Ocular Rhinosporidiosis Presenting as Chronic Follicular Conjunctivitis in a Contact Lens Wearer

Rhinosporidiosis is a rare granulomatous disease of the mucosal membranes of the eye and nose caused by *Rhinosporidium seeberi*. Although commonly found in southern India and Sri Lanka, it is rare in North America. We report a case of chronic follicular conjunctivitis diagnosed by histopathological analysis, transmission electron microscopy, and polymerase chain reaction analysis to be ocular rhinosporidiosis in a young contact lens wearer from Louisiana.

**Report of a Case.** A healthy 15-year-old boy from Louisiana had chronic irritation in his left eye without discharge. His social history was remarkable for recent, frequent swimming in a bayou while wearing daily disposable contact lenses. His best-corrected visual acuity was 20/20 OU. Examination revealed a follicular conjunctivitis bilaterally. On his left palpebral conjunctiva, there were numerous pedunculated lesions with white spots on their surfaces (Figure 1A). Otherwise, his examination results were unremarkable. His contact lenses were unavailable for examination. Excisional biopsy of these lesions showed a chronic inflammatory infiltrate with circular periodic acid–Schiff–positive encapsulated lesions (Figure 1B-D) of varying sizes with central nuclear elements or spores, consistent with rhinosporidiosis. The patient stopped his contact lens wear, but the lesions recurred in his left conjunctiva in 11 months. Otolaryngological examination revealed no nasopharyngeal involvement. Another excisional biopsy of the left conjunctival lesions with cryotherapy of the excised margins and an incisional biopsy of the right palpebral conjunctiva were performed. Only the left conjunctiva showed encapsulated lesions. Conjunctival cultures were negative. Transmission electron microscopy of the medium-sized sporangia (Figure 2A) showed a cell wall with radiating filaments (Figure 2B) encapsulating globules of lipidlike material (Figure 2A) around a single nucleus. Polymerase chain reaction analysis of the tissue with genus-specific *R seeberi* primers correctly amplified a 400–base pair product (Figure 2C). The lesions recurred in the left eye after 3 months, and another biopsy revealed *R seeberi*.

**Comment.** First described as a fungal entity, *R seeberi* is now classified in its own class, Mesomycetozoea. It undergoes a life cycle from trophocyte to mature sporangium, which produces endospores that are the infective units. Spores are transmitted from contaminated water and dust through epithelial defects on mucosal surfaces. The patient’s use of soft contact lenses while swimming likely produced epithelial defects for transmission. Infection causes formation of highly vascular polyps most commonly found in the nasal mucosa and the conjunctiva. Ocular involvement occurs in 15% of cases, mostly in the conjunctiva, but dacryocystitis and scleral melting have been reported. Involvement of other mucosal membranes, skin, and internal organs has been reported. The polyps are covered with white spherules representing the sporangia, described as resembling a strawberry. In this patient the follicular conjunctivitis developed after exposure to the bayou water, and the presence of these strawberry lesions differentiated his condition from other inflammatory conditions such as giant papillary conjunctivitis commonly seen in contact lens wearers. Histopathologically, there is a fibromyxomatous stroma containing trophocytes and sporangia in all stages of development. Treatment involves excision with cryotherapy, but recurrences are common and medical therapy such as with dapsone remains controversial. In this patient, the follicular conjunctivitis recurred despite cessation of contact lens wear, also differentiating this condition from other contact lens–related inflammation. Culturing specimens usually does not yield growth of this organism and histopathological analysis has been the mainstay of diagnosis. This report shows identification with histopathological analysis, transmission electron microscopy, and polymerase chain reaction analysis.
Interestingly, sequence data of polymerase chain reaction from this organism showed this isolate to be more similar to *R. seeberi* from a canine sample than that of other human isolates. This patient did not have any dogs at home. As such, these data suggest that the genus *Rhinosporidium* may possess isolates capable of infecting multiple host types.

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**Novel Compound Heterozygous Mutations in CERKL Cause Autosomal Recessive Retinitis Pigmentosa in a Nonconsanguineous Chinese Family**

Retinitis pigmentosa (RP) (OMIM 268000) is characterized by night blindness, progressive constriction of the visual fields, and fundus changes, including bony spicule pigmentation. To date, a number of RP loci or genes have been reported. One of the autosomal recessive RP (arRP) loci, RP26, was mapped to chromosome 2q31-q33 in 1998. The disease-causing gene at this locus has been recently identified as the *CERKL* gene (ceramide kinase–like) (GenBank NM_201548), encoding a 532–amino acid protein that shares 29% identity with ceramide kinase. Only 3 *CERKL* mutations have been reported, including p.E257X in Spanish families. In a consanguineous Pakistani family, and a splicing mutation in Yemeni Jewish families. We now report 2 novel compound heterozygous mutations in *CERKL*, c.156_157insT and c.758delT, which were found in a nonconsanguineous Chinese family with arRP. Our data indicate that compound heterozygous mutations of *CERKL* can cause RP.

**Methods.** A family with arRP was enrolled in a study in China and diagnosed by clinical and ophthalmological examinations. Peripheral blood was collected and genomic DNA was isolated. Linkage and haplotype analyses were carried out with microsatellite markers flanking 21 known arRP loci, including RP26. After the pathogenic gene was mapped to chromosome 2q31-q33, all 13 exons of *CERKL* were sequenced using both forward and reverse polymerase chain reaction primers as described. To discern 2 mutant alleles in the proband, polymerase chain reaction products of exon 1 and exon 5 of *CERKL* were cloned using the pGEM-T Easy Vector System (Promega Corp, Madison, Wisconsin) and multiple clones were sequenced.

**Results.** The nonconsanguineous Chinese family with arRP includes 5 siblings. Both parents and their 2 daughters do not show any RP symptoms, but 3 sons are affected with RP (Figure 1). The proband (III:3) had night blindness at age 18 years and progressively lost visual acuity. Funduscopic examinations revealed attenuation of the retinal arteries and bony spicule pigmentation in the midperipheral retina but normal color of optic discs. A bull’s-eye–like appearance in the macular region was observed (Figure 1). Family members

![Figure 1](image-url)