CASE REPORT

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Bilateral keratoconus hydrops in a patient with Down Syndrome: a case report

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ABSTRACT

Background: Keratoconus has been reported in as many as 8% to 36% of people with Down syndrome. Corneal hydrops, a complication of advanced keratoconus, is characterized by the sudden onset of severe stromal and epithelial edema resulting from descemet membrane rupture. According to previous studies, acute corneal hydrops occurs in between 0.2% and 2.8% of keratoconus patients, while the incidence of corneal hydrops in Down syndrome patients with keratoconus is unclear. This study aims to evaluate the bilateral keratoconus hydrops in a patient with Down Syndrome.

Case Presentation: We reported a case of bilateral corneal hydrops in a Down syndrome patient aged 28. Upon arrival, his bilateral visual acuity was fixed to follow the object. Slit-lamp examination revealed bilateral corneal edema and apical corneal scarring, and grade 4 plus keratoconus were seen on the Scheimpflug camera system. The patient receives medical treatment to lessen the patient's symptoms while they wait for a cornea donor to do a transplant.

Conclusion: Patients with Down syndrome could experience bilateral corneal hydrops. Strong symptom communication, quick referral, and good cooperation during eye examinations are needed for early detection of this eye condition.

Keywords: corneal hydrops, down syndrome, genetics, keratoconus.
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INTRODUCTION

A non-inflammatory corneal ectatic condition known as keratoconus leads the cornea to steepen, along with gradual stromal thinning and loss of the best spectacle-corrected visual acuity.[1] Early estimates suggested that the prevalence of keratoconus was approximately 1 in 2000 individuals worldwide, while more recent research has estimated its frequency to be 1 in 350 individuals in the general population. Keratoconus has been reported in as many as 8% to 36% of people with Down syndrome.[2] Corneal hydrops, a complication of advanced keratoconus, is characterized by the sudden onset of severe stromal and epithelial edema resulting from descemet membrane rupture.[1] In patients with keratoconus, the incidence of acute corneal hydrops is between 0.2% and 2.8%.[3] The incidence of corneal hydrops in Down syndrome patients with keratoconus is unclear. According to the authors' knowledge and a database search of the literature, there aren't any case reports of bilateral hydrops in Downs syndrome patients at this date.

Given that the identification of this eye condition depends on adequate symptom communication, prompt referral, and good cooperation during eye examinations, it seems reasonable to suspect that there are a significant number of undiagnosed cases of keratoconus, especially in individuals with Down syndrome. Early keratoconus can be difficult to diagnose with a slit lamp alone, and Down Syndrome patients frequently have trouble cooperating with slit lamp examinations or corneal tomography.[4]

Ophthalmological examinations should be carried out at birth and periodically during life, ideally every 1-2 years, because individuals with Down's Syndrome are more likely to develop several ocular disorders such as keratoconus and amblyopia.[5] Based ion those mentioned above, this study aims to evaluate a rare case of Down syndrome patients with bilateral corneal hydrops.

CASE REPORT

A 28-year-old man who has whitened the center cornea of his eyes is present. The man has Down's syndrome. One and a half months ago, the patient's father also saw that his son seemed uncomfortable, with slight redness and tearing of both eyes. Although there are spots, it is no longer painful. The patient's father claims that his movements have slowed since then. Additionally, he was known to rub his eyes. Despite his limitations, the patient could still perform daily functions like eating and bathing before that stage. The patient received his formal education at a special-needs school.

Initial examination revealed small hands and feet, small ears, a short neck, a flattened face and a nasal bridge on the systemic side, as shown in Figure 1. On ocular examination, visual acuity was fixed to following the object with good eye movement in all directions. The intraocular pressure of the right eye was 21 mmHg, and that of the left was 11 mmHg. The palpebral fissure appears up-slanting; there is no injection of the conjunctiva. The right and left corneas appear edematous. The right eye appears like a pseudocyst due to fluid accumulation in the intrastromal spaces.

There were apical scars and Munson's sign on both eyes, but the Brushfield spots on the iris are not visible, as seen in Figure 2. On examination, the Scheimpflug camera system on the right eye K1 = 56.3 D, K2 = 58/5D, and Km = 57.3 D. The pachymetry vertex was 836 micrometers, as we can see in Figure 3. In contrast, left eye tomography was unavailable since the patient examination was challenging.

