CASE REPORT

Neurofibromatosis II: A Disease of Multicentric Central Nervous System Masses

Arvinder Singh[1], Manjeet Kaur[2], Sumanjeet Kaur[3]

Abstract

Neurofibromatosis (NF) is an autosomal dominant disorder with two reported types. Neurofibromatosis type I presents with the cutaneous manifestations and involves astrocytes and the neurons. Neurofibromatosis II involves meninges and the Schwann cells in the central nervous system with presentation as bilateral schwannomas, multiple meningiomas, and cord ependymomas.

Our present case was presented with bilateral hearing loss and chronic headache. MRI of the brain showed bilateral vestibular schwannomas extending up to bilateral cerebellopontine angle cisterns, mass lesion in relation to the right trigeminal nerve, multiple extra-axial dura-based mass lesions, posterior fossa mixed signal lesion, optic nerve sheath tumors, and two intradural intramedullary cervical cord ependymomas with surrounding cord edema.

Keywords: Magnetic resonance imaging, Neurofibromatosis, Schwannomas.

AMEI's Current Trends in Diagnosis & Treatment (2021): 10.5005/jp-journals-10055-0137

Introduction

Neurofibromatosis (NF) consists of genetic disorders in which there are benign and malignant tumors of the nervous system.[1] It is classified into two types, i.e., NF-I and NF-II, and may involve brain, spinal cord, and exiting nerves, bones, soft tissue, kidneys, and the mediastinum.[2]

Neurofibromatosis type I was first found by von Recklinghausen in 1882. This disorder was presented with neurocutaneous tumors and vasculitis. Neurofibromatosis type II is a relatively rare disorder, presenting frequently with central nervous system tumors.[3]

Notably, several patients have a range of organs affected as there is an extreme prevalence of multiple tumors happening in the same patient.[2] The diagnostic imaging techniques with precise clinical diagnosis help in distinguishing the above two types.[1] Magnetic resonance imaging (MRI) is the best imaging modality for the diagnosis and management of neurofibromatosis type I and II.[4]

[1]Department of Radiodiagnosis, Sri Guru Ram Das Institute of Medical Sciences and Research, Amritsar, Punjab, India

[2]Department of Physiology, Sri Guru Ram Das Institute of Medical Sciences and Research, Amritsar, Punjab, India

[3]Department of Radiodiagnois and Imaging, Sri Guru Ram Das Institute of Medical Sciences and Research, Amritsar, Punjab, India

Corresponding Author: Arvinder Singh, Department of Radiodiagnosis, Sri Guru Ram Das Institute of Medical Sciences and Research, Amritsar, Punjab, India, Phone: +91 9878655028, e-mail: arvinderdr@rediffmail.com

How to cite this article: Singh A, Kaur M, Kaur S. Neurofibromatosis II: A Disease of Multicentric Central Nervous System Masses. AMEI's Curr Trends Diagn Treat 2021;5(2):110–113.

Source of support: Nil

Conflict of interest: None

Case Descriptions

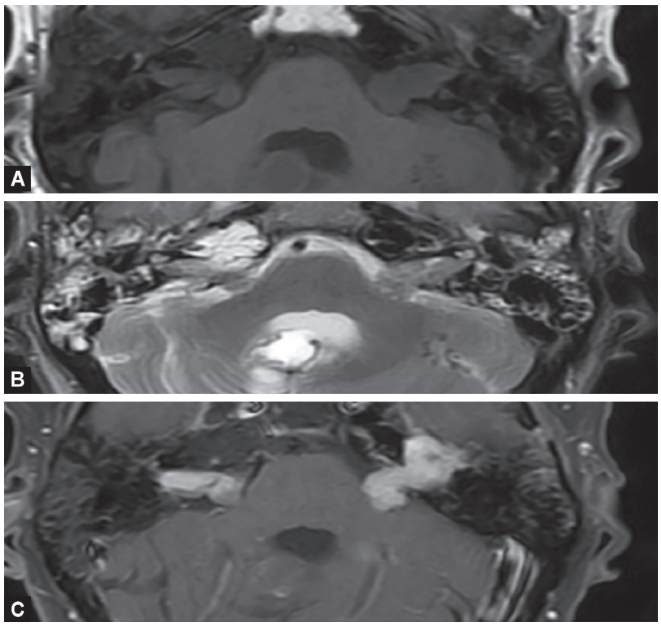
An 18-year-old male presented us with a history of progressive bilateral hearing loss, diminished visual acuity on the left side, chronic headache, vertigo, and past history of seizures since 1 year. He was normotensive, afebrile, and the pulse rate measured was 72 bpm at the time of general physical examination. There was no focal neurological deficit.

