Case Report
pISSN 1738-2637 / eISSN 2288-2928
J Korean Soc Radiol 2014;70(2):83-86
http://dx.doi.org/10.3348/jksr.2014.70.2.83

대한영상의학회지

Multiple Solid Pilocytic Astrocytomas in Cerebellum with Neurofibromatosis Type I: A Case Report

제1형 신경섬유종 환자에서 소뇌에 발생한 다발성 고형성 털모양별아교세포종: 증례 보고

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Pilocytic astrocytoma usually has a classic imaging manifestation of a solitary, cyst-like mass with a strong contrast-enhancing mural nodule. There is only one published report so far of multiple solid and cyst type pilocytic astrocytomas in the cerebellum in neurofibromatosis type 1 (NF1) patient from the United States in 2007. We report a case of pilocytic astrocytoma presenting with only solid, multiple pilocytic astrocytomas in the cerebellum in NF1 patient.

Index terms
Pilocytic Astrocytoma, Multiple
Cerebellum
Neurofibromatosis Type 1

Received September 30, 2013; **Accepted** November 8, 2013
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INTRODUCTION

The most common giloma in patients with neurofibromatosis type 1 (NF1) is the pilocytic astrocystoma (1, 2). It commonly involves the optic tract and hypothalamus, in the order of frequency (2). However, the pilocytic astrocytoma is rarely found in other intracranial regions of NF1 patient (3).

Also, pilocytic astrocytoma usually has a classic imaging manifestation of a solitary, cyst-like mass with an strong contrast-enhancing mural nodule (4). Therefore, while the isolated pilocytic astrocytoma in NF1 is being well-described and frequently reported, multiple pilocytic astrocytomas in an individual patient is a less common form of manifestation. There has only been one single publication on the multiple involvements of pilocytic astrocytoma, which was located within the cerebellum of NF1 patient recorded in the United States in 2007 (2). According to our knowledge, no such case has been described since, thus, herein, we report a case of pilocytic astrocytoma presenting with only solid, multiple pilocytic astrocytoma in the cerebellum of the NF1 patient.

CASE REPORT

A 10-year-old male patient with a known history of NF1 was originally diagnosed from the presence of axillary freckling, multiple café-au-lait macules (> 6), and an affected first-degree relative presented with 1 month of headache. He underwent computed tomography (CT) and magnetic resonance imaging (MRI) of the brain in another hospital, and was found with multiple cerebellar masses. He visited our hospital for further evaluations and treatments.

His initial symptoms only included headaches, and there were no other neurological symptoms. We reviewed the external CT, which showed bilateral low densities with central iso- or high-density in both cerebellums (Fig. 1A). MRI found multiple well-demarcated nodular contrasts enhancing solid lesions in both cerebellar hemispheres with heterogenous low-signal intensity on T1-weighted images, and heterogenous high-signal intensity on T2-weighted images (Fig. 1B–E). Perilesional vasogenic edema surrounding these multiple enhancing solid lesions were also

found, which did not cause obstructive hydrocephalus. Generally, findings known as hamartomas or heterotopias (1), typically found in NF1 patients and presented as iso-signal intensities on T1-weighted images, and high-signal intensities on T2-weighted images without contrast enhancements, were found in the frontal subcortex, the right periventricular white matter, the posterior limb of right internal capsule, both the basal ganglia, the right thalamus, pons and the left superior cerebellum (Fig. 1F).

For the histological analysis, a left suboccipital craniotomy and an open biopsy was performed for one of the enhancing lesions in left cerebellar hemisphere. Pathology was consistent with low graded pilocytic astrocytoma (World Health Organization grade I), according to standard features of biphasic appearance together with loose glial components of multifocal myxoid changes and more compact piloid tissues with elongated nuclei (Fig. 1G). Cytologically, the tissues were positive for glial fibrilary acidic protein.

The patient was discharged without neurological deficits, and after 2 months, he underwent a gamma-knife surgery for multiple enhancing cerebellar solid masses. He was discharged after displaying no remarkable postoperative complications.

