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CLINICAL INVESTIGATIONS

Severe Optic Disc Edema Without Hydrocephalus in Neurofibromatosis 2

Takayuki Harada,* Yutaka Sawamura,†
Tsutomu Ohashi,* Chikako Harada,*
Yasuhiro Shinmei,* Kazuhiko Yoshida,* and Hidehiko Matsuda*

**Department of Ophthalmology, †Department of Neurosurgery,*
Hokkaido University School of Medicine, N15 W7, Kita-ku, Sapporo 060-8638, Japan

Abstract: A 26-year-old man who had neurofibromatosis type-2 with symptoms of unexplained optic disc edema is reported. Magnetic resonance imaging (MRI) revealed bilateral acoustic schwannomas. Obstructive hydrocephalus, however, was not evident in spite of his severe disc edema and visual loss. After partial removal of the right acoustic schwannoma, symptoms of intracranial hypertension, such as vomiting and headache, developed and MRI demonstrated evidence of obstructive hydrocephalus. Placement of a ventricular-peritoneal shunt relieved the symptoms of intracranial hypertension, but visual acuity in his left eye was reduced to hand motion due to secondary optic atrophy. In patients with similar symptoms it is suggested that, in addition to tumor removal, early treatment to decrease intracranial pressure should be considered when visual function is progressively impaired by the symptoms of prolonged papilledema. **Jpn J Ophthalmol 1998;42:381–384** © 1998 Japanese Ophthalmological Society

Key Words: Acoustic schwannoma, neurofibromatosis 2, optic atrophy, papilledema.

Introduction

Neurofibromatoses are genetic disorders that primarily affect the cell growth of neural tissues. These disorders can cause tumors to grow on the nerves at any location and at any time. Neurofibromatosis 1 (NF 1) (von Recklinghausen disease) and neurofibromatosis 2 (NF 2) (bilateral acoustic neurofibromatosis) have been established by the National Institutes of Health (NIH) Consensus Development Conference as distinct disorders among several proposed categories of NF.[1] Moreover, genetic loci associated with these diseases have been linked to dif–ferent chromosomes; NF 1 with chromosome 17,[2,3] and NF 2 with chromosome 22.[4–6] Bilateral acoustic schwannomas are common in NF 2, but not in NF 1.[7] Ocular manifestations in NF 2 include cataracts, retinal hamartomas, nystagmus, diminished corneal reflex; however, papilledema is reported to be less than 10%.[8–10] Papilledema is a sign of increased intracranial pressure. When papilledema is associated with posterior fossa tumor(s) in patients with NF 2, an obstructive hydrocephalus is usually the cause. Here we report an unusual case of NF 2 presenting optic neuropathy secondary to severe disc edema without typical findings of obstructive hydrocephalus.

Case Report

In 1993, a 26-year-old man with a 5-year history of hearing loss and tinnitus visited a hospital of otolaryngology. Otologic examinations revealed bilateral sensorineural hearing loss; right side was dominant. No cafe-au-lait spots or skin tumors were observed. He was otherwise in good health. Magnetic resonance imaging of the brain revealed bilateral acoustic tumors located in the cerebellopontine angle. To preserve hearing on the left side, the left

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Address correspondence and reprint requests to: Takayuki HARADA, MD, Department of Degenerative Neurological Disease, National Institute of Neuroscience, National Center of Neurology and Psychiatry, 4-1-1 Ogawahigashi, Kodaira, Tokyo 187-8502, Japan

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cerebellopontine angle tumor was treated with fractionated stereotaxis radiotherapy. However, the hearing loss progressively deteriorated.

He was referred to our clinic in January 1995. Our initial examinations revealed a best-corrected visual acuity of 20/20 in the right eye and 20/15 in the left eye. There were no Lisch nodules, cataracts, or glaucoma. No abnormalities were found in either fundus.

On September 5, 1995, the patient complained of decreasing visual acuity. Physical examinations revealed a large painless mass located in the right side of his neck. No additional neurologic abnormalities were found. His best-corrected visual acuity was 20/40 in the right eye and 20/30 in the left eye. Light reflex was normal. Fundus examination disclosed severe bilateral disc edema (Figure 1A). Magnetic resonance imaging revealed no change in size of the bilateral tumors on the eighth nerve (Figure 2A). Although the fourth ventricles were partially compressed by the mass effect due to the posterior fossa tumors, obstructive hydrocephalus was not evident (Figure 2B). No abnormalities, such as optic nerve glioma and meningioma, were found in the cavernous sinuses or in the orbits. On bilateral carotid angiograms, the sagittal sinus or jugular veins were patent.