Topical hypertonic saline eye (5%) drops were first administered to him. The patient arrived 1.5 months after the initial episode, so the pressure bandage was not applied. The patient is awaiting a corneal donor to have Deep Anterior Lamellar Keratoplasty (DALK), or Penetrating Keratoplasty (PKP), first on the right eye and then on the left due to the lack of corneal donors in our nation.

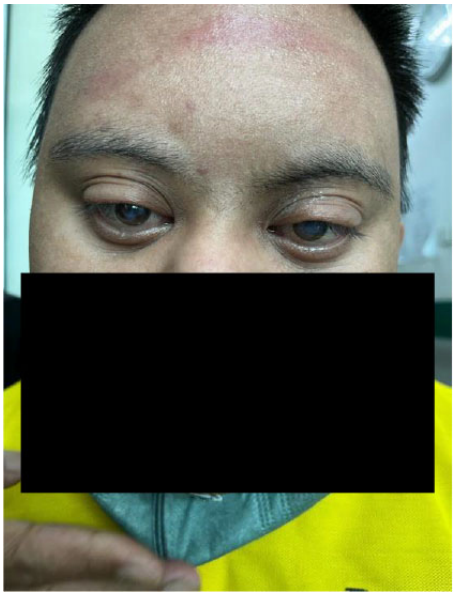

Figure 1. The facial characteristics show a slanting palpebral fissure and a flattened nasal bridge.

DISCUSSION

Multiple body systems are affected by the characteristics of the Down syndrome phenotype. Additionally, they have a higher risk of developing autoimmune diseases, hypothyroidism, and hearing and visual impairments. As is true for all human autosomal trisomies, advanced maternal age at conception is a major risk factor for trisomy 21. Due to the inherent difficulty in identifying each environmental element's exposure, dosage, and timing, non-disjunction risk is also influenced by environmental factors. However, these factors are challenging to define.[5]

Down syndrome has also been associated with numerous ophthalmologic manifestations, including patterns of strabismus, amblyopia, nystagmus, nasolacrimal duct obstruction, keratoconus, eyelid abnormalities, cataract, optic nerve abnormalities, glaucoma, retinal abnormalities. Previous research has shown that individuals with Down syndrome have different physical corneal characteristics from healthy people. These patients' corneas, in particular, are thinner and steeper than healthy individuals, which probably adds to the likelihood of keratoconus in this population. Furthermore, it has been proposed that individuals with Down syndrome are more prone to rubbing their eyes, which can lead to mechanical stress on the cornea. Relatedly, a small percentage of Down syndrome patients have been documented to have corneal hydrops, a keratoconus consequence in which Descemet's membrane and corneal endothelium abnormalities allow aqueous fluid to enter the stroma.[4]

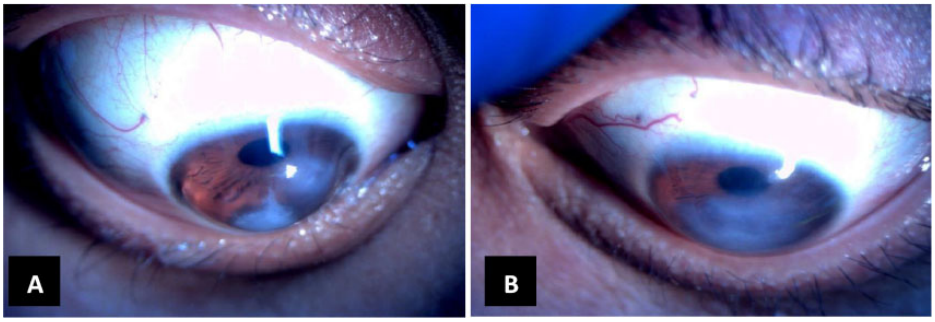

Figure 2. The cone-shaped cornea and bowing of the lower eyelid, with corneal edema and apical scarring on the right eye (A) and left eye (B).