MRI brain showed mass lesions in relation to bilateral internal auditory canal causing their widening and extending into bilateral cerebellopontine (CP) angle cisterns. These lesions were hypointense on T1W and hyperintense on T2W images and showed intense postcontrast enhancement (Fig. 1). The lesion on right side measured about 2.2 × 1.0 cm and 2.0 × 1.0 cm on left side. About three dura-based extra-axial lesions were seen, one in right frontal and one each in parietal regions, appearing hypointense on T1W and hyperintense on T2W images with avid contrast uptake on T1 postcontrast images (Fig. 2). A well-defined homogeneously enhancing lesion was also seen in relation to the cisternal segment of the right trigeminal nerve which was not seen separately from this mass (Fig. 3).

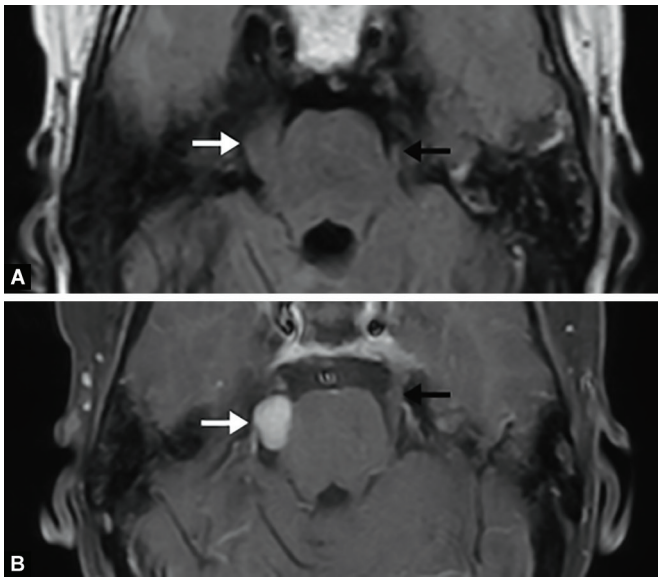
A solid cystic mass lesion measuring about 3.0 × 2.8 × 2.0 cm in size was seen in relation to the right cerebellar hemisphere abutting the fourth ventricle. The cystic component appeared hyperintense on T2W images and hypointense on T1W images. The solid component appeared heterogeneous on T2W images and T1W images and showed variable enhancement on postcontrast images (Fig. 4).

A T2W hyperintense mass lesion was seen around the left optic nerve measuring 2.0 × 1.7 cm in size which showed significant enhancement on the postcontrast study. The optic nerve was seen traversing through the lesion (Fig. 5).

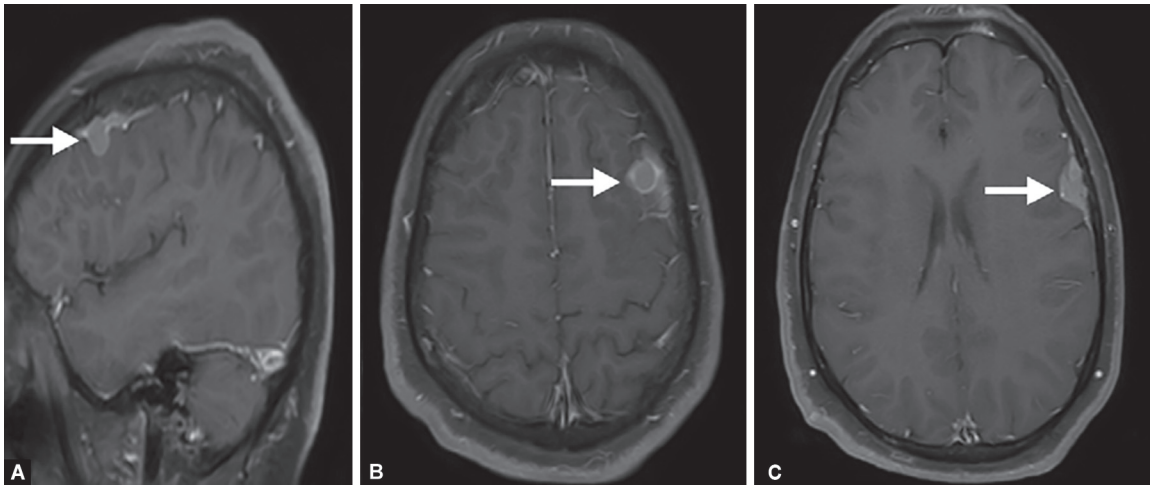
MRI evaluation of cervical spine revealed two intramedullary lesions opposite to C2 and C3 vertebral bodies, appearing isointense to hypointense on T1W, isointense to hyperintense on T2W images, and showed homogeneous enhancement on postcontrast study (Fig. 6).



