DOI:10.14744/bej.2022.87487
Beyoglu Eye J 2023; 8(1): 60-63

Case Report

Wilson Disease Combined with Keratoconus: A Case Report

Adem Tellioglu, Derya Ozkan, Burcin Kepez Yildiz

Department of Ophthalmology, University of Health Sciences Beyoglu Eye Training and Research Hospital, İstanbul, Türkiye

Abstract

A case of Wilson disease (WD) combined with keratoconus (KC) is described. A 30-year-old male diagnosed with WD presented to Ophthalmology Department due to progressive bilateral vision loss. Biomicroscopy revealed copper depositional ring and mild central corneal ectasia in both eyes. The patient had essential tremors and mild speech disturbance. The keratometric values were K1 = 45.94 diopters (D), K2 = 49.10 D in the right eye, and K1 = 47.14 D, K2 = 51.22 D in the left eye. The maximal elevation points on the posterior elevation maps were 98 mm for the right eye and 94 mm for the left eye. The typical KC pattern was seen on corneal topography bilaterally. Based on these findings, the patient was diagnosed with KC, and corneal cross-linking treatment was recommended. WD rarely occurs in combination with KC, and only two cases have been reported; this is the third case of WD combined with KC so far.

Keywords: Corneal diseases, hepatolenticular degeneration, keratoconus, liver diseases

Introduction

Keratoconus (KC) is a non-inflammatory, bilateral, and progressive eye disorder characterized by corneal thinning and ectasia. KC is associated with some conditions, including Down syndrome, Leber congenital amaurosis, and connective tissue disorders including Marfan syndrome and Ehlers-Danlos syndrome (1-3). Hepatolenticular degeneration (Wilson disease [WD], OMIM No. 277900) is a hereditary disease (caused by a mutation in the copper transporter gene ATP 7B) in which the body accumulates excessive levels of copper, primarily in the liver, brain, and eyes. WD rarely occurs in combination with KC, and only two cases have been reported (4,5). To the best of our knowledge, this is the third case of WD combined with KC so far.

The patient signed written informed consent for the research use of clinical records and data included in the study.

Case Report

In February 2021, a 30-year-old man with WD was referred to the ophthalmology department with bilateral visual disturbance. He has no family history of KC or WD. His uncorrected visual acuity was 0.2, spectacle best-corrected visual acuity was 0.6 in the right eye; uncorrected visual acuity was 0.2, and spectacle best-corrected visual acuity was 0.5 in the left eye. Marked scissoring was noted on retinoscopy and, intraocular pressure was 16 mm Hg bilaterally. Ophthalmologic examination using the slit-lamp showed copper depositional ring (Kayser-Fleischer [KF], KF, ring) and mild central corneal ectasia in both eyes (Figs. 1, 2). The KF rings widest

How to cite this article: Tellioglu A, Ozkan D, Kepez Yildiz B. Wilson Disease Combined with Keratoconus: A Case Report. Beyoglu Eye J 2023; 8(1): 60-63.

Address for correspondence: Adem Tellioglu, MD. Department of Ophthalmology, University of Health Sciences Beyoglu Eye Training and Research Hospital, İstanbul, Türkiye
Phone: +90 506 610 73 61 **E-mail:** ademoztel@hotmail.com

Submitted Date: July 17, 2022 **Revised Date:** July 17, 2022 **Accepted Date:** September 07, 2022 **Available Online Date:** March 01, 2023

Beyoglu Eye Training and Research Hospital - Available online at www.beyoglueye.com

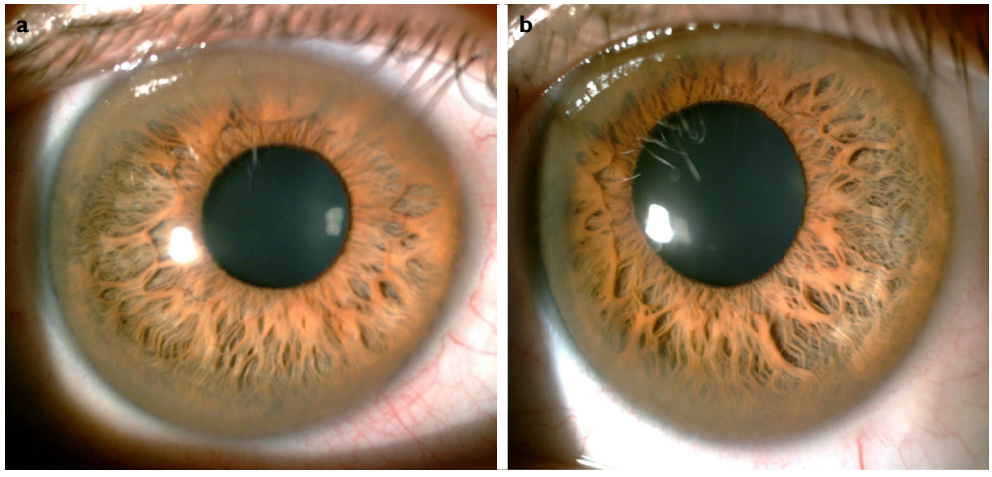

Figure 1. Anterior segment photograph showing the Kayser-Fleischer ring over 360° of the peripheral cornea.

