

ENUCLEATION IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1 WITH BUPHTHALMOS AND OCULAR DEFORMITY

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Abstract- In infants with neurofibromatosis type 1, plexiform neurofibroma with altering anterior segment outflow system may result in raised intraocular pressure. Here we report a case of buphthalmos with neurofibromatosis (NF-1) who suffered with blindness suffered enucleation. Physical examination and histopathology were performed. Physical examination revealed Buphthalmos and vascularized cornea with lower lid ectropion and orbital deformity. Histopathologic findings showed plexiform neurofibroma in orbital tissue with scleral wall involvement. And hamartomatous proliferation in choroid and ciliary bodies with melanocytic elements. Few NF-1 cases with buphthalmos have been reported. But cases with complicationg buphthalmos (retinal detachment and optic nerve atrophy) suffering enucleation are very rare.

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Key words: Neurofibromatosis-1, plexiform neurofibroma, buphthalmos, enucleation.

INTRODUCTION

Neurofibromatosis type 1 (NF-1) is an autosomal dominant inherited syndrome caused by a gene mutation on chromosome 17. It is characterized by the presence of multiple café-au-lait spots, Lisch nodules and multiple benign neurofibroma (1). Neurofibromas subdivide into fibroma molluscum, plexiform neurofibroma and elephantiasis neurofibroma (2). Another ophthalmic findings is congenital glaucoma (2). In infants plexiform neurofibroma with altering anterior segment outflow system result in raised intraocular pressure. This can produce a uniform enlargement (Buphthalmos) (3).

In this paper we present a case with NF-1 and chronic ocular plexiform neurofibroma and complicating Buphthalmos suffering enucleation.

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CASE REPORT

A 21-years old man presented to the Tehran farabi hospital complaining of left eye blindness and orbital deformity from the birth. Ophthalmic exploration revealed buphthalmos (globe antero-posterior Diameter is 3.8 cm and corneal Diameter is 1.4cm), with opacified and vascularized cornea with lower lid Ectropion. CT scan findings revealed orbital wall defect and periscleral mass with scleral thickening and extension to the brain. He underwent enucleation because of severity and chronicity of disease and unilateral visual loss.

Pathology

Gross examination revealed diffuse enlargement of globe (3.8 cm in ant-posterior diameter) and cornea (1.4 cm in maximal Diameter) in section. There was diffuse thickening of choroid and opacification of ocular lens was obvious (Fig. 1).

Histologically periscleral tissue showed proliferation of all elements of a peripheral nerve:

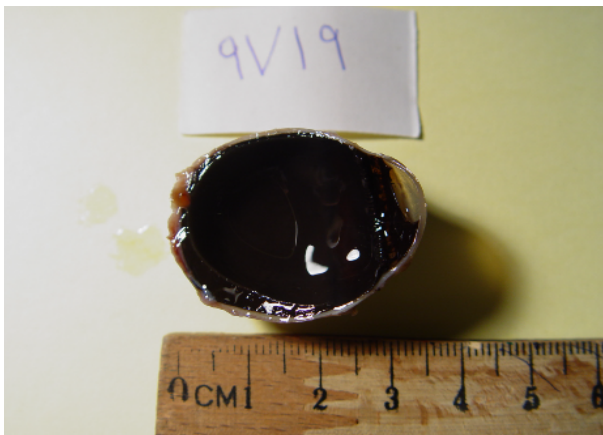


Fig. 1. Macroscopy, eye enucleation, enlargement of globe, diffuse thickening of choroid.

axons, schwann's cell, fibroblasts and perineurial cells that expands contiguous nerves, which appear as small nodules with a dense eosinophilic rim. (plexiform neurofibroma (Fig. 2). In some microscopic foci thickening of sclera with scleral wall involvement with neurofibroma is overt.

Choroid and ciliary bodies shows diffuse hamartomatous proliferation of neural and melanocytic elements (Fig. 3, 4). Retina revealed exudative detachment. Optic disk with cup-like appearance and optic nerve atrophy.

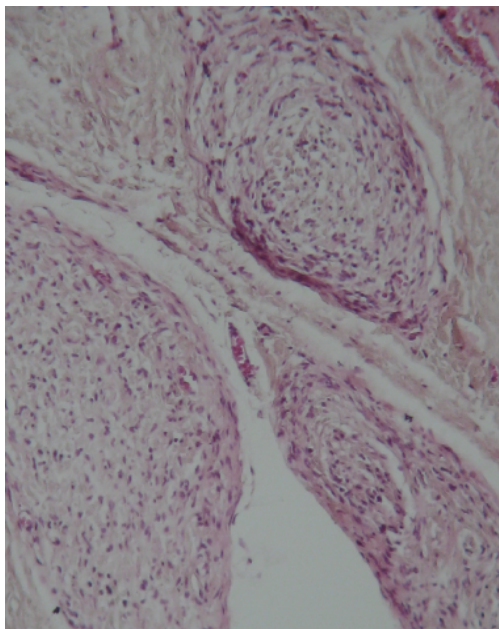


Fig. 2. Periscleral tissue showed proliferation of all elements of a peripheral nerve: axons, schwann's cell, fibroblasts and perineurial cells that expands contiguous nerves, which appear as small nodules with a dense eosinophilic rim. (plexiform neurofibroma).