Figs 1A to C: (A) Axial T1W image showing hypointense mass lesions in bilateral internal auditory canal and bilateral CP angle cisterns, causing widening of the bilateral internal auditory canal; (B) Same lesion appearing hypointense T2W images; (C) Axial T1 + C fat-suppressed image showing contrast uptake in the above-described lesion



Figs 3A and B: (A) Axial T1W image showing a well-defined hypointense lesion in relation to the cisternal segment of the right trigeminal nerve (white arrow). The right trigeminal nerve at this level is not separately seen; (B) Axial T1 + C FS image showing homogeneous enhancement in the right trigeminal nerve lesion. Normal traversing trigeminal nerve can be seen on the left side (black arrow)



Figs 2A to C: (A) Sagittal T1 + C FS image showing dura-based extra-axial lesion showing enhancement along the high parietal convexity; (B and C) Axial sagittal T1 + C FS images at different levels showing enhancing dura-based lesions

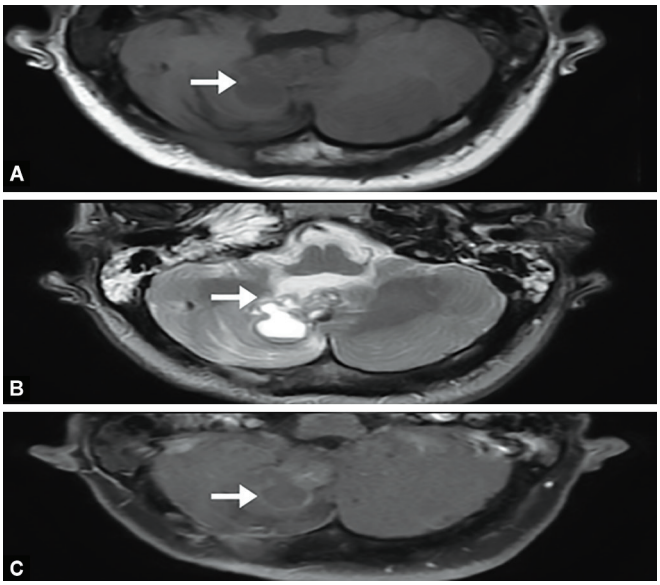
Discussion

Neurofibromatosis (NF) is an autosomal dominant disease with involvement of the cutaneous, nervous, and musculoskeletal systems.[5] It may manifest immediately after birth or later in the life.[2] It has been classified into two types, with chromosome 17 in NF-I and defect in chromosome 22 in NF-II.[5]

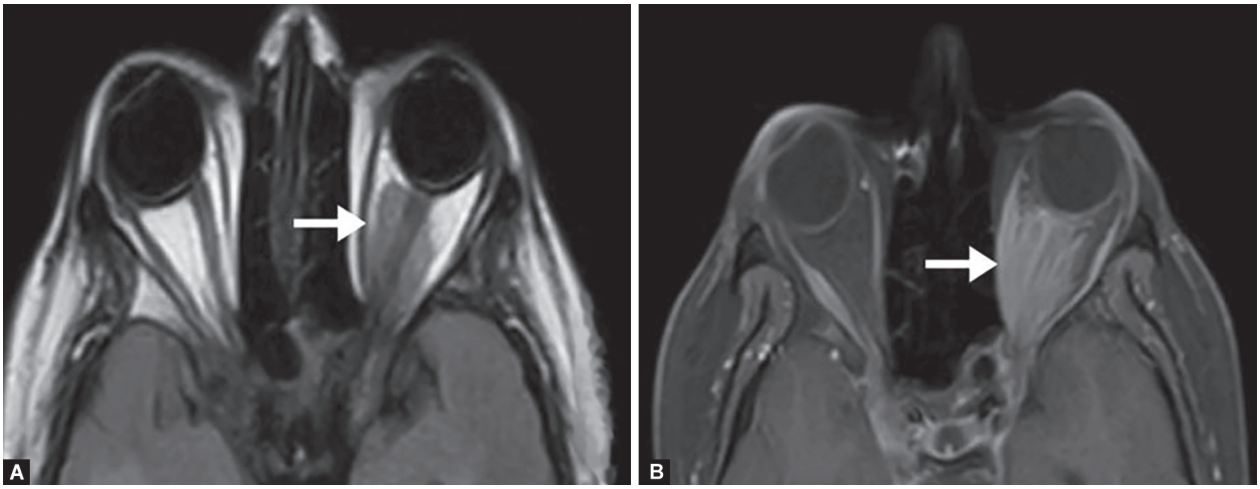
NF-I has a tumor-suppressor gene that encodes the protein neurofibromin, which normally regulates the cell growth and differentiation. NF-II encodes a tumor-suppressor gene which encodes the protein merlin that regulates the cell growth, especially the Schwann cells.[3]

