

Case Report

A rare case of a solitary intraocular neurofibroma

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Abstract

Background: Solitary neurofibroma in the absence of neurofibromatosis is of rare occurrence and very few cases have been reported till date. **Objective:** To report a case of a solitary intra-ocular neurofibroma **Case:** A 65-year-old man presented to us with a large swelling appearing to arise from right pthysical eye for the past one and a half years. After knowing the extent and origin of mass lesion, right eyeball was enucleated and subjected to histopathological examination which revealed intraocular neurofibroma in the absence of neurofibromatosis which is of very rare occurrence. **Conclusion:** The isolated neurofibroma of intraocular origin can present as an isolated orbital mass without systemic features.

Keyword: Neurofibromatosis, solitary neurofibroma

Introduction

A neurofibroma is a benign tumor arising from Schwann cells of the peripheral nervous system. It is usually associated with neurofibromatosis type I, a multisystem autosomal dominant disorder in which the nerve tissue grows benign tumors that may cause disfiguration and serious damage by compressing nerves and other tissues. The disorder affects all neural crest cells, i.e., Schwann cells, melanocytes and endoneural fibroblasts. Affected cells exhibit biallelic inactivation of the NF1 gene at 17q chromosome that codes for the protein neurofibromin (Muir et al 2003). Apart from occurring in association with neurofibromatosis, isolated neurofibromas may occur. However, the exact incidence of true solitary or isolated neurofibroma occurring intraocularly or in the orbit is difficult to obtain because of its relation to neurofibromatosis in most of the cases. Because of its rarity, it is often difficult for an ophthalmologist to diagnose it on a presumptive basis.

Case report

A 65-year-old gentleman presented with a large, pedunculated swelling arising from the right pthysical eyeball which had developed during the past one and a half years. The patient correlated its occurrence with a minor trauma to the right pthysical eyeball one and a half years back, following which he noticed a small nodule which was painless and gradually progressed to the present extent of approximately 4 x 3 cm mass (Figure 1).

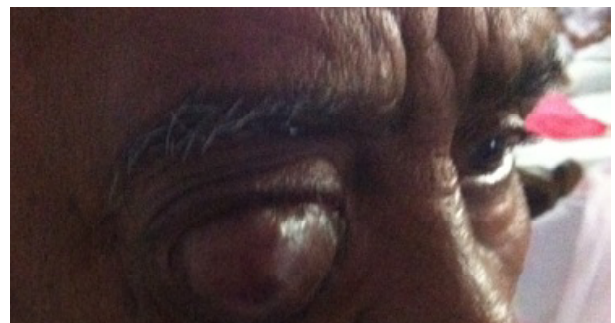


Figure 1: Showing the large swelling in the right eye.

The mass was hard in consistency with keratinization of the overlying surface. It appeared to arise from the superomedial aspect of the pthysical

eyeball, effacing whole of the remaining structure.

A slit-lamp biomicroscopy examination of the left eye revealed a normal anterior segment except for grade I nuclear sclerosis. The fundus examination was normal.

The systemic examination presented with no significant findings. Systemic features of neurofibromatosis like multiple, multifocal neurofibromas, *café au lait* spots or axillary freckling were absent. The family history was not significant.

The USG of the right orbit revealed a hypoechoic mass lesion arising from the right pthysical eyeball and extending anteriorly into the right lower lid. It showed flow on Color Doppler and foci of calcification were also present. The CT scan revealed a heterogenous, fairly enhanced soft tissue mass occupying the whole of the right eyeball with foci of calcification in it. Stranding of retrobulbar fat in the region of the optic nerve head was present. No involvement of the opposite eye and brain was noted (Figure 2).

Informed consent was taken, and after knowing the extent and origin of the mass lesion, the right eyeball was enucleated and sent for gross and histopathological examination.

On histopathological examination, a proliferation of the nerve cells and Schwann cells forming lobules was seen, along with the foci of calcification. The result was an effacing of the structures of the eyeball. Several myelinated axons, bundles of Schwann cells and collagen fibres were distributed within a myxoid stroma which was positive for Alcian blue. Scattered perivascular lymphocytes were also seen. All these features confirmed the presence of an intraocular neurofibroma (Figure 3).

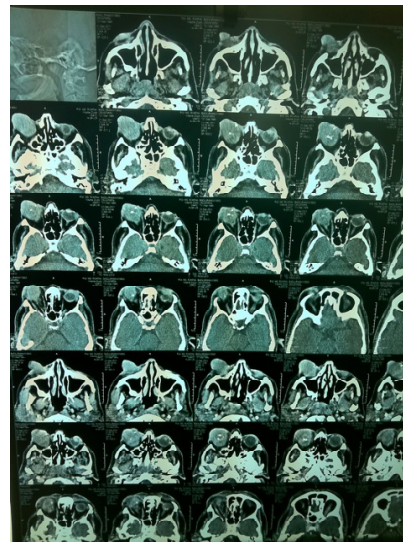


Figure 2: The CT scan showing the effaced right eyeball

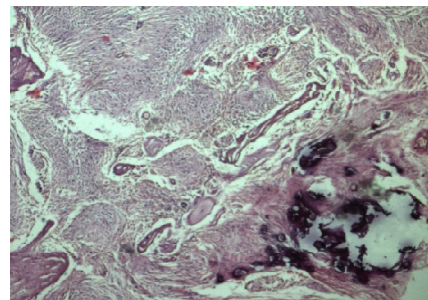


Figure 3: Photomicrographs showing lobules of nerve cells and Schwann cells (arrows). H&E, 20x

Discussion

Neurofibromas are benign tumors of the nerves characterized by proliferation of nerve cells and Schwann cells along with intervening fibrous components. These are most commonly associated with neurofibromatosis I along with other systemic findings. Those associated with NF I usually present in early childhood. Neurofibromas are radio-resistant and widely-infiltrating ones are difficult to excise surgically.

The exact incidence of isolated neurofibroma especially of those of intraocular origin is difficult to obtain; however, its occurrence is relatively rare. No case of isolated intraocular neurofibroma has

yet been reported as per our knowledge. Very few cases of isolated orbital neurofibroma have been reported. A study revealed a 93 % incidence of benign neurilemmoma or neurofibroma among orbital peripheral nerve sheath tumors (Rose et al 1991). They defined a family history of systemic neurofibromatosis in one quarter of the patients with a solitary neurofibroma. In a series of orbital tumor cases only three cases of a solitary neurofibroma, occurring in middle aged persons as a slow growing upper quadrant mass, have been reported (Rootman J 2003). Alkatan HM (2007) reported a case of an isolated neurofibroma of orbit in a 25-year-old male. Shields et al (1990) reported a case in which the patient had no manifestations of neurofibromatosis with three separate right orbital lesions.

In our case, it was difficult to determine the nerve and the underlying tissue from which the neurofibroma arose and whether the ptosis of the same eye preceded the development of the neurofibroma and was independent of it or not.

Conclusion

The isolated neurofibroma of intraocular origin can present as an isolated orbital mass without systemic features.

References

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