Published in final edited form as:

Cornea. 2016 March ; 35(3): 392–394. doi:10.1097/ICO.0000000000000722.

High-Resolution Optical Coherence Tomography Findings of Lisch Epithelial Corneal Dystrophy

Cameron Pole, BS¹, Adam Sise, MD¹, Madhura Joag, MD¹, Anat Galor, MD², Jose Antonio Bermudez-Magner, MD^{1,3}, Sander Dubovy, MD^{1,3}, and Carol L. Karp, MD¹

¹Bascom Palmer Eye Institute, University of Miami Miller School of Medicine, Miami, Florida, USA

²Department of Ophthalmology, Miami Veteran Affairs Medical Center

³Florida Lions Ocular Pathology Laboratory

Abstract

Purpose—To describe a case of Lisch epithelial corneal dystrophy (LECD) and present its unique characteristics on high-resolution optical coherence tomography (HR-OCT).

Methods—A 78-year-old man with whorled corneal epithelial opacities in the right eye was referred for evaluation of ocular surface squamous neoplasia. Clinical evaluation, photos, and HR-OCT images of involved cornea were obtained and scrapings of the affected cornea were sent for histopathologic analysis.

Results—Clinically the patient presented with a opalescent whirling epithelium in a linear pattern encroaching on the visual axis. HR-OCT showed normal thickness epithelial hyperreflectivity of involved cornea without stromal involvement, along with sharply demarcated borders of unaffected tissue. Histopathologic findings demonstrated vacuolated PAS-positive cells throughout the epithelial layers consistent with LECD.

Conclusions—HR-OCT was able to provide useful information to rule out ocular surface squamous neoplasia, and confirm the clinical impression of LECD at the time of clinical examination. HR-OCT shows promise as an adjunctive diagnostic tool for ocular surface lesions and pathologies.

Keywords

Lisch epithelial corneal dystrophy; OCT; optical coherence tomography; corneal dystrophy

Introduction

Lisch epithelial corneal dystrophy (LECD), also known as band-shaped and whorled microcystic dystrophy, is a corneal epithelial dystrophy first described in 1992 by Lisch et al¹ in five members of a single family. It was described on slit-lamp examination as “gray, band-shaped, and feathery opacities that sometimes appear in whorled patterns” presenting

Corresponding Author: Mailing Address: 900 NW 17th Street, Miami, FL, 33136; Phone: 305-326-6156; ckarp@med.miami.edu.

Conflict of Interest: All contributing authors have no financial or legal conflicts of interest to disclose.

both unilaterally and bilaterally. Further analysis and linkage studies have suggested an X-linked dominant inheritance pattern, mapping the gene to Xp22.3².

Histopathologic analysis of superficial scrapings from affected corneal epithelium demonstrates sheets of cells with a bubbly cytoplasm and vacuoles made of glycogen, prevalent in the suprabasilar cell layers^{1,3-7}. Slit-lamp examination reveals varying shapes of homogenous gray defects often in whorled and fimbriated patterns¹⁻⁹. A few small studies have investigated various diagnostic modalities for this dystrophy. In vivo confocal microscopy has shown well-demarcated regions of uniformly affected cells with hyporeflective nuclei and hyperreflective cytoplasm^{3,4,6,9}. Additionally, electron microscopy reveals the vacuoles to contain a laminated and clumped substance or whorled material^{3,4,7}.

High resolution optical coherence tomography (HR-OCT) has markedly improved our imaging capabilities of the anterior segment and ocular surface¹⁰. Technological advancements have allowed for precise visualization of corneal layers, tear films, and pathologies, with resolutions of up to 2 μm ^{10,11}. Although HR-OCT has been used to image tumors, benign lesions^{8,10,12}, and some dystrophies^{10,13}, this is the first known report of Lisch epithelial corneal dystrophy imaged with HR-OCT.

Case

A 78-year-old man presented to the ocular surface oncology clinic at the Bascom Palmer Eye Institute for evaluation of a corneal opacity in his right eye suspicious for ocular surface squamous neoplasia (OSSN). He reported that the lesion had advanced slowly since it was first observed 9 years earlier. He denied pain, photophobia, or any visual disturbances. His past medical history included diet-controlled diabetes mellitus, hypertension controlled with losartan, and gout. His surgical history was significant for cataract surgery and placement of bilateral posterior chamber intraocular lenses five years prior to presentation, as well as a history of two basal cell carcinomas excised from his scalp. The patient's son was noted to have a history of recurrent corneal erosions but it was not known if he had been diagnosed with an underlying condition (and was not available for examination).

