Kaohsiung Journal of Medical Sciences (2014) 30, 215–216

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: http://www.kjms-online.com

LETTER TO THE EDITOR

Co-occurrence of choroidal pigmented ganglioneuroma and plexiform neurofibroma in a patient with neurofibromatosis 1

Dear Editor,

Neurofibromatosis 1 (NF1) is an autosomal dominant disorder characterized by a wide variety of lesions. Ganglioneuromas are well-differentiated benign tumors originating from the ganglion cells of the sympathetic and parasympathetic nervous system.

A 21-year-old woman presented to the eye clinic of our institution with severe pain in her congenitally blind left eye for 6 months. She had asymmetry of the face and had undergone an operation for an asymmetrical left upper eyelid, the pathologic diagnosis of which was plexiform neurofibroma. She had multiple facial café au lait spots. An evisceration procedure for the painful blind eye using an implant was considered. At surgery, the surgeon noted no evident intraocular tissue.

Biopsy from the retrobulbar area indicated a clear diagnosis of plexiform neurofibroma on histopathologic examination. The evisceration material revealed diffuse thickening and replacement of normal choroidal tissue with bundles of spindle-shaped cells arranged in a fascicular fashion (Fig. 1A). Between these fascicles were numerous ganglion cells with abundant cytoplasm and large vesicular nuclei with prominent nucleoli. The cells were arranged in clusters and individual cells, which were positive for synaptophysin and neuron-specific enolase. There were also abundant pigment-containing cells showing strong positivity for HMB-45 and Mart-1. No atypia, necroses, or pleomorphisms were observed (Fig. 1B). We diagnosed the lesion that was in the evisceration as a ganglioneuroma. Ki-67 proliferation indexes were low in both the plexiform neurofibroma and the pigmented ganglioneuroma.

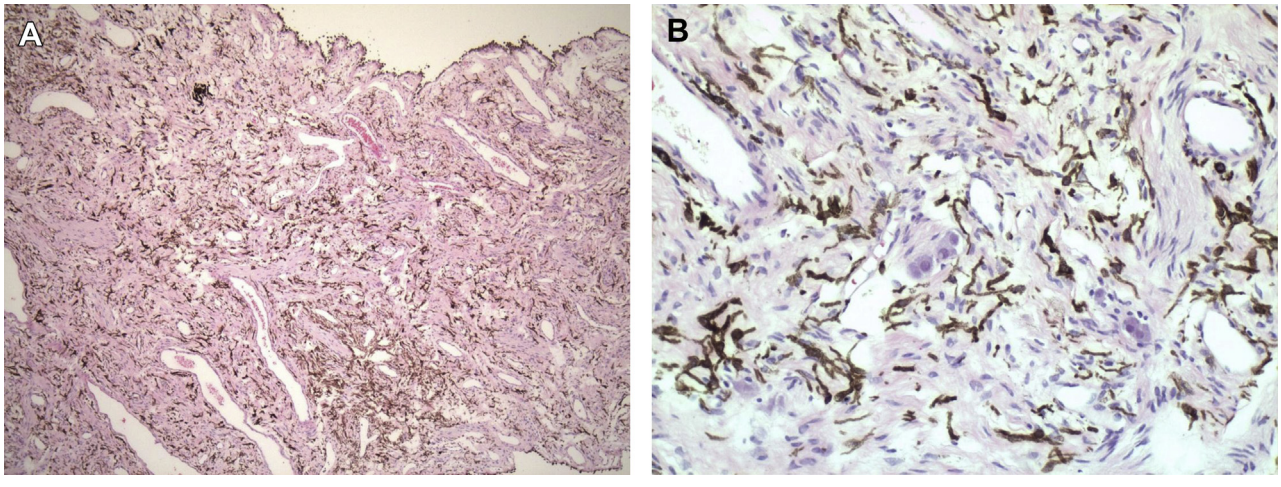

Figure 1. (A) Under choroidal lining, there are bundles of spindle-shaped neural tissue cells in a fascicular fashion (H&E ×40). (B) There are numerous ganglion cells and abundant pigment-containing cells (H&E ×200). H&E = hematoxylin and eosin.

http://dx.doi.org/10.1016/j.kjms.2013.02.001

Table 1 Clinical profiles of choroidal ganglioneuroma previously.

