Case Report

Isolated Orbital Neurofibroma in NF-1 Negative Patients

Panrapee Funarunart MD*, Kanita Kayasut MD**,
Siriporn Hirunpat MD***, Passorn Preechawai MD*

This study was presented as poster in part of the World Ophthalmology Congress (WOC) annual meeting 2008, June 28 to July 2, 2008; Hong Kong, China

* Department of Ophthalmology, Faculty of Medicine, Prince of Songkla University, Hat Yai, Songkhla, Thailand
** Department of Pathology, Faculty of Medicine, Prince of Songkla University, Hat Yai, Songkhla, Thailand
*** Department of Radiology, Faculty of Medicine, Prince of Songkla University, Hat Yai, Songkhla, Thailand

Isolated orbital neurofibroma unassociated with systemic neurofibromatosis is relatively rare and may be difficult to clinically differentiate from other orbital tumors. The authors report a case of isolated orbital neurofibroma without neurofibromatosis type 1.

Keywords: *Orbital neurofibroma, Orbital tumor, Peripheral nerve tumor*

J Med Assoc Thai 2011; 94 (10): 1281-2
Full text. e-Journal: *http://www.mat.or.th/journal*

Case Report

A 54-year-old Thai woman presented with painless, slowly progressive downward proptosis about 6 mm and a palpable rubbery lobulated mass of the left upper eyelid for 10 years. Motility was normal in all directions. Her best-corrected visual acuity was 20/20 both eyes, and the clinical examination was otherwise unremarkable.

Computed tomography (CT) scan orbit revealed a well-circumscribed, homogenous enhancing extraconal mass within the superior part of the left orbit (Fig. 1). She underwent an anterior superior orbitotomy with total tumor removal. During the surgical operation, the finding revealed a grey rubbery well-encapsulated tubular mass of the supratrocheal nerve with minimal cystic component, as demonstrated in Fig. 2A. There was no adjacent tissue invasion. The tumor was totally removed without significant bleeding. Microscopic examination showed a typical neurofibroma with a positive reaction for S-100 protein in an immunohistochemical test, as shown in Fig. 2B.

She did not have any features or family history of neurofibromatosis type I (NF-1). Post-operatively, she reported a sensation of numbness on her forehead that spontaneously improved within 6 months. As of 10-month follow-up, there had been no other abnormal symptoms or tumor recurrence.

Discussion

A number of isolated cases of intraorbital solitary neurofibroma have been reported since 1985[1]. Isolated orbital neurofibroma unassociated with systemic neurofibromatosis is relatively rare and may be difficult to clinically differentiate from other orbital tumors. Patients often present with slowly progressive proptosis without pain. Visual acuity is often not affected unless the tumor compresses the optic nerve. As in the present case, the superior orbital location is the most common site as the tumor often arises from branches of the frontal nerve[2,3]. Bony involvement has been reported in previous literature[3].

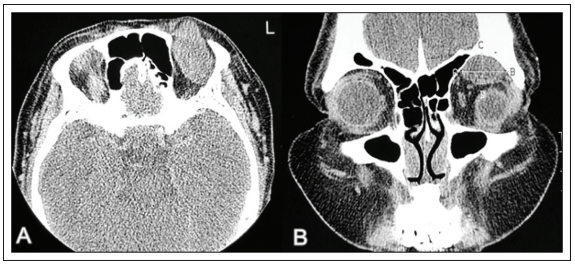

Fig. 1 Contrast-enhanced axial (A) and coronal (B) images revealed a well-circumscribed, homogenous extra-conal mass within the superior part of the left orbit

Correspondence to:
Funarunart P, Department of Ophthalmology, Faculty of Medicine, Prince of Songkla University, Songkhla 90110, Thailand.
Phone: 074-451-381, Fax: 074-429-619
E-mail: panf81@hotomail.com