point was at twelve o'clock and, the narrowest point was at the 3 o'clock position. Fleischer ring (iron deposition ring in deep epithelium), Vogt's striae, hydrops scars, and Munson sign were absent. Pupils were isochoric, the lens was transparent, and there was no pathology on fundus examination. The patient had essential tremors and mild speech disturbance. Laboratory findings were also significant for the low level of serum copper concentration (20 µg/day), low level of serum ceruloplasmin (4.4 mg/dL), and elevated urine copper excretion (248 µg/day). The patient had undergone hepatic transplantation 3 years before. Hepatic ultrasonography before transplantation showed fatty steatotic changes and fibrosis.

During Sirius corneal topography (Costruzione Strumenti Oftalmici, Florence, Italy) assessments, central corneal thickness was detected to be 456 µm in the right eye and 449 µm in the left eye. The keratometric values were K1 = 45.94 diopters (D), K2 = 49.10 D in the right eye, and K1 = 47.14 D, K2 = 51.22 D in the left eye (Fig. 3). On the posterior elevation maps, the maximum elevation points were 98 mm for the right eye and 94 mm for the left. The characteristic pattern of KC was seen on corneal topography bilaterally (Fig. 3). Based on these findings, the patient was diagnosed with KC.

Considering the laboratory values and corneal topography of our case, corneal cross-linking treatment was recommended but the patient discontinued follow-up.

Discussion

KC is a non-inflammatory corneal ectatic condition that is progressive and predominantly bilateral. It causes myopia and irregular astigmatism that frequently results in visual loss with an onset typically in early adulthood. Complex interactions between genetic and environmental factors

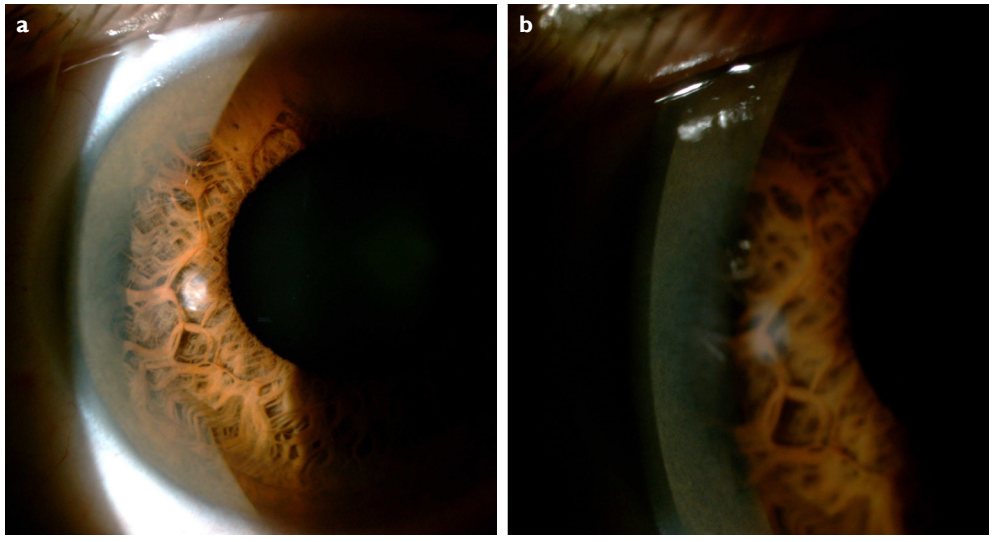

Figure 2. Copper deposits in the Descemet's membrane of the cornea.

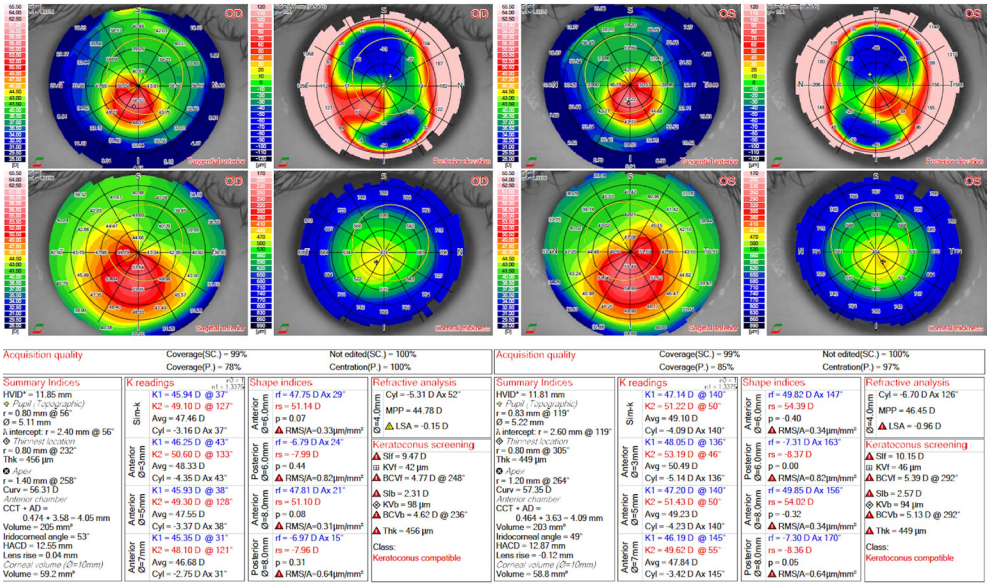

Figure 3. Corneal topographic maps showed significantly steep keratometry values and thinning in the inferior paracentral cornea bilaterally.