Best corrected visual acuity was 20/20 in both eyes. Slit-lamp examination of the right eye showed a gray-white feathery streak opacity within the epithelium without noticeable elevation or depression of the corneal surface (Figure 1). There was no limbal involvement or corneal neovascularization. Both fluorescein and Rose Bengal staining were negative. The slit lamp examination of the left eye was normal, and no other ocular abnormalities were noted in either eye. HR-OCT images of the lesion done with the Rtxvue (Optovue, Fresno, CA) showed homogenous areas of hyperreflectivity within the epithelium with normal epithelial thickness throughout. The opacities created mild shadowing of the underlying corneal stroma. The remaining stroma and endothelium were within normal limits (Figure 2). Notably absent were duplication of the basement membrane, epithelial thickening, and stromal opacities. While the clinical and HR-OCT findings suggested a benign etiology, the patient was concerned about progression and requested biopsy. Following application of topical anesthesia, a biopsy was performed by scraping the lesion

with a #69 blade. Histopathologic evaluation disclosed epithelial tissue with foci of foamy PAS-positive epithelial cells (Figure 3) consistent with Lisch epithelial corneal dystrophy.

Discussion

The differential diagnosis of a corneal epithelial opacity includes OSSN, epithelial basement membrane dystrophy (EBMD), pannus, limbal stem cell deficiency, and other epithelial dystrophies. Lisch epithelial corneal dystrophy is a rare entity, with characteristic whorling pattern on clinical exam with distinct pathological findings that includes the presence of giant vacuoles replacing the epithelium.

Treatment of LECD lesions consists mainly of observation unless there is an associated change in vision or comfort. Alcohol-assisted epithelial debridement and contact lens placement have both been successfully attempted, with some degree of recurrence reported^{7,9}. Treatment using photorefractive keratectomy (PRK) combined with mitomycin C (MMC) application has also been described with minimal recurrence^{3,9}.

Our patient presented with the prior diagnosis of EBMD and possible OSSN. The patient was anxious to rule out the possibility of dysplasia. The lesion in this case had an opaque, whorled epithelial pattern. Familial patterns can be supportive of LECD as it is associated with an X-linked dominant inheritance. Unfortunately the patient had no siblings, and to his knowledge no other family members had this entity. By utilizing HR-OCT, we were able to identify key characteristics to rule out OSSN. Histopathologic analysis demonstrated features that supported the initial diagnosis of LECD.

LECD displayed a unique pattern on HR-OCT that correlated to findings on histopathology. Most notably there was homogenous hyper-reflectivity within the epithelium, without associated epithelial thickening. These lesions occurred in an intermittent pattern that corresponded to the patient's whorls seen on clinical exam. In addition, there was no involvement of the epithelial basement membrane to suggest EBMD, the stroma, or Descemet's membrane. Interestingly, the LECD lesion caused some shadowing not typically seen in corneal OSSN. The authors presume that the vacuolated material seen in LECD causes some additional blocking of the A scan not seen with OSSN.

HR-OCT is playing an increasing role in the evaluation of ocular surface lesions¹⁰. It has been particularly helpful in the diagnosis and management of OSSN^{12,14}, but various other pathologies have also been described^{8,13}. The appearance of LECD on HR-OCT from this case shows distinct characteristics from images of other epithelial lesions we have seen including EBMD, pannus, and spheroidal degeneration¹³. Elucidation of the findings seen on HR-OCT can help others with the future differentiation of this lesion.

While HR-OCT is emerging as a promising imaging modality, it does have some limitations. Any calcification or cellular characteristics may cause some shadowing of underlying structures. Image quality is affected by wavelength, resolution, and patient cooperation. In addition, limited availability and experience with the technology has restricted its use from standard practice. By describing the characteristics of LECD on HR-OCT and demonstrating

its unique features, we hope to add to the library of images available for this technology and demonstrate its usefulness as an adjunctive diagnostic tool.

