CASE REPORT

A neurofibromatosis type 2 case with vestibular, trigeminal and facial schwannomas: Magnetic resonance imaging findings

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Abstract

Neurofibromatosis type 2 is characterized by the development of multiple nervous system tumors. This disorder is also called multiple inherited schwannomas, meningiomas, and ependymomas syndrome. In this report, we discuss the magnetic resonance imaging findings in a patient with Neurofibromatosis type 2 who had right and left vestibular and trigeminal schwannomas, unilateral facial schwannoma, multiple meningiomas, and cervical intramedullary spinal cord tumors.

Key words

Neurofibromatosis type 2, Magnetic resonance imaging, Schwannoma, Cranial nerve, Meningioma, Intramedullary

1 Introduction

Neurofibromatosis type 2 (NF2) is a rare autosomal dominant disorder characterized by the development of multiple nervous system tumors [1]; it affects 1 in 33,000 to 40,000 people worldwide [2]. The presence of right and left vestibular schwannomas is a defining feature, but patients with NF2 also develop other cranial, spinal, and peripheral schwannomas; cranial and spinal meningiomas; and cataract [3]. This disease is also called the multiple inherited schwannomas (MIS), meningiomas (M), and ependymomas (E) (MISME) syndrome [4, 5]. NF2 is characterized by bilateral vestibular schwannomas. Schwannomas of the other cranial nerves (CNs) occur more frequently in NF2, and the presence of one of the rare cranial nerve schwannomas usually indicates the possibility of NF2 [6].

In this report, we present magnetic resonance (MR) imaging findings of an NF2 patient with vestibular, trigeminal, and facial schwannomas; multiple meningiomas; and cervical spinal intramedullary lesions.

2 Case presentation

A 23-year-old male patient complaining of tinnitus and imbalance for three years presented at the Ear-Nose-Throat

Department of our hospital. After the physical examination, the patient was referred to Radiology Department for further work up with MR imaging.

The MR imaging is performed with a 3.0 Tesla scanner (Philips, Achieva 3.0 T X-series, Best, The Netherlands). After the integration of 8-channel head coil, the examination started in supine position. Our department's routine protocol for the patients with a doubt of intracranial mass was applied. The sequences and the scan parameters; Axial T2-weighted Turbo spin echo (TSE) (Repetition time [TR] = 3,000 ms, Echo time [TE] = 80 ms, slice thickness [ST] = 5 mm, Number of excitations [NEX] = 2), axial T2-weighted Fluid Attenuated Inversion Recovery (FLAIR) (TR = 11,000 ms, TE = 125 ms, ST = 5 mm, NEX = 1), axial T1-weighted Inversion Recovery TSE (TR = 2,000 ms, TE = 20 ms, ST = 5 mm, NEX = 1), coronal T2-weighted TSE SPectral Attenuated Inversion Recovery (SPAIR) (TR = 3,467 ms, TE = 80 ms, ST = 5 mm, NEX = 2), pre-contrast axial, sagittal and coronal T1-weighted 3-dimensional (3D) Turbo field echo (TFE) (TR = 8.223 ms, TE = 3.773 ms, ST = 1 mm for sagittal, 3 mm for axial and coronal images, NEX= 1), Diffusion-weighted imaging and apparent diffusion coefficient map (TR = 2,808.5 ms, TE = 69.488 ms, b values = 0 and 1,000 s/mm^2), and post-contrast axial, sagittal and coronal T1-weighted 3D TFE (TR = 25 ms, TE = 4.6 ms, ST = 1 mm for axial, 1.5 mm for sagittal and coronal images, NEX = 1). Field of view was 230 mm×184 mm×139 mm (AP×RL×FH) for axial, 153 mm×184 mm×230 mm (AP×RL×FH) for coronal, and 250 mm×155 mm×250 mm (AP×RL×FH) for sagittal images. Total scan time was approximately 30 minutes.

