

Corneal Myofibroma (Keloid) in a Young Patient with Neurofibromatosis Type 2

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Established Facts

- Neurofibroma occurs in neurofibromatosis type 2.
- Lagophthalmus occurs in neurofibromatosis type 2 due to facial palsy.

Novel Insights

- Corneal myofibroma (keloid) occurs in neurofibromatosis type 2 in association with facial palsy.

Keywords

Corneal myofibroma · Keloid · Neurofibromatosis type 2

Abstract

We present a 27-year-old male patient with neurofibromatosis type 2 (NF2), facial palsy, and lagophthalmos following acoustic neuroma removal and an impressive vascularized corneal tumor, which was excised. Histology showed a fibrous tumor with small vessels, and immunohistochemistry was positive for vimentin and negative for smooth muscle actin, S100, and GFAP. We assume a corneal myofibroma (keloid), which in this case rather represents a reactive lesion. This entity has not been described before in NF2 or in facial palsy-associated lagophthalmos in general.

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Background

Neurofibromatosis type 2 (NF2) is an autosomal dominant disease resulting from a mutation in the NF2 tumor suppressor gene. It occurs in one out of every 60,000 live births [1] and is characterized by multiple neoplasms such as spinal cord schwannomas, meningiomas, ependymomas, and vestibular schwannomas [2]. Ocular involvement in NF2 is common and in about 10% the first presenting sign. These ocular manifestations include anomalies in the anterior segment of the eye such as corneal opacities, intrascleral schwannomas, Lisch nodules, and melanocytic lesions of the iris [3] and most frequently subcapsular cataract formation with a frequency of occurrence of 38–81% [4]. To our knowledge, the formation of a corneal myofibroma (keloid) has not yet been described in the literature in NF2 in the setting of lagophthalmos following surgery for an acoustic neuroma so far.



Fig. 1. A whitish tumor on the cornea of the left eye with multiple vessel tufts within the tumor stroma of a 27-year-old male patient with known neurofibromatosis type 2.

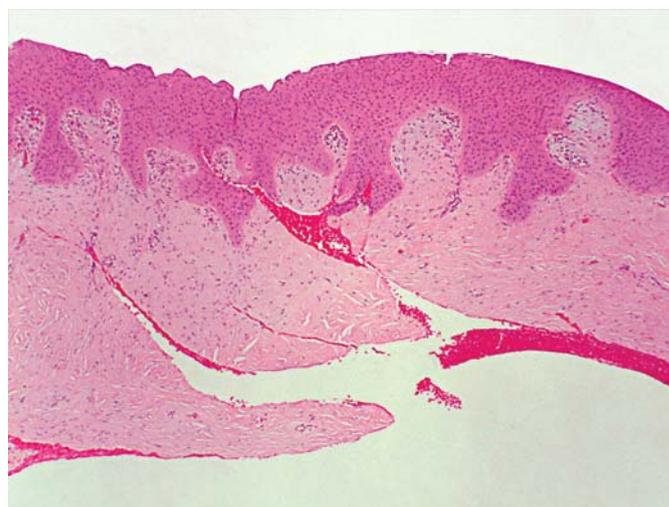


Fig. 2. Histology showing hyperplastic, non-keratinizing corneal epithelium with metaplasia towards keratinizing epithelium and dense vascularized fibromatous tissue (hematoxylin-eosin, original magnification $\times 100$).

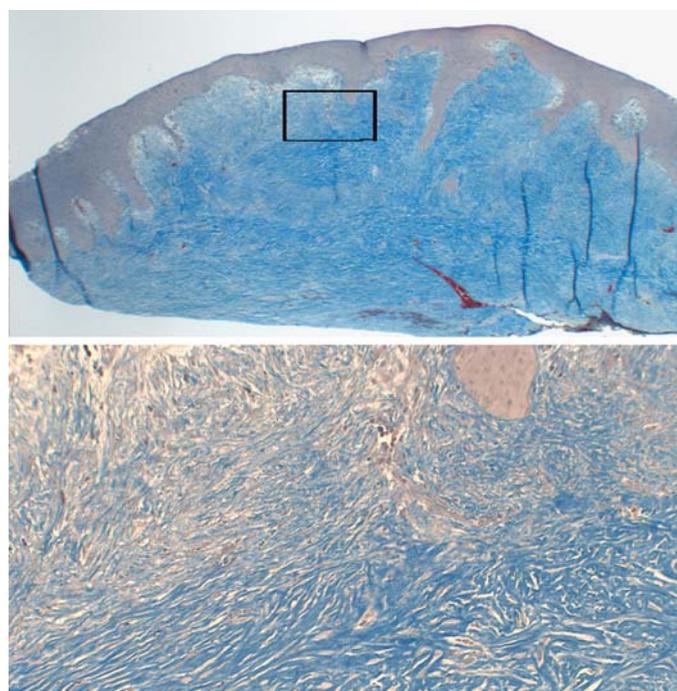


Fig. 3. Upper half: low-power magnification of the specimen with the fibrous tissue highlighted by bluish positivity for Masson trichrome (original magnification $\times 40$). Lower half: high-power magnification of the tumor stroma with small blood vessels of the area highlighted by the square (Masson trichrome, original magnification $\times 200$).