Acknowledgments

Funding: Supported by the Department of Veterans Affairs, Veterans Health Administration, Office of Research and Development, Clinical Sciences Research and Development's Career Development Award CDA-2-024-10S (Dr. Galor), NIH Center Core Grant P30EY014801, Research to Prevent Blindness Unrestricted Grant, Department of Defense (DOD- Grant#W81XWH-09-1-0675 and Grant# W81XWH-13-1-0048 ONOVA) (institutional), The Florida Lions Eye Bank, The Ronald and Alicia Lepke Grant, The Lee and Claire Hager Grant, The Jimmy and Gaye Bryan Grant, The Gordon Charitable Trust, and the Richard Azar Family Grant (institutional grants).

References

1. Lisch W, Steuhl KP, Lisch C, et al. A new band-shaped and whorled microcystic dystrophy of the corneal epithelium. *Am J Ophthalmol*. 1992; 114:35–44. [PubMed: 1621784]
2. Lisch W, Buttner A, Oeffner F, et al. Lisch corneal dystrophy is genetically distinct from Meesmann corneal dystrophy and maps to xp22.3. *Am J Ophthalmol*. 2000; 130:461–468. [PubMed: 11024418]
3. Wessel MM, Sarkar JS, Jakobiec FA, et al. Treatment of lisch corneal dystrophy with photorefractive keratectomy and mitomycin C. *Cornea*. 2011; 30:481–485. [PubMed: 21045666]
4. Weiss JS, Moller HU, Aldave AJ, et al. IC3D classification of corneal dystrophies--edition 2. *Cornea*. 2015; 34:117–159. [PubMed: 25564336]
5. Robin SB, Epstein RJ, Kornmehl EW. Band-shaped, whorled microcystic corneal dystrophy. *Am J Ophthalmol*. 1994; 117:543–544. [PubMed: 8154546]
6. Kurbanyan K, Sejpal KD, Aldave AJ, Deng SX. In vivo confocal microscopic findings in Lisch corneal dystrophy. *Cornea*. 2012; 31:437–441. [PubMed: 22222997]
7. Charles NC, Young JA, Kumar A, et al. Band-shaped and whorled microcystic dystrophy of the corneal epithelium. *Ophthalmology*. 2000; 107:1761–1764. [PubMed: 10964841]
8. Medina CA, Plesec T, Singh AD. Optical coherence tomography imaging of ocular and periocular tumours. *Br J Ophthalmol*. 2014; 98(Suppl 2):ii40–ii46. [PubMed: 24599420]
9. Lisch W, Wasielica-Poslednik J, Lisch C, Saikia P, Pitz S. Contact lens-induced regression of Lisch epithelial corneal dystrophy. *Cornea*. 2010; 29:342–345. [PubMed: 20118787]
10. Wang J, Abou Shousha M, Perez VL, et al. Ultra-high resolution optical coherence tomography for imaging the anterior segment of the eye. *Ophthalmic Surg Lasers Imaging*. 2011; 42(Suppl):S15–S27. [PubMed: 21790108]
11. Wu S, Tao A, Jiang H, Xu Z, Perez V, Wang J. Vertical and horizontal corneal epithelial thickness profile using ultra-high resolution and long scan depth optical coherence tomography. *PloS One*. 2014; 9:e97962. [PubMed: 24844566]
12. Shousha MA, Karp CL, Canto AP, et al. Diagnosis of ocular surface lesions using ultra-high-resolution optical coherence tomography. *Ophthalmology*. 2013; 120:883–891. [PubMed: 23347984]
13. Vajzovic LM, Karp CL, Haft P, et al. Ultra high-resolution anterior segment optical coherence tomography in the evaluation of anterior corneal dystrophies and degenerations. *Ophthalmology*. 2011; 118:1291–1296. [PubMed: 21420175]
14. Thomas BJ, Galor A, Nanji AA, et al. Ultra high-resolution anterior segment optical coherence tomography in the diagnosis and management of ocular surface squamous neoplasia. *Ocul Surf*. 2014; 12:46–58. [PubMed: 24439046]

Figure 1.

A. Slit-lamp direct illumination of the cornea showing a grayish, whorled, epithelial opacification in a streak configuration. B. Slit-lamp indirect illumination of the same lesion, highlighting individual microcysts within the lesion.

Figure 2.
HR-OCT image of the lesion (*arrow*) showing normal thickness epithelium with hyperreflectivity and posterior shadowing (*arrowhead*). Upper left inset image shows the orientation of the cross-section.

Figure 3. Histopathology demonstrating foci of foamy, vacuolated cells (*arrow*). (Hematoxylin and eosin (H&E) stain, original magnification x60)