Review of the brain MR imaging findings showed bilateral acoustic schwannoma that extended through the internal acoustic canals (IACs) and compress pons, bulbus, the left half of the inferior mesencephalon, right and left middle cerebellar peduncles, cerebellar hemispheres, and the forth ventricle (see Figure A-C, G, H: white arrows). The brain stem and the forth ventricle were deviated to the left because of the compression effect of the mass located in the left pontocerebellar angle. Additionally, bilateral symmetric homogeneously enhanced masses were detected at the trigeminal nerve localizations in Meckel's caves (see Figure A, C: thin white arrow). A similar 1-mm enhancing lesion was seen at the level of the geniculate ganglion of the right facial nerve, as well (see Figure A, D: white arrowhead). Eight other homogeneously enhancing mass lesions consistent with meningiomas, involving the falx cerebri, parafalcin region, cribriform plate, left frontal dural surface, and tentorium cerebelli, were observed (see Figure B, D-H, J: black arrows). The same examination showed two enhancing intramedullary well-defined small foci in the proximal cervical spinal cord (see Figure I, J: ellipse). Ependymomas or intraparanchimal schwannomas were initially suspected. The presence of bilateral schwannomas were diagnostic indicators of NF2 and there was no need for a biopsy. The patient was then referred to the Neurosurgery Department for surgical treatment.

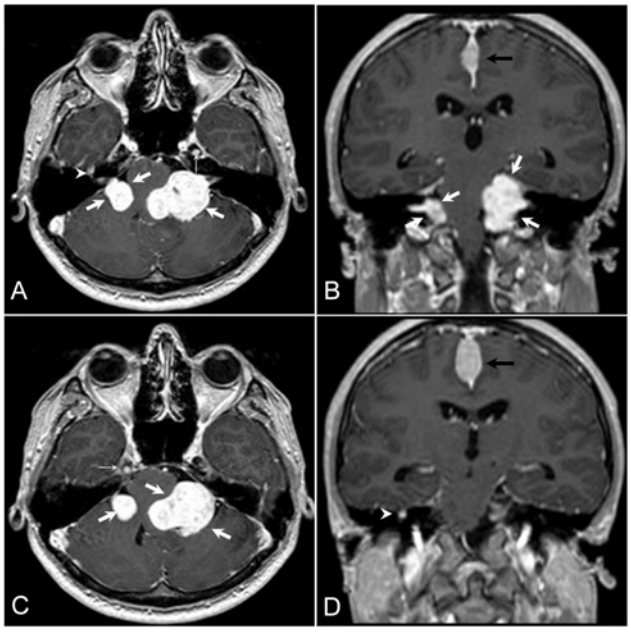

Figure A-D. Multiple varied cranial nerve schwannomas. Axial (A, C) and coronal (B, D) postcontrast three-dimensional T1 weighted images demonstrate homogeneously enhanced, well-defined, bilateral vestibular (thick white arrows) and trigeminal nerve schwannomas (thin white arrow). The pronounced mass affect of the left vestibular schwannoma to the pons, left middle cerebellar peduncle, and the fourth ventricle is observed. In (B), extension of the right and left vestibular schwannomas to the internal acoustic canals can be seen clearly. Additionally, facial nerve schwannoma at the level of the geniculate ganglion (white arrowhead) and a meningioma located at the falx cerebri (black arrow) are observed.

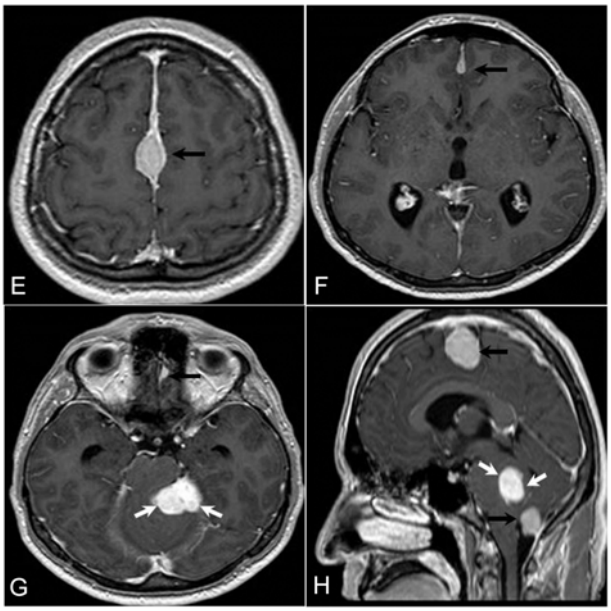

Figure E-H. Multiple meningiomas located at different sites. Axial (E-G) and sagittal (H) postcontrast three-dimensional T1 weighted images show well-defined, homogeneously enhanced lesions suggestive of meningiomas at the falx cerebri and posterior fossa, and adjacent to the cribriform plate (black arrow). A left vestibular schwannoma is also seen clearly (thick white arrows).

