

Ocular manifestations in lipid proteinosis: A rare clinical entity

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Lipoid proteinosis is a rare autosomal recessive genodermatosis with abnormal lipid protein complexes deposition in different parts of the body, especially in the skin and mucus membranes of the upper aerodigestive tract. Though ocular involvement in lipoid proteinosis is rare, ophthalmologists may encounter diverse ocular complications accompanying this syndrome in clinical practice. We describe a case of lipoid proteinosis involving bilateral eyelids with pathognomonic moniliform blepharosis in a 33-year-old gentleman who presented with the complaints of itching of eye lids on and off since 10 years.

Key words: Lipoid proteinosis, moniliform blepharosis, Urbach–Wiethe syndrome

Lipoid proteinosis, also known as Urbach–Wiethe syndrome or hyalinosus cutis et mucosae, first described by a dermatologist

Urbach and an otolaryngologist Wiethe in 1929. It is a rare autosomal recessive multisystem disorder with dermatological, otorhinolaryngological, ocular, and neurological manifestations due to mutation of extracellular-matrix protein-1 gene located on chromosome 1q21.^[1] It is characterized by the deposition of an amorphous hyaline material with a glycoprotein constitution in various tissues in the body mainly the larynx, skin, and tongue.^[2] Though this entity has been well known to dermatologists and otorhinolaryngologists, few reports have appeared in the ophthalmic literature with the eyelids involvement in the majority of the cases.^[2,3] We describe a case of lipoid proteinosis involving bilateral eyelids with pathognomonic moniliform blepharosis in a 33-year-old gentleman who presented with the complaints of itching of eyelids on and off since 10 years.

Case Report

This study conforms to the principles outlined in the Declaration of Helsinki and was conducted after obtaining approval from the Institutional Ethics Committee on Human Research and informed written consent from the patient.

A 33-year-old gentleman presented to our outpatient department with complaints of itching of eyelids in both eyes on and off since 10 years. On ophthalmic examination, unaided visual acuity was 20/20 in both eyes, anterior segment examination revealed rows of yellow-white color, round or oval, bead-like excrescences on the margins of all four lids and the caruncle, resembling a string of pearls, the pathognomonic,

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moniliform blepharosis [Figs. 1 and 2]. The lesions were continuous, overlying the entire lid margin and obscuring the details of the lid margin with thickening of the free ends of the lids considerably. Rest of the anterior segment examination was otherwise normal including intraocular pressure measurement. Fundus evaluation of both eyes was within normal limits. Systemic examination revealed thickened and hard tongue with teeth indentations [Fig. 3], hoarseness of voice, multiple small yellow-colored soft and partially translucent nodules over the skin in bilateral axilla [Fig. 4], and normal neurological and psychiatric evaluation. Otorhynolaryngologist's evaluation for hoarseness of voice showed thickening and irregularities of the vocal cords' mucosa on laryngoscopy. Similar clinical features including pathognomonic moniliform blepharosis and hoarseness of voice were noted in his sibling who was asymptomatic. Baseline laboratory data including biochemical and hematological parameters were within normal limits. Biopsy of the axillary skin lesions performed by a dermatologist was consistent with the histopathological diagnosis of lipid proteinosis. The patient has been managed with artificial tear supplements and antihistamine eye drops with the symptomatic improvement of ocular manifestations within 4 weeks' time. He has been further advised and counseled regarding the chronic nature of the disease,

other ocular manifestations, multisystem involvement, and the need for follow-up under a multidisciplinary team of specialists including ophthalmologist, dentist, dermatologist, otorhynolaryngologist, and neurologist.

Discussion

Lipoid proteinosis usually starts in infancy or early childhood due to the involvement of upper aerodigestive tract mucosa with hoarseness of voice as presenting feature, may be accompanied by swelling of the tongue and lips, with associated difficulty in swallowing and respiratory distress in some cases.^[4] Next, in sequence occurs a skin rash, involving the face, scalp, and trunk with a thickened, pale yellow-brown, pock marked appearance of the lesions.^[5] It may also involve axilla as seen in our case or the extensor surfaces (elbows and knees) with marked hyperkeratosis in response to minor trauma. Neurological manifestations include longstanding memory impairment, paranoia, rage attacks, mental retardation, and temporal lobe epilepsy.^[6]

Though ocular manifestations in lipid proteinosis are rare, they were described since early descriptions of the disease. Along with moniliform blepharosis as a pathognomonic feature