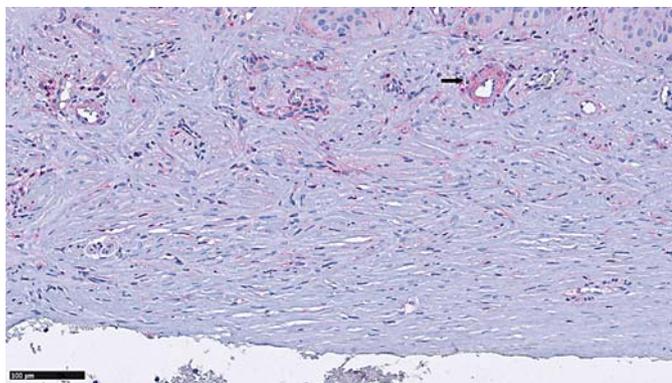
Case Report

Clinical Findings

A 27-year-old male patient with known NF2 presented with an enlarging whitish tumor measuring 8×5 mm on the cornea of his left eye with multiple vessel tufts within the tumor stroma (Fig. 1). According to our charts, the tumor had not been present 7 years earlier, and no other ocular signs of NF2 were present. The anterior OCT showed dense tissue within the tumor area without clear demarcation between tumor and corneal stroma. The adjacent limbal region showed deep vascularization besides pronounced conjunctival hyperemia. Visual acuity was reduced to 0.4. His medical history included bilateral excisions of acoustic neuromas with subsequent bilateral facial nerve palsy, bilateral lagophthalmos, which was more pronounced on the left side, and deafness 8 years earlier. Due to lagophthalmos, previous corneal problems had been chronic irritation and conjunctivalization in the lower half of both corneas. There was no ocular trauma reported by the patient.

Therapy and Diagnosis

Initially, conjunctival intraepithelial neoplasia was suspected, but intraoperatively, a collagenous tumor was found, which did not show an obvious “dissection plane” towards the cornea. Due to our positive experience in using mitomycin C 0.02% in fibrovascular proliferative conjunctival lesions as symblepharon or pterygia, we applied mitomycin C 0.02% for 2 min intraoperatively. Postoperatively, haze in the central and lower part of the cornea resulted in limited visual acuity of 0.32. Bilateral lagophthalmos improved after ophthalmoplastic surgery following corneal tumor excision, but was still present. Within 3 months of follow-up, no recurrence developed.



Color version available online

Fig. 4. Mainly negative staining of tumor cells for smooth muscle actin, whilst adjacent vessels were positive (arrow).

The histology of the specimen showed hyperplastic non-keratinizing corneal epithelium with metaplasia towards keratinizing epithelium and dense vascularized fibromatous tissue (Fig. 2) with intense bluish Masson trichrome positivity (Fig. 3). The tumor cells were positive for vimentin, indicating cells of mesenchymal origin, but negative for S100 and neurofilament, excluding neurofibroma. Immunohistochemistry against smooth muscle actin was mainly negative as well, only the vessel walls and few cells stained positively (Fig. 4).

Discussion and Conclusion

Initially, we favored the diagnosis of a neurofibroma as this tumor entity had been described at other body locations in association with neurofibromatosis. Never-

theless, neither S100 nor neurofilament positivity was found. According to the histological findings, i.e., strong vimentin positivity and smooth muscle actin negativity, this corneal tumor is consistent with a corneal myofibroma (keloid). Corneal myofibromas generally occur without preceding trauma, even in very young children [5].

Our patient suffered from chronic corneal exposure caused by a long-standing lagophthalmos following resection of an acoustic neuroma, which may have induced the formation of corneal keloid or hypertrophic scar. Nevertheless, no association between lagophthalmos and the formation of keloid has been reported in the literature up to now. Thus, we assume that the underlying neurofibromatosis is a predisposing factor for the corneal myofibroma in the setting of lagophthalmos following surgery for an acoustic neuroma.

Statement of Ethics

All subjects have given their informed consent to publish (in print and online) patient descriptions, photographs, and pedigrees.

Disclosure Statement

All authors had full access to all of the data in the observation and take responsibility for the integrity of the data and the accuracy of the data analysis. None of the authors has any relevant financial interest, activities, or relationships to disclose.

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