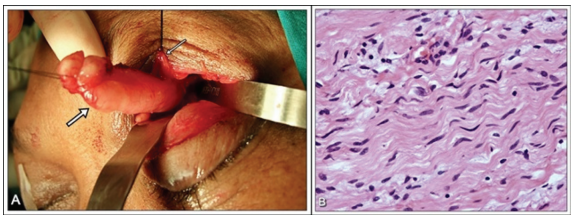

Fig. 2 A) The gross appearance of the tumor showed a grey rubbery well-encapsulated tubular mass of the supratrocheal nerve with minimal cystic component (large arrow). The intact supraorbital nerve was also demonstrated (small arrow)
B) Histopathology revealed a neurofibroma seen as Schwann cells with typical elongated nuclei and wavy pink cytoplasm and fibroblasts (hematoxylin-eosin stain)

The best imaging technique for diagnosis of orbital neurofibroma is controversial. CT appears to be most useful for delineating bony involvement, while Magnetic resonance imaging is most useful for defining the intra-structure of the tumor and relationship to adjacent tissue. A variety of radiologic patterns have been noted such as cystic lesions, solid lesions and mixed cystic-solid lesions. Such findings may result from a large spectrum of complex cellular differentiation modalities with the resulting variability in stromal composition[2].

Incisional biopsy with tissue pathology is required for definite diagnosis. Most cases do not need specific management, and surgical excision is considered depending on the size of the tumor and the degree of severity of the symptoms, such as clinical diplopia, severe proptosis and its complications, or optic nerve compression. Preoperative imaging studies are helpful in surgical planning. Total tumor removal can be achieved without serious complications. Radiation therapy is not advisable because a transformation into a neural sarcoma has been reported during radiation therapy[4].

It has been suggested that some neurofibromas may be related to somatic mutations or mosaicism, producing a segmental pattern of involvement as in isolated orbital neurofibroma[5]. Genetic analysis of surgical specimens may prove it.

Potential conflicts of interest

None.

References

1. Krohel GB, Rosenberg PN, Wright JE, Smith RS. Localized orbital neurofibromas. Am J Ophthalmol 1985; 100: 458-64.
2. Kottler UB, Conway RM, Schlotzer-Schrehardt U, Holbach LM. Isolated neurofibroma of the orbit with extensive myxoid change: a clinicopathologic study including MRI and electron microscopic findings. Orbit 2004; 23: 59-64.
3. Park WC, White WA, Woog JJ, Garrity JA, Kim YD, Lane J, et al. The role of high-resolution computed tomography and magnetic resonance imaging in the evaluation of isolated orbital neurofibromas. Am J Ophthalmol 2006; 142: 456-63.
4. Jamjoom AB, Malabarey T, Jamjoom ZA, al-Sohaibani M, Hulailah A, Kolawole T. Cerebrovasculopathy and malignancy: catastrophic complications of radiotherapy for optic nerve glioma in a von Recklinghausen neurofibromatosis patient. Neurosurg Rev 1996; 19: 47-51.
5. Kyprianou IG, Mantry S, Reuser T. Segmental neurofibromatosis. Eye (Lond) 2004; 18: 950-2.

ก้อนเนื้อชนิด neurofibroma ในเบ้าตาในผู้ป่วยที่ไม่ได้เป็นโรคท้าวแสนปม

พรรณรพี ฟูนฤนารถ, คณิตา กายะสุต, สิริพร หิรัญแพทย์, ภัสสร ปรีชาไว

ก้อนเนื้อในเบ้าตาชนิด neurofibroma ที่ไม่สัมพันธ์กับโรคท้าวแสนปมนั้นพบได้น้อยมาก การแยกโรคจากก้อนเนื้อในเบ้าตาชนิดอื่น ๆ ทำได้ยาก คณะผู้นิพนธ์ได้รายงานผู้ป่วย 1 ราย ซึ่งตรวจพบก้อนเนื้อในเบ้าตาชนิด neurofibroma ที่ไม่สัมพันธ์กับโรคท้าวแสนปม