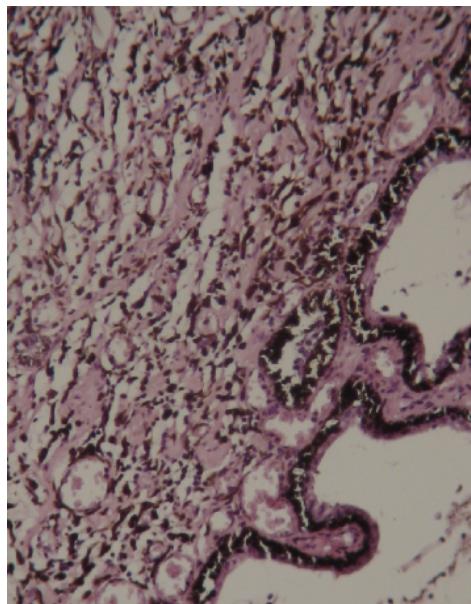


Fig. 3. Choroid body showing diffuse hamartomatous proliferation of neural and melanocytic elements.

DISCUSSION

Neurofibromatosis is one of the most common autosomal dominant central nervous system disorders (1). it had divided into at least two major subtypes (NF-1 and NF-2) with gene defects of different chromosomes (2) the prevalence being 1 in 2500 to 3300 (3).

The primary defect is related to disorder of neural crest – derived cells (melanocytes and Schwann and chromaffin cells) (4) despite the advances in molecular biology, the diagnosis of NF-1 can be done on a clinical basis. Clinical criteria for the diagnosis (5) is made if two or more of the following are found:

1. six or more café-au-lait spots over 5mm in greatest diameter (over 15 mm in postpuberal individuals).
2. two or more neurofibromas of any type or plexiform neurofibroma
3. freckling in the axillary or inguinal areas.
4. optic glioma
5. two or more lisch nodules (pigmented hamartomas of the iris).
6. A distinctive osseous lesion, such as sphenoid dysplasia or thinning of long bone cortex.
7. A first degree relative with NF-I.

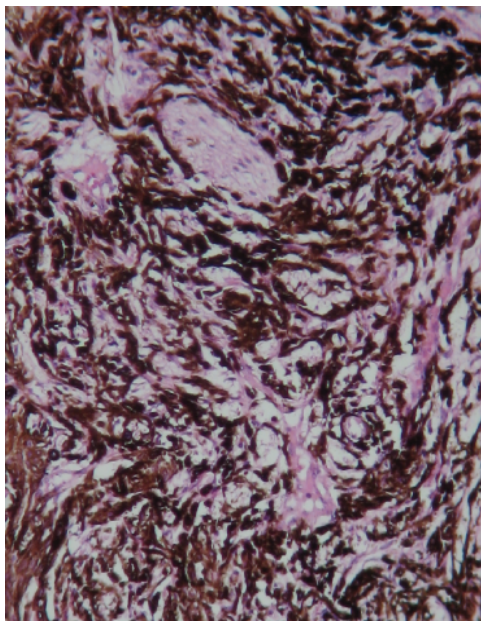


Fig. 4. Ciliary body showing diffuse hamartomatous proliferation of neural and melanocytic elements.

Ophthalmic finding in NF-1 include: (6).

1. café-au- lait spots.
2. Neurofibromas subdivides into:
 - a. fibroma molluscum, the common neurofibroma, results from proliferation of all element of the distal end of peripheral nerve: axon, schwann's cells, fibroblasts, and produces a small, localized skin tumor.
 - b. plxiform neurofibroma (bag of worms) is a diffuse proliferation of all components of conventional neurofibroma and perineurial cells (are Immunoreactive for EMA & negative for S-100) in the nerve sheath and produces a thicked and tortuous nerve.
 - c. Elephantiasis neuromatosa is a diffuse proliferation outside the nerve sheath that produces a thickening and folding of the skin.
3. Hamartomas in trabecular meshwork, uvea, neural retina, and optic nerve head:
 - a. melanocytic nevi in trabecular meshwork and uvea (clinically, the multiple, small, spider-like, melanocytic Iris nevi(lisch nodules) are the most common clinical feature of adult NF-1, Found in 93% of adults.
 - b. glial hamartomas in neural retina and optic Nerve head.
- C. Retinal capillary hemangiomas and combined pigment epithelial and retinal hamartomas.
4. sectoral neural retinal pigmentation.

5. optic Nerve glioma (Juvenile pilocytic astrocytoma).

6. thickening of corneal and conjunctival nerves and congenital glaucoma. If a plexiform neurofibroma of the eyelid is present (specially in the upper eyelid) 50% of the eyes will have glaucoma.

If the out flow system and the structures of the anterior segment fail to develop normally, the resistance to aqueous outflow is impaired and even before birth the intraocular pressure is abnormally high.

In contrast to the adult eye, the corneoscleral envelope of the infant and child is elastic and distensible so that a raised intraocular pressure can produce a uniform enlargement. The Globe may achieve diameter of the order of 40mm (Buphthalmos). NF-1 cases with buphthalmos have been Reported. But very rare cases with complicating buphthalmos suffering enucleation.

Chamber angle malformation often occurs as part of more complex diseases involving either other ocular tissues and/or other organ systems e.g. in NF and the Sturge-Weber syndrome (7).

This case qualified two out of six criteria for diagnosis of NF-1 (plexiform neurofibroma and pigmented hamartomas of iris and ciliary body).

Few NF-1 cases with buphthalmos have been reported (8). But cases with complicating buphthalmos suffering enucleation are very rare. Because of early outset of the disease in childhood and its progression, buphthalmos occurs and gradually followed by retinal detachment and optic nerve Atrophy and visual loss.

Thus scleral and orbital wall invasion by plexiform neurofibroma leads to orbital and eyelid deformity with all of these complications enucleation is indicated.

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