have a role in disease development. Contact lens usage, recurrent eye rubbing, and allergic eye disease are currently recognized environmental factors for KC (6). Despite a great amount of research, the pathogenesis of KC is poorly understood. KC has been associated with some systemic and ocular diseases. Individuals with Down syndrome have a greater prevalence of KC (7). KC is also associated with Leber congenital amaurosis, Brittle cornea disease, and connective tissue disorders (3,8).

Excess copper deposition in the Descemet membrane causes KF rings, which are a typical ophthalmologic sign in WD patients. KF rings are present in the majority of WD patients with neurologic impairment. There are correlations between KF at diagnosis and neuropsychiatric symptoms (9). Essential tremors and speech impairment are also present in our case.

Another ocular sign of WD is sunflower cataract which is a centralized, thin opacification that appears directly beneath the anterior capsule. Sunflower cataracts have a limited effect on patient visual acuity. In our case, there is no sunflower cataract (10).

Gharaee and Soleimani reported a 15-year-old female patient with KC and KF ring who was referred to neurology 2 years later because of hand tremors and diagnosed with WD (5).

Hu et al. reported a 19-year old male patient with KF ring combined with KC; the corneal morphology and thickness remained stable after 5 years of follow-up. The patient was given a low-copper diet, penicillamine for copper discharge, and zinc gluconate to inhibit copper absorption, and recent examination revealed that the KF ring had lightened in color. Because of the insufficient control of WD's disease and corneal topography values, we recommended corneal cross-linking to our patient, even though it had not been used in the other two reported cases in the literature.

Conclusion

There is not enough evidence about the effect of copper deposition in the Descemet membrane on corneal biomechanics and KC progression so far. The future case reports on the course of the disease and the efficacy of corneal cross-linking therapy in patients with KC combined with Wilson's disease are needed.

Disclosures

Informed consent: Written informed consent was obtained from the patient for the publication of the case report and the accompanying images.

Peer-review: Externally peer-reviewed.

Conflict of Interest: None declared.

Authorship Contributions: Concept – A.T., D.O., B.K.Y.; Design – B.K.Y., A.T., D.O.; Supervision – B.K.Y., D.O., A.T.; Resource – A.T., B.K.Y.; Materials – B.K.Y., A.T.; Data Collection and/or Processing – A.T., D.O.; Analysis and/or Interpretation – B.K.Y., A.T.; Literature Search – A.T., B.K.Y.; Writing – B.K.Y., A.T., D.O.; Critical Reviews – B.K.Y., A.T.

References

1. Robertson I. Keratoconus and the Ehlers-Danlos syndrome: A new aspect of keratoconus. Med J Aust 1975;1:571–3.[CrossRef]
2. Kristianslund O, Drolsum L. Prevalence of keratoconus in persons with Down syndrome: A review. BMJ Open Ophthalmol 2021;6:e000754. [CrossRef]

3. Elder MJ. Leber congenital amaurosis and its association with keratoconus and keratoglobus. J Pediatr Ophthalmol Strabismus 1994;31:38–40. [CrossRef]
4. Hu P, Lin L, Wu Z, Jin X, Ni H. Kayser-Fleischer ring with keratoconus: A coincidence? A case report. BMC Ophthalmol 2020;20:190. [CrossRef]
5. Gharaee H, Soleimani T. Double corneal ring: Report of simultaneous coincidence of kayser-fleischer ring and fleischer ring in a keratoconus patient prior to appearance of Wilson's diseaseno title. Iran J Ophthalmol 2012;24:57–62.
6. Rabinowitz YS. Keratoconus. Surv Ophthalmol 1998;42:297–319. [CrossRef]
7. Shapiro BL. Down syndrome and associated congenital malformations. J Neural Transm Suppl 2003;67:207–14. [CrossRef]
8. Abu A, Frydman M, Marek D, Pras E, Nir U, Reznik-Wolf H, et al. Deleterious mutations in the Zinc-Finger 469 gene cause brittle cornea syndrome. Am J Hum Genet 2008;82:1217–22.
9. Fenu M, Liggi M, Demelia E, Sorbello O, Civolani A, Demelia L. Kayser-Fleischer ring in Wilson's disease: A cohort study. Eur J Intern Med 2012;23:e150–6. [CrossRef]
10. Langwińska-Wośko E, Litwin T, Dzieżyc K, Członkowska A. The sunflower cataract in Wilson's disease: Pathognomonic sign or rare finding? Acta Neurol Belg 2016;116:325–8. [CrossRef]