No.	Age (y)	R/L	Sex	IOP (mmHg)	VH	CNV	Other ophthalmic findings	Past treatments	Refs
1	21	L	F	ND	−	−	Cataract, total RD, globe enlargement, anterior staphyloma	Retinopexy	[3]
2	11	L	M	34	−	−	Cataract, globe enlargement	Trabeculectomy	[4]
3	7	L	M	25	+	+	Cataract, total RD, bullous keratopathy, globe enlargement	Trabeculectomy	[5]
4	21	L	F	ND	ND	ND	Cataract, globe enlargement	Partial eyelid excision for plexiform NF	Present case

CNV = choroidal neovascularization; IOP = intraocular pressure; ND = not described; R/L = right/left; RD = retinal detachment; VH = vitreous hemorrhage.

NF1 is an autosomal dominant disorder with a high penetrance and mutation rate caused by a single gene abnormality. Individuals with NF1 develop tumors, including neurofibromas, malignant nerve sheath tumors, and pilocytic astrocytomas, at increased frequency. The most common peripheral nerve tumor of the orbit is plexiform neurofibroma, and this tumor is pathognomonic of NF1. It may be responsible for orbital enlargement, thickening of the adjacent soft tissues (eyelids, periorbits, and face), and enlargement of the extraocular muscles [1].

Ganglioneuromas are rare, benign tumors, forming part of a spectrum of tumors arising from primordial neural crest cells in the sympathetic nervous system. Malignant transformation in ganglioneuromas has been only rarely documented in the literature. These few cases transformed into malignant peripheral nerve sheath tumors and neuroblastomas [2]. The association of extraocular ganglioneuroma with cutaneous manifestations of NF1 and with fully expressed neurofibromatosis is well-documented. However, choroidal ganglioneuroma is extremely rare; a MEDLINE search revealed only three published case reports. Table 1 [3—5] summarizes the clinical demographics of the patients with choroidal ganglioneuroma and NF1 reported previously [3—5].

We report a patient with NF1 who underwent evisceration for severe ocular pain in her left eye. We diagnosed a plexiform neurofibromatosis in the retrobulbar area and an unexpected choroidal ganglioneuroma in the same eye. It is likely that choroidal ganglioneuromas can present within the spectrum of NF1 in some patients. It should be kept in mind that in an NF1 patient with eye symptoms, a hitherto undiagnosed choroidal ganglioneuroma could exist. Close follow up is strongly recommended in these kinds of patients as well.

References

[1] Sippel KC. Ocular findings in neurofibromatosis type 1. Int Ophthalmol Clin 2001;41:25—40.
[2] Shimada H, Abros IM, Dehner LP, Hata J, Joshi W, Roald B, et al. The International Neuroblastoma Pathology Classification (the Shimada system). Cancer 1999;86:364—72.
[3] Woog JJ, Albert DM, Craft J, Silberman N, Horns D. Choroidal ganglioneuroma in neurofibromatosis. Graefes Arch Clin Exp Ophthalmol 1983;220:25—31.
[4] Shome D, Vemuganti GK, Honavar SG. Choroidal ganglioneuroma in a patient with neurofibromatosis type 1: a case report. Eye (Lond) 2006;20:1450—1.
[5] Ishijima K, Kase S, Noda M, Ishida S. Intraocular neovascularization associated with choroidal ganglioneuroma in neurofibromatosis type 1. Eur J Ophthalmol 2011;21:837—40.

Gonca Ozgun*
Saduman Balaban Adim
Nesrin Ugras
Department of Pathology, Faculty of Medicine, Uludag University, Bursa, Turkey

Bulent Yazici
Department of Ophthalmology, Faculty of Medicine, Uludag University, Bursa, Turkey

*Corresponding author. Department of Pathology, Uludag University, 16059 Görükle, Nilufer, Bursa, Turkey.
E-mail address: goncabarit@hotmail.com (G. Ozgun)