He underwent a partial removal of the right cerebellopontine angle tumor on October 9, 1995. The histopathologic diagnosis was ordinary schwannoma. Several days after the surgery, the patient complained of severe headache and vomiting. Magnetic resonance imaging depicted apparent obstructive hydrocephalus with swelling of the right cerebellar hemisphere, presumably due to surgical intervention. Re-examination revealed the best-corrected visual acuity to be 20/30 in the right eye and hand motion in the left eye. Disc edema became progressive with symptoms of intracranial hypertension. A ventricular-peritoneal shunt was placed on November 9, 1995. Although hydrocephalus was resolved after the shunting, the patient continued to complain of visual loss in his left eye. Fundus re-examination re-

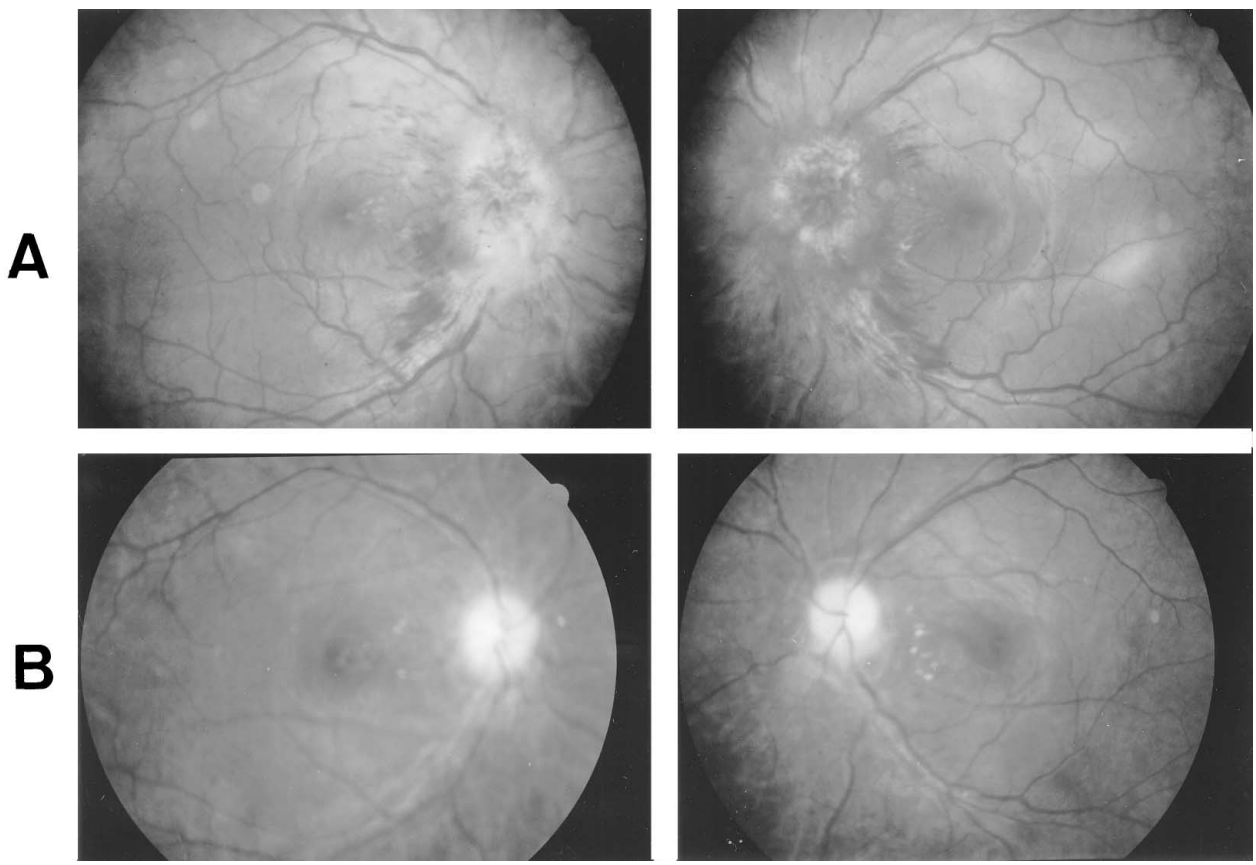

Figure 1. Fundus photographs of both eyes reveal severe papilledema (**A:** September 1995) and optic atrophy (**B:** December 1995).