DISCUSSION

NF1, formerly known as von Recklinghausen's disease or peripheral neurofibromatosis, is a relatively common (incidence of

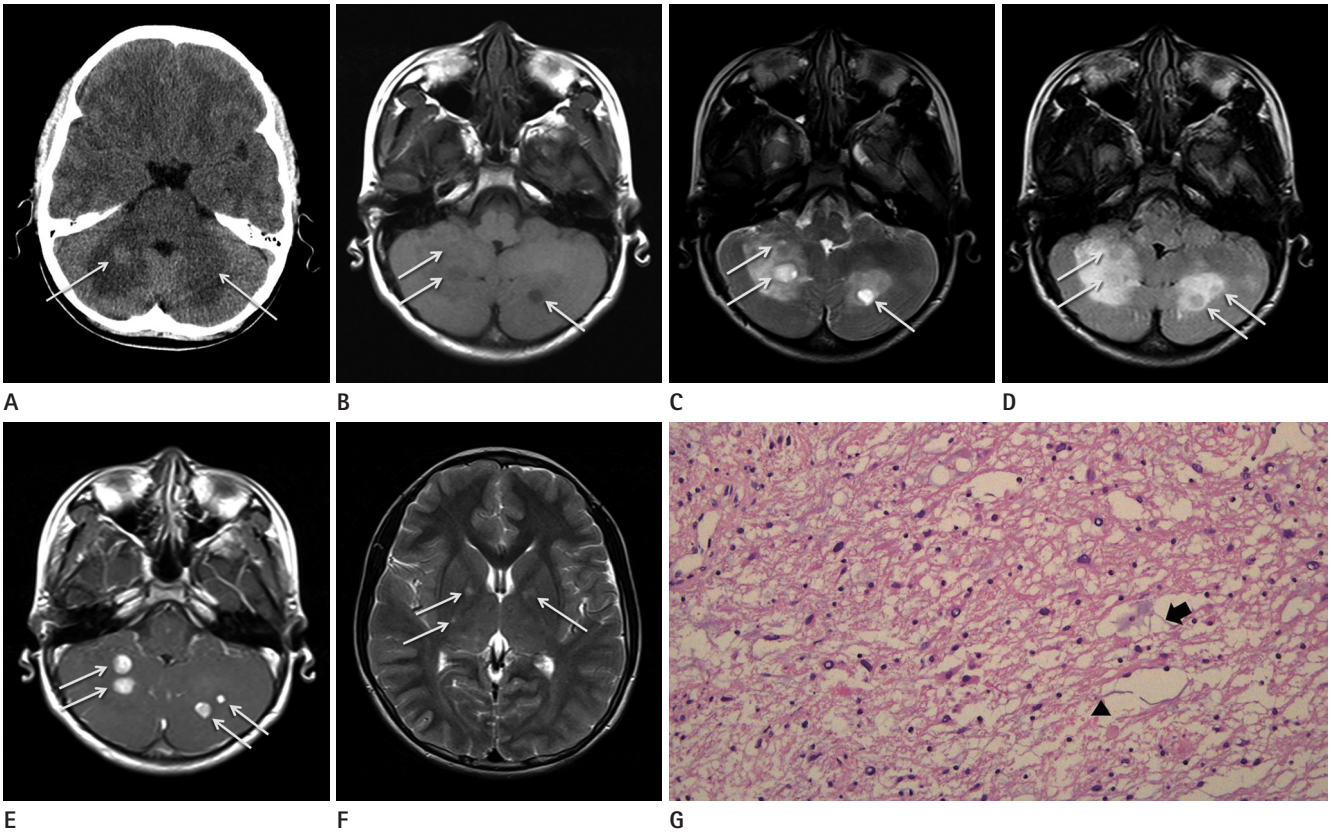

Fig. 1. A 10-year-old male with a known history of neurofibromatosis type 1.
A. Axial CT scan of the brain with brain window image reveals bilateral low density with central iso or high density in both cerebellum (arrows).
B–E. Axial T1 weighted **(B)**, T2 weighted **(C)**, T2 fluid attenuated inversion recovery **(D)** and post-contrast enhanced T1 weighted images **(E)** of the MRI of the brain show multiple well-demarcated nodular contrast enhancing solid lesions in both cerebellar hemisphere with heterogenous low signal intensity on the T1 weighted images, high signal intensity on the T2 weighted images (arrows). Perielesional vasogenic edema was surrounding these lesions without causing obstructive hydrocephalus.
F. Axial T2 weighted images of the MRI of the brain shows multiple high signal intensity involves both basal ganglia, posterior limb of right internal capsule (arrows).
G. Photomicrograph of the surgical specimen shows loose component of the tumor. Eosinophilic granulor bodies (arrowhead) and multifocal areas of myxoid change (arrow) are seen in the hematoxylin-eosin stain (× 200). Cytologically, glial fibrilary acidic protein (+) glial cells with elongated nuclei were seen.

