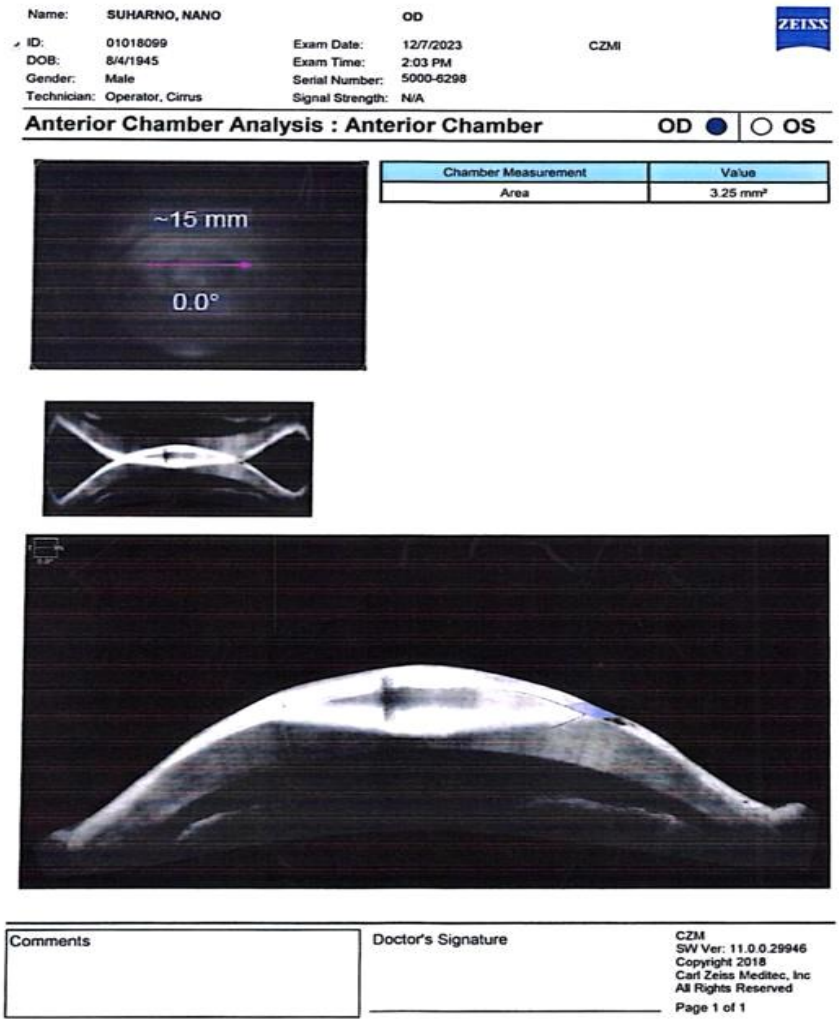

Figure 3: Anterior Chamber Examination: Anterior Segment-Optical Coherence Tomography (AS-OCT) on the right eye with density in all half anterior cornea

{

Figure 4: Histopathological examination of corneal tissue samples.. A.*Bullae* (*) dan stroma edema ({). B. Endothelial Cell flat (↑). C.Loss *basket weave* ({). D.Inflamatory cell ({)

A sample of right-eye corneal tissue was taken for histopathological examination. Stroma edema of fibrous connective tissue found with the loss of basket weave, embedded in polymorphonuclear inflammatory leukocyte cells (PMN), Lymphocytes, plasma cells and histiocytes. The results support the diagnosis of FECD in patients.

Case Summary:

A man, eighty-eight, arrived with double vision. For the previous three months, the left eye has been blurry with a history of recurrent redness and stinging. After being exposed to diesel fog fourteen years ago, the right eye started to turn white and become blurry. Examination revealed keratopathy in the right eye and lens opacity, deschemet folds, bullae, and guttae in the left eye, indicating FECD in both eyes as well as cataract. The histopathology result revealed the classic signs of FECD, including flattened endothelial cells, thickened deschemet membrane, lifted epithelium off Bowman's layer, and edematous stroma. For the right eye, a triple procedure was performed.