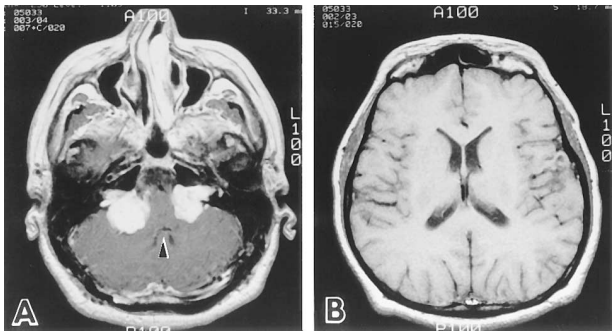

Figure 2. T1-weighted magnetic resonance images with gadolinium enhancement showing bilateral acoustic tumors located at cerebellopontine angle (September 1995). Note fourth ventricle was patent although partially compressed (arrowhead) (**A**). Lateral ventricles and cortical sulci were normal in size, and obstructive hydrocephalus was not evident (**B**).

vealed bilateral optic atrophy (Figure 1B). Visual field testing showed concentric contraction, and visual evoked response testing demonstrated a decreased amplitude in both eyes. The loss of visual functions remained stable in March 1996.

Discussion

Neurofibromatosis 2 (NF 2) frequently presents with ocular manifestations. Ragge et al[11] reported that ocular abnormalities were present in 42 (86%) of 49 patients with NF 2, but were the initial feature in only 5 (10%) patients. Juvenile cataracts (67%) and retinal hamartomas (22%) were common. Optic atrophy was, however, found in only 4 cases and 1 of them was attributed to a large intracranial meningioma. Chronic papilledema with an extensive serious retinal detachment and optic disk drusen was found in 1 patient.

Papilledema is a swelling of the optic nerve head produced by elevated intracranial pressure. In cases of neurofibromatosis, increased intracranial pressure is the consequence of either obstructive hydrocephalus due to neoplasm(s) in the posterior fossa, including multiple cranial nerve schwannomas, or of the mass effect of a large supratentorial meningioma. However, in the present case, disc edema was present in the absence of both these findings. Intracranial pressure was not measured at the first disclosure of disc edema, because the size of lateral ventricles was small, normal in size, and because a lumbar puncture is generally contraindicated in patients with a tumor in the posterior fossa, for fear of tonsilar herniation. Thus, strictly speaking, we cannot judge the disc edema seen in our patient as papilledema. We were not aware of the cause of disc edema before posterior fossa surgery, however, disc edema became progressive with an apparent evidence of obstructive hydrocephalus after unilateral removal of schwannoma.

In young people, ventricular obstruction sufficient to raise intracranial pressure may not necessarily enlarge the ventricles beyond the normal range.[8] The obstruction may be inconstant, but still be frequent or severe enough to cause papilledema. In the present case, the fourth ventricles seemed to be partially compressed by the tumors before surgery (Figure 2A). Thus, we cannot exclude the possibility that disc edema may have been just a result of the obstruction of ventricular cerebrospinal fluid (CSF) outflow due to the tumors. However, the intermittent obstruction of CSF outflow usually provokes nausea and headache, which did occur in this patient. In addition, this postulate is inconsistent with the postoperative findings; the patient developed symptoms of hydrocephalus after the removal of the schwannoma. Another possibility is that disc edema before posterior fossa surgery may have been papilledema due to the communicating hydrocephalus. Elevated CSF protein level or unusual protein constituent produced by spinal cord schwannoma can lead to intracranial hypertension by impairing CSF absorption.[12,13] This hypothesis may explain the clinical improvement associated with shunting without complete tumor removal, although we did not carry out an examination of CSF proteins.

We were unable to prevent bilateral optic atrophy, thus resulting in a visual loss to hand motion in the left eye. Although visual acuity in patients with papilledema is usually normal except for the enlargement of the blind spot, chronic papilledema can cause secondary optic atrophy. In this stage, nerve fiber damage causes a progressive loss of visual field in the form of an irregular constriction. In addition, secondary changes in the retinal pigment epithelium contribute to impairment of central vision. The results in this reported case suggest that, in addition to tumor removal, early progressive treatment for decreasing intracranial pressure (e.g., ventricular-peritoneal shunt) should be considered when visual function is progressively impaired by the symptoms of prolonged papilledema.

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