1 in 3000–4000 births) autosomal dominant disorder, and is related to mutations in chromosomes 17 (17q11.2) which are encoded for the protein neurofibromin (1).

Although occurring less frequent than peripheral tumors, the central nervous system tumors are important for NF1 patients because they may lead to major morbidity and mortality rates (1, 2). The most common intracranial tumors are optic gliomas, which occur in about 5–15% of NF1 patients. Brain gliomas are observed in about 1–3% of the patients, and the most common glioma in NF1 is pilocystic astrocytoma (1, 2).

The association of pilocytic astrocytoma with NF1 has been well-documented (3, 4). For example, the cytogenetic analysis of pilocytic astrocytomas have detected losses of genetic materials involving the long arm of chromosomes 17 (17q) near the same locus for the NF1 tumor suppressor genes. Furthermore, the lack of expression on an NF1 gene product, the neurofibromin, has been documented in NF1-associated pilocytic astrocytomas (3, 4). These findings have fueled the speculations that NF1 tumor suppressor genes are linked with the expressions of pilocytic astrocytoma.

Pilocytic astrocytoma in NF1 patients typically involves the optic nerves or chiasms, which comprise up to about 14% to 15% of all tumors associated with NF1 (1-5), and 3.7% of brain stem tumors (1, 3, 5), but the actual percentages vary among different studies. Pilocytic astrocytoma is rarely found in other intracranial regions of NF1 patients. Cerebellum is an uncommon location for tumors in patients with NF1, and rarely surpasses a prevalence of 1% (3).

On the other hand, regardless of associations with NF1, the pilocytic astrocytoma is generally the most common primary brain tumor in children, comprising around 85% of all cerebellar astrocytomas and 10% of all cerebral astrocytomas within this age group (4). Most lesions arise from the cerebellum, the optic nerve and chiasm, or the region of the hypothalamus-thalamus, and occur within or near the midline (4) sections.

The clinical presentations of pilocytic astrocytoma vary with its site of origin. Symptoms, commonly found in cerebellar pilocytic astrocytoma, include headaches, vomiting, gait disturbances, blurred visions, diplopia, and neck pains (4, 6). Clinical signs usually include hydrocephalus, papilledema, truncal ataxia, appendicular dysmetria, head tilt, sixth nerve palsy, and nystagmus (4, 6).

The treatment of pilocytic astrocytoma also varies according its origin. Surgical resection is considered the treatment of choice for cerebellar pilocytic astrocytomas and is generally regarded as curative after gross removals of the tumor (4, 6). For lesion locations involving difficult approaches, the stereotactic resection may be used (4). Overall, the prognosis for patients with a pilocytic astrocytoma is excellent, usually with a 94% of 10-year survival rates and a 79% of 20-year survival rates (4).

On CT imaging, most pilocytic astrocytomas have well-defined borders with either a round or oval shape, smaller than 4 cm in size, cyst-like features, smooth margins, and occasional calcifications (4). In a study by Coakley et al. (7), most tumors (82% in one series) were located near the ventricle, and almost all of them (94%) showed intense enhancements on post-contrast images which were obtained after intravenous administration of contrast materials (4).

Numerous studies have described four predominant imaging patterns of pilocytic astrocytoma on MRI-I: mass with a nonenhancing cyst and an intensely enhancing mural nodule, II: mass with an enhancing cyst wall and an intensely enhancing mural nodule, III: necrotic mass with a central nonenhancing zone, and IV: predominantly solid mass with minimal to no cyst-like component (4, 6). Most studies have also shown that approximately two-thirds of all pilocytic astrocytoma demonstrated the typical imaging manifestation of a cyst-like mass with strong contrast-enhancing mural nodule. The degree of surrounding vasogenic edema are being diminished, which the expectation for a tumor with low biological activity (4).