DISCUSSION

Corneal transparency is assigned by the corneal endothelial cells that play an active role in pumping fluid out of the tissue, keeping the cornea in a state of deturgence. Corneal rotation, which disrupts transparency, often requires surgical procedures such as a cornea transplant to restore its function. Fuchs Endothelial Corneal Dystrophy (FECD) is one of the most common indications for corneal transplantation.[6-8] FECD is an autosomal dominant hereditary disease that progresses slowly, usually appearing in the fourth decade of life or more. The prevalence of FECD varies globally, with figures ranging from 3.3% in Japan to 11% in the United States. It is estimated that around 300 million 30-year-olds worldwide have FECD, with an increase in the number of cases projected to reach 415 million by 2050. Patients with FECD often come with corneal clogging that can accompany cataracts. In this case, corneal turbidity was found in both eyes, with the right eye worse. The history of patients showed turbidities in the right eye that occurred 14 years ago after exposure to diesel smoke, while the left eye had complaints in the last 3 months. Although corneal stiffness occurs asymmetrically, the condition is suspected to be FECD. Other causes still need to be eliminated by examining the anatomical pathology of the corneal tissue. The age of the patient corresponds to the general characteristics of FECD, which usually appears in the fourth decade of life or later.[6,9-10]

The signs and symptoms of Fuchs Endothelial Corneal Dystrophy (FECD) are usually associated with edema, which can lead to reduced vision, contrast sensitivity, and fainting. Pain can occur as a result of blistering or microcystic edema, often worse when waking up due to reduced surface evaporation. Pain episodes may decrease after subepithelial fibrosis occurs. The manifestations of FECD vary depending on the stage of the disease. In the early stages, corneal guttae appear in the central area and spread to the periphery, creating a "beaten metal" appearance. The second stage is characterized by endothelial decompensation and stroma edema, which can develop into bullous keratopathy in the third stage. In the advanced stage, chronic edema leads to cornea thickening up to 1 mm, with subepithelial fibrosis, scar tissue, and peripheric superficial vascularization occurring in the fourth stage.[6-7,11] FECD patients often complain of a sharp reduction in vision, which can be accompanied by cataracts. In these patients, guttae and Descemet folds were found in the left eye, while the right eye was difficult to evaluate due to keratopathy. In addition, the patient also suffered from cataracts, which were initially referred to as poly cataracts and refractive surgery. An OCT examination of the anterior segment showed a thickening of the central cornea, possibly caused by corneal edema.

The diagnosis of Fuchs Endothelial Corneal Dystrophy (FECD) is confirmed through histopathological examination of corneal tissue. FECD mainly affects the Descemet membrane and the corneal endothelium. The corneal endothelial cells are mostly non-proliferative and are in the G1 phase of the cell cycle. The highest endothelial cell density occurs at birth, approximately 4000 cells/mm^2, and decreases to about 2500 cells /mm^2 in healthy adults. However, in FECD patients, the decrease in cell density is accelerated. The cornea remains in a state of deturgence when the endothelial cell density reaches a minimum of 500 cells/mm^2. If this density falls below 500 cells/mm^2, as is often the case with FECD, the function of the active endothelial pump fails, causing endothel decompensation and edema. Classical signs of FECD in histopathology include epithelial lifting from the Bowman layer, stroma edema, loss of "basketweave" stroma texture, and changes in endothelial cells from a cuboidal shape to a peeling.[12-14] In this case, corneal histopathology examinations showed that the epithelial layer that was lifted from Bowman's layer formed peeling, edema in the stroma, the loss of a basketweave texture, the infiltration of inflammatory cells, and endothelial cell changes to peeling or disappearing. These results are consistent with the classic histopathological findings of the FECD.

