Case Report

Cutaneous Neurofibroma of the Lacrimal Caruncle: A Case Report

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Keywords

Neurofibromatosis type 1 · Cutaneous neurofibroma · Lacrimal caruncle

Abstract

We describe the case of a neurofibroma on the lacrimal caruncle of a female patient with neurofibromatosis type 1 (NF1). NF1 is an autosomal dominant genetic disease with a wide variety of clinical manifestations, one of the most common of which is neurofibroma. The lesion was removed surgically under general anesthesia and sent to histopathological analysis, which confirmed the clinical diagnosis of a neurofibroma.

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Introduction

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder with widely varying clinical manifestations affecting 1:2,500 individuals worldwide [1, 2]. NF1 arises as a result of mutations to the *NF1* gene, located on chromosome 17q11.21, a tumor suppressor gene coding for the protein neurofibromin [3]. The most common lesion is the cutaneous neurofibroma, a benign solid tumor appearing on the skin of 90% of adults with NF1. The number of cutaneous neurofibromas in an affected individual can vary from a few to several

thousand [3, 4]. Cutaneous neurofibroma size generally ranges from one millimeter to several centimeters [3].

Case Report

A 57-year-old female patient with confirmed clinical and genetic diagnosis of NF1 and extensive cutaneous neurofibromas throughout the body surface presented with a mass on the caruncle of the left eye. The mass was round, mobile, and could be manipulated within the eye so as to no longer be visible but occasionally spontaneously extruded from the eye (Fig. 1).

The subject underwent surgical removal of the neurofibroma under general anesthesia and the excised lesion was sent to histopathology. Histopathology analysis revealed a polypoid fragment with a small pedicle measuring $0.8 \times 0.7 \times 0.5$ cm, with firm consistency and light brownish coloration. Slicing revealed a homogenous solid nodule, whitish in color.

Microscopic analysis of histological sections revealed a nonencapsulated nodule composed of a proliferation of cells with elongated and undulated nuclei, with little individualized cytoplasm within a pale pink-colored matrix (Fig. 2, Fig. 3). No atypical features or atypical figures of mitosis were noted. The lesion was well vascularized, with dilated and congested capillary vessels, and between the conjunctiva and the epithelial surface, there was loose connective tissue that was also vascularized. The conjunctival epithelium covering the lesion exhibited squamous metaplasia and mucosecreting cells. The small pedicle contained sparse smooth muscle fibers.

Discussion

Cutaneous neurofibromas are benign lesions that can appear anywhere on the body [1–3]. While benign, these lesions can be present in number anywhere from a few to thousands, leading to significant cosmetic concerns for the affected patient [2]. To date, treatment of cutaneous neurofibromas relies on surgical removal, which, while effective, may lead to scarring and in cases where large numbers of neurofibromas are present may not be viable [5]. This case report highlights an unusual neurofibroma location within the left lacrimal caruncle. To our knowledge, this is the second case of a neurofibroma involving the caruncle reported in the literature [4] and the first in Brazil. Ophthalmological manifestations of NF1 include iris Lisch nodules, eyelid and orbital neurofibromas, congenital glaucoma, optic nerve gliomas, and meningiomas [5].

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Statement of Ethics

The patient described in this case report agreed to this publication and provided written informed consent.

Disclosure Statement

The authors have no disclosures to make.

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Fig. 1. Neurofibroma located on the caruncle of the left eye with visible extrusion.

Fig. 2. Pseudoencapsulated nodular proliferation located in the corium beneath the conjunctival epithelium. Cells with ill-defined cytoplasm, arranged in bundles, presenting regular comma-shaped nuclei and stroma with collagen deposits. Squamous metaplasia on suprajacent conjunctival epithelium. Staining: H&E.

Fig. 3. Tumor detail presenting peripheral nerve sheath cells with comma-shaped nuclei. Intermingled dilated and congested capillaries. Staining: H&E.