Double trouble: entropion repair complicated by a conjunctival neurofibroma

Pooja Sethi¹, Son T. Ho¹, Hillary Z Kimbrell², Alejandra A. Valenzuela³

¹. Department of Ophthalmology, Tulane University Health Sciences Center, New Orleans, Louisiana, USA
². Department of Pathology and Laboratory Medicine, Tulane University Health Sciences Center, New Orleans, Louisiana, USA
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Abstract
While ocular involvement is well documented in association with neurofibromatosis type 1 (NF-1), conjunctival neurofibromas are very rare. We describe a challenging NF-1 case of a patient with a conjunctival neurofibroma in association with lower lid involutional entropion and severe conjunctival chemosis.

A 65-year-old man presented with age-related left lower lid laxity, retractor dehiscence, inferior conjunctival chemosis and significant ocular discomfort. The patient underwent a left lower lid lateral tarsal strip procedure with retractor plication without much improvement in the chemosis and discomfort. However post-operatively it became evident that an injected mass was present in the left inferior fornix. Surgical debulking of this mass allowed for resolution of the patient’s signs and symptoms as well as proper lower lid alignment. Histopathologic diagnosis with S-100 and CD34 positivity confirmed a conjunctival neurofibroma. Although uncommon, it is important to consider the possibility of a conjunctival mass in a patient with persistent chemosis and recurrent lower lid instability, particularly when confronted with a neurocutaneous syndrome.

Key words: neurofibroma; conjunctiva; entropion; neurofibromatosis; chemosis.

Case Presentation
A 65-year-old man with NF-1 and history of poor vision in his left eye, secondary to previous trauma, presented to the orbital and oculoplastic service complaining of burning and tearing of his left eye associated with rare mucus discharge and foreign body sensation that had slowly worsened over many months. On exam, visual acuity was 20/70 in the right and 20/200 on the left with normal optic nerve function. Of note, the patient had a left traumatic mydriasis with inferior corneal opacity, bilateral and multiple Lisch nodules, left angle recession, and bilateral nuclear cataracts. He also presented with bilateral deep superior sulci bilaterally and a left lower lid involutional entropion with vertical and horizontal laxity (Figure 1a), making it difficult to restore this eyelid into its anatomical position. Extra-ocular motility was significant for restriction of left down gaze with mild left hypertropia in primary gaze.

The left inferior conjunctiva was significantly injected with chemosis present towards the inferior fornix (Figure 1b). A thorough physical exam revealed numerous dermal neurofibromas of the face, trunk, and extremities along with multiple large café-au-lait spots. Computerized tomography imaging of the brain and orbits did not reveal any other abnormalities.

The patient underwent left lower lid entropion repair with a lateral tarsal strip and lower lid retractor plication. Post-operatively the lower lid showed improvement in position, but it was still not properly apposed to the globe. Conjunctival chemosis persisted, however, a slight improvement in chemosis allowed for visualization of a firm, non-mobile, painless inferior bulbar mass approximately 1.5x1.0cm. Excisional biopsy was performed and histopathology

Figure 1a-b: Clinical photographs of a male with extensive left inferior bulbar conjunctival chemosis and left lower lid entropion.
demonstrated a collagenous and fibrillary matrix on H&E stain (Figure 2a-b), and S-100 and CD34 positivity without SMA staining on immunohistochemistry (Figure 3). Neurofilament marker was focally positive, confirming the diagnosis of a diffuse isolated bulbar conjunctival neurofibroma (Figure 4). The patient’s lid position improved immediately after the debulking and his symptoms and chemosis resolved.

Discussion
Neurofibromatosis type I (NF-1) is an autosomal-dominant multi-system phakomatosis with complete penetrance but variable expressivity. Patients with this entity are encountered in ophthalmology with findings in the eyes and ocular adnexa including: café-au-lait spots, neurofibromas, glaucoma, optic nerve gliomas, Lisch nodules, choroidal hamartomas, retinal astrocytomas, and dysplasia of the greater wing of the sphenoid. Neurofibromas, the hallmark feature of NF-1, are benign peripheral nerve sheath tumors derived from Schwann cells. Although neurofibromas are benign histologically, they are cosmetically disfiguring and may compromise the function of nearby structures. Three types of neurofibromas occur around the eye or in the orbit: solitary, diffuse, or plexiform with the latter typically associated with NF-1. The solitary variant of conjunctival neurofibromas are associated with neurofibromatosis type 2b and are associated with prominent corneal nerves in 100% of cases.

While the presence of neurofibromas is well documented in the orbit, they have only infrequently been reported to occur in the caruncle and episclera. Conjunctival neurofibromas are soft mucosal neural tumors occurring in the conjunctival stroma and have rarely been described. Kalina et al describe only 13 cases of isolated neurofibromas of the conjunctiva dating from 1898 and of these cases 10 had systemic neurofibromatosis. A thorough history and physical examination is prompted in any patient with an isolated neurofibroma without a diagnosis of NF-1.

The challenge in this case was diagnosing a conjunctival neurofibroma in the setting of significant chemosis. Chemosis is a well-documented presentation of peri-ocular malignant metastases such as breast cancer, adenocarcinoma, lymphoma and multiple myeloma; however, in most instances it occurs with mechanical irritation of
the bulbar conjunctiva. Conjunctival neurofibromas cause a plaque-like elevation of the overlying conjunctiva which is a nidus for irritation. We speculate that the chronic irritation caused by the conjunctival neurofibroma may have incited a spastic entropion phenomenon superimposed with his age related lower lid laxity, ultimately leading him to seek medical attention.

The treatments described for symptomatic neurofibromas of the conjunctiva include surgical excision of the tumor and eyelid repositioning surgery. Prior to intervention, it is important to define the extent of the lesion and to know the presence of orbital involvement. In our case the initial surgical repair of an entropion, potentially spastic and/or senile, was hampered due to a large conjunctival neurofibroma. This surgical repair allowed for a mild improvement in chemosis, which allowed for the identification of a conjunctival neurofibroma. Subsequent surgical debulking allowed for proper eyelid apposition against the globe and resolution of the patient’s signs and symptoms. Lower eyelid entropion has only been reported as a post-surgical complication after debulking a conjunctival neurofibroma, however, it has not been described upon presentation of a neurofibroma and associated with conjunctival chemosis. Consideration of a conjunctival neurofibroma should be warranted when confronted with eyelid malposition, particularly when refractory to surgical treatment, in the setting of a phakomatosis.

REFERENCES