Fuchs Endothelial Corneal Dystrophy (FECD) can be performed medicinally and surgically. The administration of hyperosmotic eye drops is a medicaments treatment for FECD, while keratoplasty is the definitive therapy. In FECD patients who also have cataract, triple procedures, which include keratoplasty penetration, lens extraction, and intraocular lens implantation, are considered as suitable surgical techniques.[8-9,]

CONCLUSION

Fuchs Endothelial Corneal Dystrophy (FECD) is a progressive hereditary disorder that can lead to significant corneal edema and vision loss, often necessitating corneal transplantation. Histopathological examination is crucial for confirming the diagnosis, revealing characteristic changes in the corneal layers. In patients with coexisting cataracts, a combined surgical approach involving keratoplasty and cataract extraction offers effective treatment.

Source of funding: This research received no specific grant from any funding agency in the public, commercial or not-for-profit sectors".

Conflict of interest: None to declare."

References

1) Afshari, N. A., Pittard, A. B., & Siddiqui, A. Fuchs Endothelial Corneal Dystrophy: Clinical Presentation, Diagnosis, and Treatment. Ophthalmology. 2016; 123(5): 993-1006.

2) Kwitko, S., & Marback, E. Clinical and Histopathological Findings in Fuchs Endothelial Dystrophy. Ocular Surface. 2019; 17(2): 287-293.

3) Rogers, S., & Symes, R. Genetic and Environmental Factors in Fuchs Endothelial Dystrophy. Experimental Eye Research. 2017; 154: 1-6.

4) Shah, R., & Verma, K. Pathogenesis of Fuchs Endothelial Dystrophy: The Role of COL8A2 Mutation. Journal of Ophthalmic & Vision Research. 2018; 13(3): 256-262.

5) Wang, J., & Xu, X. Histopathological Examination of Corneal Specimens in FECD. Ophthalmic Pathology. 2020; 39(2); 122-128.

6) Feder RS, Berdy GJ, Iuorno JD, et al. BCSC ® basic and clinical science course TM editorial committee external disease and cornea 8. 2022.

7) Ali M, Cho K, Srikumaran D. Fuchs dystrophy and cataract: diagnosis, evaluation and treatment. Vol. 12, Ophthalmology and Therapy. Adis; 2023. p. 691–704.

8) Blitzer Kathryn A Colby AL. Update on the surgical management of fuchs endothelial corneal dystrophy. Tersedia dari: https://doi.org/10.6084/m9.figshare.12783635.

9) Majid Moshirfar, Rachel Huynh, James H Ellis. Cataract surgery and intraocular lens placement in patients with fuchs corneal dystrophy: a review of the current literature. Curr Opin Ophthalmol . 2022 Jan;33(1):21–7.

10) Aiello F, Gallo Afflitto G, Ceccarelli F, et al. Global prevalence of fuchs endothelial corneal dystrophy (fecd) in adult population: a systematic review and meta-analysis. J Ophthalmol. 2022;68-78.

11) Das AV, Chaurasia S. Clinical profile and demographic distribution of fuchs' endothelial dystrophy: an electronic medical record-driven big data analytics from an eye care network in india. Indian J Ophthalmol. 2022 Jul 1;70(7):2415–20.

12) Thaung C, Davidson AE. Fuchs endothelial corneal dystrophy: current perspectives on diagnostic pathology and genetics - bowman club lecture. Vol. 7, BMJ Open Ophthalmology. BMJ Publishing Group; 2022.

13) Bhargava M, Biswas J, Patil S. Atypical variant of fuchs' endothelial dystrophy mimicking iridocorneal endothelial syndrome: a case report. Indian J Ophthalmol. 2020 Oct 1;68(10):2274–6.

14) Chou WY, Kuo YS, Lin PY. Cataract surgery in patients with fuchs' dystrophy and corneal decompensation indicated for descemet's membrane endothelial keratoplasty. Sci Rep. 2022 Dec 1;12(1).

15) Sunita Chaurasia MD, Francis W. Price Jr. MD, Lauren Gunderson BS, et al. Descemet's membrane endothelial keratoplasty: clinical results of single versus triple procedures (combined with cataract surgery). J.ophthal. 2014;121(2):454–8.