Lipoid proteinosis: A rare entity

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Urbach–Wiethe syndrome or lipoid proteinosis is a rare autosomal recessive disorder characterized histologically by infiltration of Periodic acid Schiff-positive hyaline material in the skin, upper aerodigestive tract, eyelids, and internal organs. Classical clinical features include scarring of the skin, beaded eyelid papules (moniliform blepharosis) and laryngeal infiltration leading to hoarseness of voice. Lipoid proteinosis can lead to life-threatening conditions such as acute respiratory distress and seizures. Awareness among ophthalmologists about this rare entity is crucial for appropriate management of these patients.

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

Urbach–Wiethe syndrome (UWS), also known as lipoid proteinosis or hyalinosis cutis et mucosae is caused by abnormal deposition of Periodic acid Schiff (PAS) positive hyaline material in the skin, mucous membranes, and internal organs. Beaded papules (moniliform blepharosis) and hoarseness of the voice are the striking features of the disease. These are sometimes mistaken for leukoplakia. We report this case of biopsy proven lipoid proteinosis for its rarity and the absence of central nervous system involvement.

Case Report

A 32-year-old male came to us with complaints of cystic lesions in both lids [Figs. 1, 2a and b]. He was diagnosed to have lipoid proteinosis elsewhere and was completely screened for systemic involvement. His urine electrophoresis showed elevated albumin, serum uric acid, and alanine transaminase. Computed tomography brain was normal. A buccal mucosal biopsy taken elsewhere showed deposition of PAS-positive hyaline material [Fig. 3]. Slides were reviewed in our center, and the diagnosis was reconfirmed. His best corrected visual acuity in both eyes was 20/20; n6. There were verrucous lesions in both upper and lower lids with cauliflower shaped lesion in the lower puncta of both eyes. He had cutaneous lesions also. Fundus examination was within normal limits. Schirmer's in both eyes were within normal limits. He had tongue fissures and hoarseness of voice. He was advised to undergo excision of the lesion over the puncta. The patient was lost to follow-up after that.

Discussion

UWS is an autosomal recessive disorder,[1] first described by Urbach and Wiethe in 1929. This disorder is extremely rare. So far, not more than 300 patients have been diagnosed with UWS.[2,3] The incidence of hyalinosis cutis et mucosae seems to be fairly high in South Africa.[4] It is a multi-system disease caused due to mutations in the gene encoding extracellular matrix protein 1 on chromosome 1q21.[5,6]

It is mostly an incidental diagnosis. High clinical suspicion with tissue biopsy gives clue to the diagnosis. We did not find any specific criteria for diagnosis in the literature.

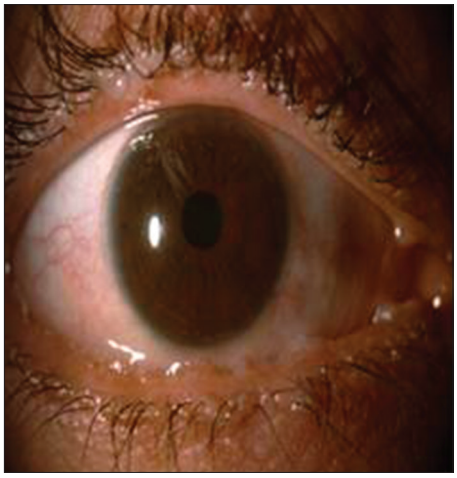

Figure 1: External photograph of the eye

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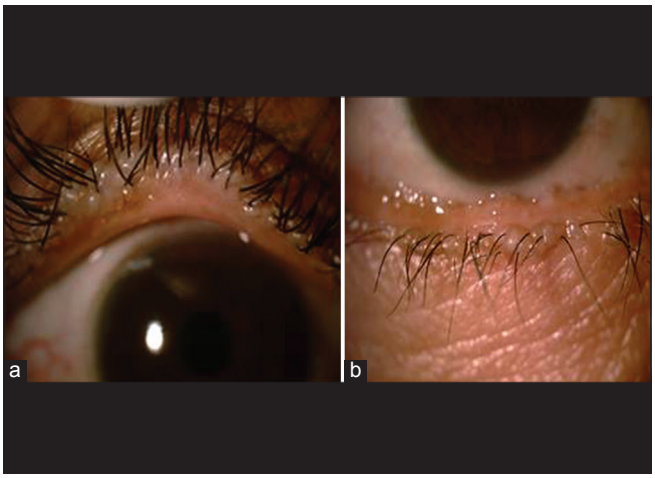

Figure 2: External photograph closer view (a) upper lid (b) lower lid

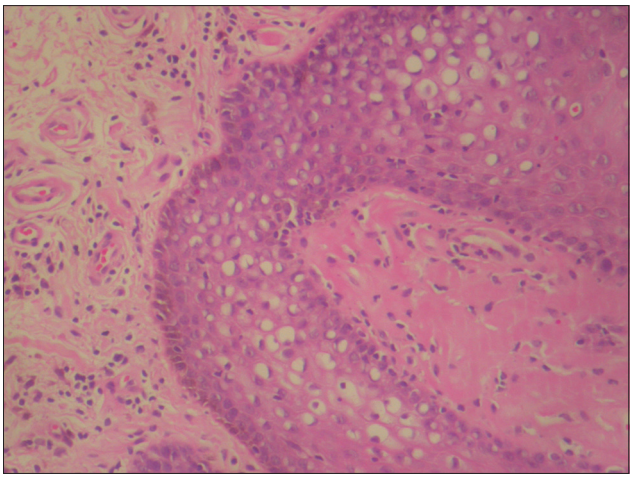

Figure 3: Histopathology picture depicting PAS positive hyaline material

Hoarseness of voice is reported to be the first manifestation of lipoid proteinosis. It was the first presentation in our patient also. Our patient had tongue fissures. Skin lesions generally appear as nodules on the face, lips (at earlier stages), and later become hyperkeratotic.[7]

The lacrimal gland can be infiltrated with hyaline material and can cause dry eyes. Schirmer's test in both eyes was normal suggesting no involvement of lacrimal gland in our case. Various ocular manifestations such as dry eyes, open angle glaucoma, drusen in the macula, retinitis pigmentosa, uveitis and subluxation of the lens has been reported along with lipoid proteinosis.[7] However, our patient had no abnormality in his eyes.

The most common radiological hallmark is the presence of bean or comma shaped intracranial calcifications in the temporal lobes in the amygdala, which is more evident in the patients who have lipoid proteinosis for a long duration.[8] Patients with neurological manifestations present with a migraine, seizures, mental retardation, anxiety, depression, and panic attacks. Our patient did not have any neurological manifestations.[9]

This disease can diminish the quality of life. It requires a multidisciplinary approach. There is no permanent cure for lipoid proteinosis. Medical treatment for the skin lesions has been reported previously by several authors.[10] Callizo *et al*. have suggested better results with surgical removal of the eyelid lesions. However, the majority of those treatments were based on single case reports.[11]

Conclusion

Though rare, systemic manifestations of lipoid proteinosis include life-threatening situations like acute respiratory distress or seizures. Awareness among ophthalmologists about this rare entity is crucial for appropriate management of these patients.

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Conflicts of interest

There are no conflicts of interest.

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