About 2.6% of eyes with keratoconus have acute corneal hydrops, which causes acute stromal and epithelial edema due to rupture in the descemet membrane. Major symptoms include rapid onset of redness, photophobia, pain, and blurred vision. Although this self-limiting condition fades away within a few months, it can lead to serious complications such as perforation, scarring, poor vision, and corneal vascularization.[6] Early onset of keratoconus, Down syndrome, microtrauma from contact lens use, eye rubbing, allergic conjunctivitis, and atopy are risk factors for the development of corneal hydrops. There may be many undetected cases of unilateral or bilateral corneal hydrops in people with Down syndrome, given that detecting this eye condition requires strong symptom communication, quick referral, and good cooperation during eye examinations. Slit lamp examinations alone can make it challenging to diagnose early keratoconus, and people with Down syndrome commonly struggle to cooperate during these procedures.[7]

Before the cornea's center area became opaque, this patient had never undergone an eye exam, and the patient also rubbed his eyes frequently. The patient's parents claim that he suffered pain a month and a half ago. Although some of the stroma of the right eye's cornea is still edematous, the scarring in the apical region has already developed. This indicates that the acute corneal hydrops phase has subsided. At the time of the initial inspection, there were no eyelid spasms or red eyes with watering as at the time of the acute phase of corneal hydrops in keratoconus.[7,8]

Corneal cross-linking (CXL), the gold standard of treatment for keratoconus once diagnosed early, blocks corneal ectasia from progressing. In 2016, the United States Food and Drug Administration (U.S. FDA) authorized corneal cross-linking as a medication and device combination for treating progressive keratoconus and corneal ectasia after laser refractive surgery.[8] Corneal cross-linking in patients with Down syndrome has been the focus of a few case studies. The outcomes of CXL performed concurrently in both eyes while under general anesthesia are shown in two case reports: one by Koppen et

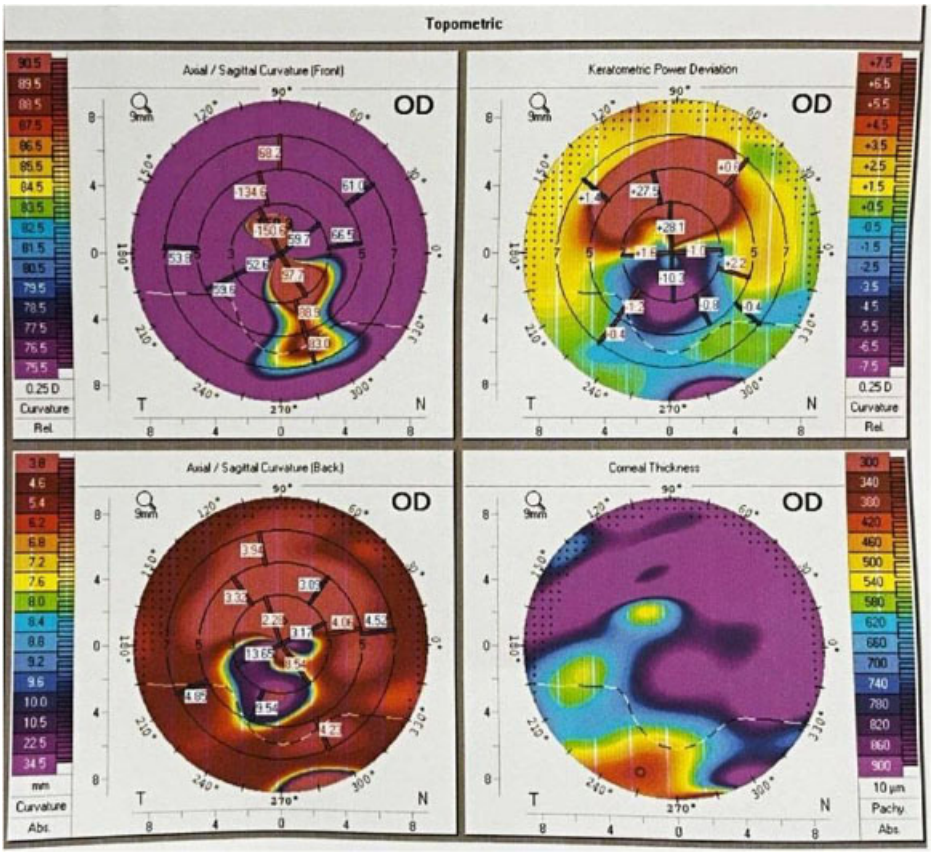

Figure 3. Corneal curvature map derived by Scheimpflug camera system, showing the axial/sagittal curvature, corneal thickness, and elevation showing keratoconus stage 4 and corneal thinning in the right eye.