However, there are occasional atypical imaging manifestations of pilocytic astrocytoma. More rarely, the multiple involvement of pilocytic astrocytomas may be presented within an individual patient (2, 8, 9). Such phenomenon has only been reported few times before—pilocytic astrocytoma in the multiple compartment (supra/infratentorial and/or spinal cord) in a patient without NF1 (8) and in same cerebral hemisphere associated with the NF1 patient (9). Also, multiple pilocytic astrocytomas in one cerebellar hemisphere of a patient with NF1 have been reported once by Dunn et al. (2). These case reports are meaningful due to the lack of literature for multiple types of pilocytic astrocytomas or posterior fossa tumors in patients with NF1.

Our case is similar to the one reported by Dunn et al., which described multiple solid and cystic type pilocytic astrocytomas in

the cerebellum of NF1 patient. However, our case is different in that there are only enhancing solid types of multiple pilocytic astrocytomas in the cerebellum, which is a less common manifestation rather than a typical manifestation of cyst-like mass with an strong contrast-enhancing mural nodule type, such as the report by Dunn et al. Thus, we believe our case to be more of a rare form of multiple pilocytic astrocytomas in the cerebellum.

In conclusion, we report a surgically and pathologically proven case of pilocytic astrocytomas which is comprised of multiple, solid enhancing nodules in both cerebellar hemispheres of the NF1 patient. Radiologists should expand the spectrum of presentation for patients with NF1 and consider multiple pilocytic asctorcytoma as a differential diagnosis when encountering multiple enhancing lesions that involve both cerebellar hemispheres in the NF1 patients.

REFERENCES

1. Mentzel HJ, Seidel J, Fitzek C, Eichhorn A, Vogt S, Reichenbach JR, et al. Pediatric brain MRI in neurofibromatosis type I. *Eur Radiol* 2005;15:814-822
2. Dunn IF, Agarwalla PK, Papanastassiou AM, Butler WE, Smith ER. Multiple pilocytic astrocytomas of the cerebellum in a 17-year-old patient with neurofibromatosis type I. *Childs Nerv Syst* 2007;23:1191-1194
3. Pascual-Castroviejo I, Pascual-Pascual SI, Viaño J, Carceller F, Gutierrez-Molina M, Morales C, et al. Posterior fossa tumors in children with neurofibromatosis type 1 (NF1). *Childs Nerv Syst* 2010;26:1599-1603
4. Koeller KK, Rushing EJ. From the archives of the AFIP: pilocytic astrocytoma: radiologic-pathologic correlation. *Radiographics* 2004;24:1693-1708
5. Guillamo JS, Créange A, Kalifa C, Grill J, Rodriguez D, Doz F, et al. Prognostic factors of CNS tumours in Neurofibromatosis 1 (NF1): a retrospective study of 104 patients. *Brain* 2003;126(Pt 1):152-160
6. Pencalet P, Maixner W, Sainte-Rose C, Lellouch-Tubiana A, Cinalli G, Zerah M, et al. Benign cerebellar astrocytomas in children. *J Neurosurg* 1999;90:265-273
7. Coakley KJ, Huston J 3rd, Scheithauer BW, Forbes G, Kelly PJ. Pilocytic astrocytomas: well-demarcated magnetic resonance appearance despite frequent infiltration histologically. *Mayo Clin Proc* 1995;70:747-751
8. Gilles FH, Leviton A, Hedley-Whyte ET, Sobel E, Tavaré CJ, Sobel RS, et al. Childhood brain tumors that occupy more than one compartment at presentation. Multiple compartment tumors. *J Neurooncol* 1992;14:45-56
9. Mukai K, Kitamura K, Asano N, Ohshima T, Hondo H, Matsumoto K. [Multifocal gliomas in cerebral hemisphere associated with von Recklinghausen's disease: case report]. *No Shinkei Geka* 1989;17:197-202

제1형 신경섬유종 환자에서 소뇌에 발생한 다발성 고형성 털모양별아교세포종: 증례 보고

최서영 · 김명순 · 김영주

Pilocytic astrocytoma는 흔히 단일 병변 형태로, 대부분 강한 조영증강을 갖는 고형 성분을 포함한 낭성 종괴의 특징을 보인다. 제1형 신경섬유종 환아에서 소뇌에 생긴 다발성 병변의 pilocytic astrocytoma는 매우 드문 예로 2007년 미국에서 증례 보고된 이후에 보고된 예가 없는 것으로 보여진다. 저자들은 제1형 신경섬유종 환아에서 소뇌에 다발성 고형성 pilocytic astrocytoma를 경험하였기에 이를 보고하고자 한다.

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