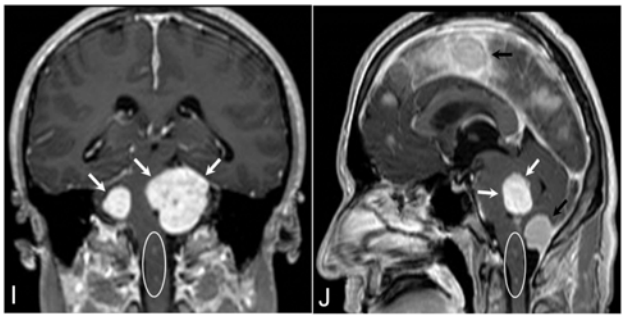

Figure I-J. Intramedullary spinal cord lesions. Coronal (I) and sagittal (J) postcontrast three-dimensional T1 weighted images demonstrate two enhancing, well-defined, small intramedullary lesions suggestive of ependymomas in the cervical spinal cord (ellipse). Bilateral vestibular schwannomas (thick white arrows), and the meningiomas (black arrow) located at the falx cerebri at the level of the vertex and posterior fossa are also seen.

3 Discussion

The diagnosis of NF2 is usually made in the second or third decade of life, with the highest number of diagnoses being made in the 20s. The clinical presentation of NF2 varies, but 30%-45% of the patients are diagnosed based on symptoms associated with CN VIII schwannomas, such as hearing loss, tinnitus, balance impairment, and weakness in CN VII distribution. The reason for this is that CN VIII schwannomas are symptomatic even when they are of a relatively small size. The symptoms present as a result of compression or stretching of the cochlear nerve, which compresses the blood supply to the nerve or to the cochlea, or causes hemorrhage into the nerve or cochlea [7].

Patients with CN schwannomas may present with loss of function of the affected nerve, but they can also be asymptomatic. In asymptomatic patients, the lesion may be incidentally discovered on computed tomography or MR scans obtained for reasons other than the evaluation of a schwannoma. In our case, the patient had tinnitus and balance impairment, both of which were attributable to vestibular nerve function loss related to vestibular schwannomas.

NF2 is characterized with bilateral vestibular schwannomas. Schwannomas of the other CNs occur more frequently in NF2, and the presence of one of the rare cranial nerve schwannomas usually indicates the possibility of NF2 [6]. CN schwannomas are usually isolated lesions, except when they are associated with NF2 [5]. Meningiomas and intramedullary ependymomas of the spinal cord also occur in NF2 [6]. Mautner *et al.* [8] reported the prevalence of vestibular schwannomas in 46 patients (96%), spinal tumors in 43 (90%), meningiomas in 28 (58%), and trigeminal schwannomas in 14 (29%) patients in a study including 48 NF2 patients. Aoki *et al.* [9] studied cranial MR images of 11 patients with NF2. In their series, all patients had acoustic schwannomas and eight had other cranial nerve tumors (five had multiple tumors and three

had a single tumor). We detected vestibular and trigeminal schwannomas on the left and right sides in our patient. The vestibular schwannomas had extended through the IACs, causing tinnitus and imbalance. Additionally, we detected a facial schwannoma at the level of the right ganglion geniculi and bilateral trigeminal nerve schwannomas, but there were no related trigeminal or facial complaints in our case.

Meningiomas are components of the MISME syndrome, and a common finding in NF2 patients [4]. They present as intradural extramedullary neoplasms that are very similar to spontaneous meningiomas and most commonly involve the thoracic spine; Frequently, more than one meningioma may be present [5]. Aoki *et al.* [9] reported the cases of eleven patients among whom six had meningiomas (four had multiple meningiomas and two had a single meningioma). In our case, we detected eight meningiomas of different sizes at various locations in the supra and infratentorial regions. The meningioma which was located in the posterior fossa was compressing the adjacent cerebellar paranchyma, and one of the meningiomas located at the level of the vertex was very close to the superior sagittal sinus.