al. A semi-structured decision tool was presented by Soeters et al. to assist the clinician in choosing Down syndrome patients for CXL therapy under local anesthesia. They evaluated whether doing CXL under local anesthesia was feasible for each patient. Alternatively, individuals who performed poorly on the pre-CXL exam can receive the therapy while under general anesthesia.[9]

Numerous treatments are available if a tear occurs in the Descemet's fold and a corneal hydrop is formed. The history and results from a slit lamp are typically used to make a diagnosis. The amount and degree of the edema and Descemet's membrane tear must be investigated to create a treatment plan, monitor the patient's reaction to the treatment, and spot any complications. These investigational techniques include optical coherence tomography of the anterior segment and ultrasound bio-microscopy. Medical treatment seeks to relieve symptoms up until a spontaneous resolution takes place. Topical lubricants, antibiotics to prevent secondary infection, cycloplegics to lessen pain and photophobia, hypertonic saline eye drops to help draw fluid, anti-glaucoma drugs to lessen the hydrodynamic force on the posterior cornea, topical steroids or nonsteroidal anti-inflammatory drugs are all included in this treatment. The best corrected visual acuity was discovered to be on par with surgical intervention in the final analysis. Sometimes a bandage contact lens may be recommended to relieve pain until the edema goes away or the patient is at ease.[10,11]

To reduce edema, this patient had a hypertonic solution. Since the patient wasn't feeling pain anymore and some of the corneas had already developed cicatricial tissue, bandage contact lenses were not prescribed for the patient. Bandage contact lenses can help with pain management during the acute phase, but extended use can also lead to hypoxia in the corneal epithelium and impair the patient's corneal ability to heal. Surgical intervention is required in cases of non-resolution corneal edema to avoid complications and for early visual rehabilitation. In addition to intracameral air injections of pneumodescemetopexy, compression sutures are used for acute corneal hydrops. It enabled corneal edema to fade quickly. It is a straightforward procedure that can be carried out with a very easy setup and doesn't require any specific gases like C3F8 or SF6.[7]

Patients with advanced keratoconus cannot achieve sufficient correction, and for such patients, keratoplasty (penetrating or lamellar) is used depending on the extent of corneal scarring. Approximately 12–20% of keratoconus patients will eventually need a corneal transplant. Keratoplasty in corneal hydrops patients provides good vision in the long term. Regardless of previous corneal hydrops, long-term allograft survival and visual results following penetrating keratoplasty in eyes with keratoconus are favorable. Endothelial rejection events are more frequent in eyes with resolved corneal hydrops. Despite excellent results with penetrating keratoplasty, deep anterior lamellar keratoplasty (DALK) may be performed for keratoconus patients with no risk of endothelial rejection to reduce the dose of steroids and the risk of secondary glaucoma and faster healing.[12,13]

This case report's limitation is that there aren't enough donors to carry out the best-recommended treatment option. Visual therapy will also be required to improve the patient's ability to do daily activities in the future, given that the patient falls within the low vision category.

CONCLUSIONS

The incidence of corneal hydrops in Down syndrome patients with keratoconus remains unclear. We found a rare case of Down syndrome patients with bilateral corneal hydrops. Strong symptom communication, quick referral, and good cooperation during eye examinations are needed for early detection of this eye condition.

CONFLICT OF INTERESTS

All authors declare no financial or conflicts of interest in this work.

ETHICAL CONSIDERATION

The patient's parents provided informed consent for the publication of this case report.

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None to declare.

AUTHOR CONTRIBUTION

All authors contributed to the study from the conceptual framework, data gathering, and analysis until the study's results were interpreted upon publication.

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