Figure 1: Lipoid proteinosis involving eyelids

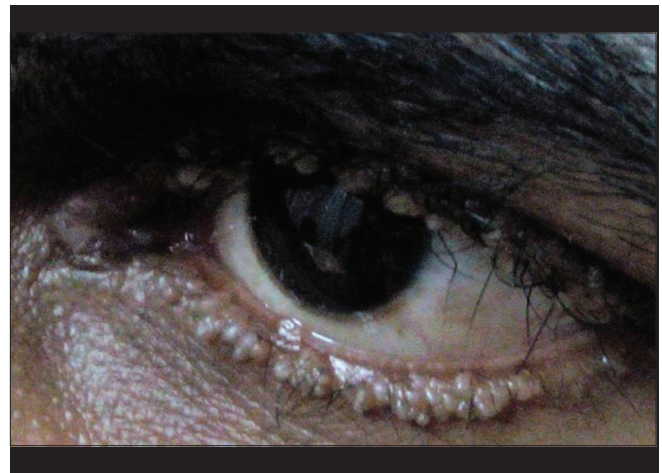


Figure 2: Lipoid proteinosis involving eyelids showing pathognomonic moniliform blepharosis

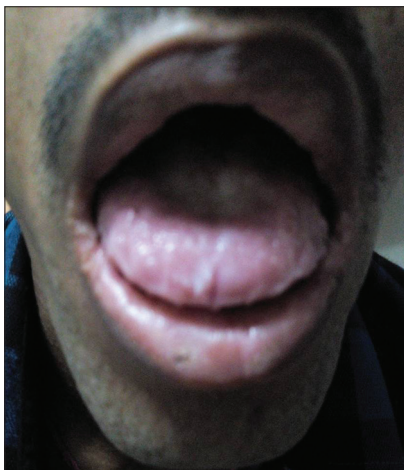


Figure 3: Lipoid proteinosis involving oral mucosa and tongue

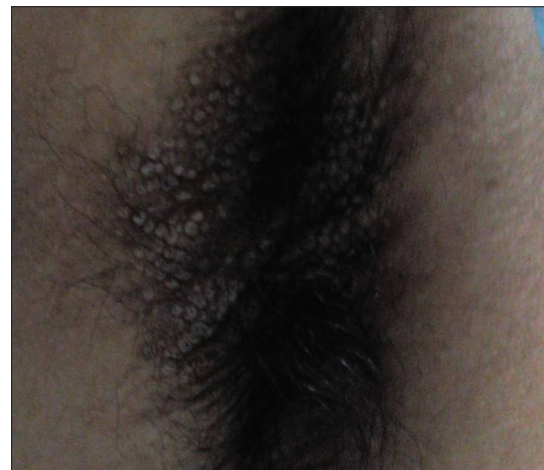


Figure 4: Lipoid proteinosis involving axillary skin

of the disease as seen in our case, an ophthalmologist may encounter other manifestations of lipoid proteinosis in any part of the eye such as cornea, conjunctiva, sclera, trabecular meshwork, iris/pupil, lens and zonular fibers, retina, and nasolacrimal duct. In general, the most common ocular lesions include eyelid lesions, as mentioned before; classic presentation of such lesions is called moniliform blepharosis, which is generally believed to be one of the most pathognomonic features of lipoid proteinosis.^[7] This hallmark presents as tiny papules on eyelid margins such as a string of yellowish and waxy beads [Figs. 1 and 2] as seen in our case and is particularly known as a strong diagnostic clue. Apart from the diagnostic value, lid lesions are reported to accompany infiltration of glands of Zeiss, Moll, and Meibomian and consequently cause madarosis, trichiasis, and sometimes distichiasis. Another frequent finding is focal degeneration of macula and drusen formation in Bruch's membrane in about 30–50% patients.^[7]

Uncommon ocular manifestations of lipoid proteinosis^[7] include glaucoma (either due to deposition of hyaline inclusions in the trabecular meshwork or due to hyalinization of scleral trabeculum with deposition of glycoproteins), lens-related complications (cataract or lens dislocation or subluxation),^[8] retinal complications (the concomitancy of retinitis pigmentosa, impaired color vision, and light hypersensitivity), corneal manifestations (corneal ulceration caused by trichiasis, corneal opacities, keratoconus^[9] as well as the deposition of hyaline on the cornea, especially at Descemet's membrane), iris- and pupil-related complications, unilateral or bilateral uveitis, dry eye or epiphora, nasolacrimal duct obstruction, and transient blindness.

As lipoid proteinosis runs a chronic, benign but progressive course, no effective treatment is known. In most of the cases, management is guided toward the type of ocular manifestation with symptomatic improvement in clinical condition. As in our case, eyelid lesions are managed with artificial tear supplements and antihistamine eye drops and may involve surgical removal and CO₂ laser therapy in some cases.^[10] Patients have been further advised and counseled regarding the chronic nature of the disease, diverse ocular manifestations, multisystem involvement, and the need for follow-up under a multidisciplinary team of specialists including ophthalmologist, dentist, dermatologist, otorhinolaryngologist, and neurologist.

Conclusion

Though ocular manifestations in lipoid proteinosis are rare, ophthalmologists may encounter diverse ocular complications as mentioned above, leading them for a more effective role in diagnosing and managing individuals with this disease as a part of a multidisciplinary team of physicians.

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Conflicts of interest

There are no conflicts of interest.

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