Neurothekeoma of the eyelid: A case report

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The purpose of this article is to describe a rare benign tumor of nerve sheath origin arising from the eyelid in an elderly male. Local excision was done and histopathological examination revealed a neurothekeoma. Six months later the patient was doing well with no recurrence. The case was unique in that the patient was an elderly male while neurothekeoma is commonly seen on the face of young adults, especially females.

Key words: Eyelid, neurothekeoma, tumor

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Neurothekeoma is a rare benign soft tissue neoplasm of probable nerve sheath origin. They usually occur on the face and extremities of young adults and show a female preponderance. Only six cases of neurothekeoma have been reported till date. We present here a case of neurothekeoma on the eyelid in an elderly male.

Case Report

A 66-year-old man presented to the out patient department with a painless swelling on the medial half of the right lower lid. It was of insidious onset, gradually increasing in size over the past eight years. Fine needle aspiration cytological examination of the lesion yielded only blood and diagnosis could not be established. He was a known diabetic and hypertensive, on treatment for the past eight years.

Examination of the lids and adnexa of the right eye revealed a single, soft, non-tender swelling measuring 2x1.9x1 cm [Fig. 1] with a lobulated surface, arising from the medial half of the lower lid. The lower lid margin along with the punctum was everted. The skin over the swelling showed prominent dilated veins. The palpebral conjunctiva in the region of the swelling showed keratinisation. Lid closure was adequate. No regional lymph nodes were palpable. Rest of the ocular examination was normal. The left eye was normal. The patient had watering of the eyes due to the exposure. The mass was causing a cosmetic deformity and so microsurgical excision with reconstruction of the lower lid was planned under local anesthesia.

A pentagon full-thickness resection of the tumor mass with microscopically clear margins on all sides was done and...
the tissue sent for histopathology examination. The defect created by this resection was a little less than half of the eyelid. Since the lower lid tissues were already stretched by the presence of the tumor, lid repair was possible with lateral cantholysis alone. The lid defect was closed in layers using 6-0 polyglactin sutures and the lids were apposed using a frost suture. Routine postoperative care was given to the patient and the frost sutures were removed after 48 h. The patient on serial follow-up was normal at six months with no evidence of recurrence.

The resected mass was a multilobulated pale bluish lesion [Fig. 2] covered by thin, stretched skin. The lesion was dissected by fibrous septae [Fig. 3A] of varying thickness into nodular masses. No adnexal structures of the eyelid were entrapped in the mass. The lesion showed spindle-shaped and stellate cells [Fig. 3B] arranged in small fascicles in an alcian blue positive myxoid stroma. In each nodule, fine wavy bundles and swirling whorls of collagen (highlighted by reticulin silver stain) enclosing the elongated Schwann cells and mast cells were observed. The spindle and stellate cells expressed S-100 protein [Fig. 3, inset] confirming its nerve sheath origin. Immunostain for neurofilament revealed occasional clusters of axons while the cells in the lesion were not immunolabeled for antibodies to desmin and cytokeratin. No ganglionic cells were found and the mitotic activity was very low. The features were characteristic of neurothekeoma-myxoid variant. The presence of axons and absence of entrapment of adnexal structures with S-100 positivity of the cells differentiated this lesion from superficial angiomyxoma which was a very close histological differential diagnosis.

**Discussion**

Myxoid neurothekeoma or nerve sheath myxoma is a rare benign neoplasm of putative peripheral nerve sheath origin first described in 1969 by Harkin and Reed and subsequently termed ‘neurothekeoma’ by Gallager and Helwig in 1980.

It often affects the skin with predilection for the face and extremities of young adults. Females seem to be more often affected than males. They are often solitary and may be either asymptomatic or may present as a painful, raised, skin-colored, well-circumscribed dome-shaped nodule less than 3 cm in diameter. The tumor occurs in a myxoid or classic variant, cellular variant and an intermediate (mixed) variant, the myxoid variant being relatively common. This lesion forms an important differential diagnosis for myxoid soft tissue tumors, especially the benign ones. Although excision is the mode of treatment for these benign myxoid tumors, proper histopathological typing is needed as some of these tumors like superficial angiomyxoma show tendencies towards local recurrence and may be associated with syndrome complex like association of superficial angiomyxoma with Carney complex, an autosomal dominant syndrome complex comprising myxomas of heart and skin, hyperpigmentation of the skin (lentiginosis) and endocrine overactivity.

The myxoid neurothekeoma lesions are consistently positive for S-100 proteins. Hence S-100 is considered to be a sensitive immunohistochemical marker for neurothekeoma. They are also known to be positive for vimentin but negative for cytokeratin,
Neurotrophic keratitis in a patient with dihydroxypyrimidine dehydrogenase deficiency

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We describe a case of neurotrophic keratitis in association with dihydroxypyrimidine dehydrogenase (DHPD) deficiency. Ocular manifestations in patients with DHPD are rare and neurotrophic keratitis has never been reported before. A six-year-old boy who was a known case of DHPD deficiency and born of a consanguineous marriage presented to our clinic with non-healing corneal ulcers in both eyes. Reduced corneal sensations were detected and the patient was started on lubricating eye drops. The patient continues to be on lubricant eye drops and there has been no recurrence of the disease.

Key words: Dihydroxypyrimidine dehydrogenase deficiency, neurotrophic keratitis, pyrimidine metabolism disorders

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Dihydroxypyrimidine dehydrogenase (DHPD) deficiency is a rare metabolic disease. The patient usually presents in early infancy with features of cerebral dysfunction.1 Though other ocular manifestations have been reported, to our knowledge corneal involvement has not been documented in the past.2 This is the first case report of DHPD deficiency with corneal involvement.

Case History

An 18-day-old baby with failure to thrive was referred to our clinic for eye examination. The patient was born of a consanguineous marriage. His parents were first cousins. At birth he was lethargic with poor cry and feeble suck. He had bilateral hydrocele and large anterior fontanelle. He was investigated for possible metabolic diseases. Blood investigations included total and differential blood count, blood sugar, serum electrolytes, serum ammonia, serum lactate, serum pyruvate, liver enzymes, thyroid function tests and arterial blood gases. Urine was tested for pH, ketones, odor and reducing substances. Karyotyping with G-banding studies was also performed. Though all above investigations were within normal range urine gas chromatography showed increased thiamine and uracil levels. Fibroblast cultures indicated deletion of the DHPD gene and diagnosis of DHPD deficiency was made. His elder brother was homozygous for the mutation and excreted increased amount of thiamine and uracil in urine; however, he was clinically asymptomatic. His eldest sister has two normal alleles but his mother was heterozygous for the condition.

Ocular assessment at that time did not detect any pathology. On subsequent follow-up his vision was found to be reduced to 20/100 (Cardiff Acuity Cards). Ophthalmic examination under general anesthesia was essentially normal apart from astigmatism in both eyes. Glasses were prescribed with orthoptic follow-up. At the age of six years his visual acuities deteriorated to 20/200 and 20/120 (Sheridan Gardner) in the right and left eye respectively. Bilateral central