Intramedullary spinal tumors may be associated with NF2. The majority of the intramedullary spinal tumors are ependymomas and arise either in the upper cervical cord or in the conus; however, astrocytomas and schwannomas may also be present [2, 5]. Patronas *et al.* [10] reported spinal cord and/or canal tumors in 31 patients (63%) in a series of 49 patients with NF2. In the same report, 26 patients (53%) had intramedullary lesions, 27 patients (55%) had intradural extramedullary tumors, and 22 patients (45%) had at least one tumor of each type. We detected two intramedullary enhancing small lesions in the proximal cervical spinal cord in our patient. We primarily suspected ependymomas or intraparanchimal schwannomas.

In this report, we have presented the case of an NF2 patient with bilateral vestibular and trigeminal schwannomas, right-sided facial schwannoma, multiple meningiomas, and two intramedullary spinal cord lesions. This case includes all the probable intracranial and spinal mass lesions which may be associated with NF2. According to our knowledge, no other paper has reported such a case with vestibular, trigeminal, and facial schwannomas, and their association with meningiomas and intramedullary spinal cord tumors in the same patient.

References

[1] Evans DG, Huson SM, Donnai D, Neary W, Blair V, Newton V, *et al*. A clinical study of type 2 neurofibromatosis. Q J Med. 1992; 84: 603-18.

[2] Mautner VF, Tatagiba M, Lindenau M, Fünsterer C, Pulst SM, Baser ME, *et al*. Spinal tumors in patients with neurofibromatosis type 2: MR imaging study of frequency, multiplicity, and variety. AJR Am J Roentgenol. 1995; 165: 951-55. PMid: 7676998. http://dx.doi.org/10.2214/ajr.165.4.7676998

[3] Rosen DR, Sapp P, O'Regan J, McKenna-Yasek D, Schlumpf KS, Haines JL, *et al*. Genetic linkage analysis of familial amyotrophic lateral sclerosis using human chromosome 21 microsatellite DNA markers. Am J Med Genet. 1994; 51: 61-69. PMid: 7913294. http://dx.doi.org/10.1002/ajmg.1320510114

[4] Gutmann DH, Aylsworth A, Carey JC, Korf B, Marks J, Pyeritz RE, *et al*. The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2. JAMA. 1997; 278: 51-7. PMid: 9207339. http://dx.doi.org/10.1001/jama.1997.03550010065042

[5] Spilberg G, Marchiori E, Gasparetto EL, Cabral RF, Takayassu TC, Batista RR, *et al*. Magnetic resonance findings of neurofibromatosis type2: a case report. Cases J. 2009; 2: 6720. PMid: 19829851. http://dx.doi.org/10.4076/1757-1626-2-6720

[6] Jain A, Suri V, Sharma BS, Sharma MC. Mixed schwannoma with meningioma of the trigeminal nerve. Indian J Pathol Microbiol. 2010; 53: 769-71. PMid: 21045412. http://dx.doi.org/10.4103/0377-4929.72084

[7] Novak MA. Hearing Loss in Neurotologic Diagnosis, in Neurotology. Jackler RK, Brackman DE, Editors. 1994, Mosby Year Book Inc: St Louis. P: 131-44.

[8] Mautner VF, Lindenau M, Baser ME, Hazim W, Tatagiba M, Haase W, *et al*. The neuroimaging and clinical spectrum of neurofibromatosis 2. Neurosurgery. 1996; 38:880-5; discussion 885-6.

[9] Aoki S, Barkovich AJ, Nishimura K, Kjos BO, Machida T, Cogen P. Neurofibromatosis types 1 and 2: cranial MR findings. Radiology. 1989; 172: 527-34.

[10] Patronas NJ, Courcoutsakis N, Bromley CM, Katzman GL, MacCollin M, Parry DM. Intramedullary and spinal canal tumors in patients with neurofibromatosis 2: MR imaging findings and correlation with genotype. Radiology. 2001; 218: 434-42.