NF-I involves multiple cell types and systems of the body.[4] The NF1 presents with cafe-au-lait spots and various cutaneous lesions with the involvement of CNS in about 15% of cases. It consists of optic nerve gliomas, cerebral, and brainstem astrocytoma.[2] The optic gliomas can affect any part of the optic pathway, with the involvement of one or both optic nerves.[4]

The most common benign neoplasms are neurofibromas with peripheral nerve sheath tumor as the most aggressive malignancy. The vascular abnormalities may involve the distal internal carotid and its branches. Skeletal manifestations consist of scoliosis, pseudoarthrosis of long bones, hemihypertrophy, and ribbon-shaped appearance of ribs.[2]



Figs 4A to C: (A and B) Axial T1W and T2W images showing a solid cystic lesion is seen in the right cerebellar hemisphere abutting the ependymal surface of the fourth ventricle posteriorly. The cystic component appears hypointense on T1W images and hyperintense on T2W images. Solid components appear isointense on T1W images and both hypointense and hyperintense on T2W images; (C) Axial T1 + C FS image showing heterogeneous enhancement in the solid component of the lesion



Figs 5A and B: (A) Axial T1W image showing a well-defined hypointense lesion in relation to the retrobulbar segment of the left optic nerve sheath; (B) This lesion shows enhancement on postcontrast images. However, the optic nerve is seen traversing through the lesion

NF-II presents with CNS lesions like bilateral acoustic schwannomas occurring primarily in relation to the internal auditory canal in about 95% of adult patients.[2,4] The definite diagnostic criterion for NF-II is bilateral vestibular schwannomas. Multiple meningiomas are reported in NF-II patients with variable size occurring in a younger age group.[4,5] Infrequently, the patient may present with Bell's palsy of the facial nerve.[6]

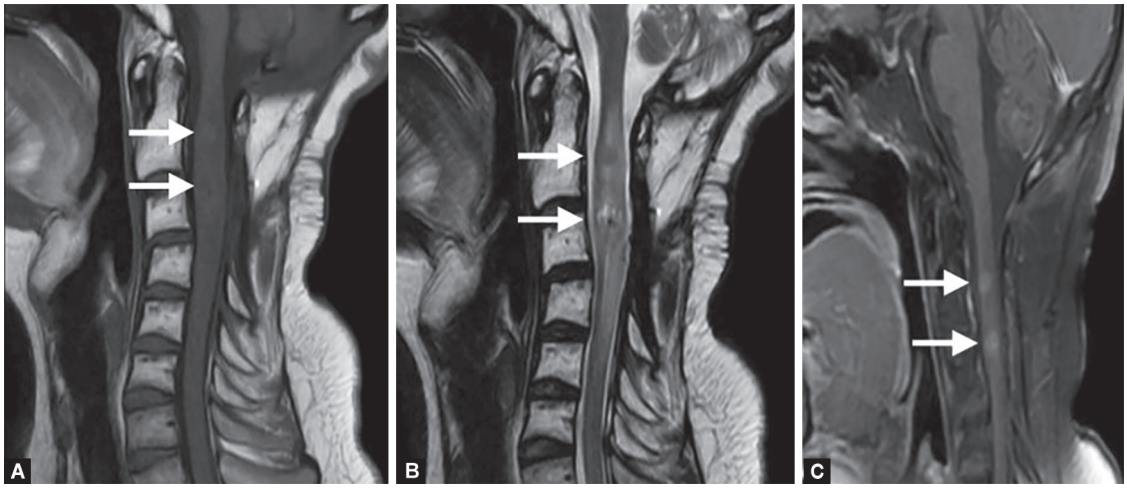
The most common intraspinal tumor is upper cervical cord intramedullary ependymomas followed by intraspinal schwannomas and meningiomas.[4] The meningiomas tend to occur in the thoracic spine.[7]

MR imaging findings in neurofibromatosis-I are often in the brain stem, cerebellum, and basal ganglia in the form of pilocytic astrocytomas, brain stem gliomas, plexiform neurofibromas, and orbital gliomas.[3]

Cervical spine ependymomas are usually centrally located within the cord and display an intense postcontrast enhancement.[4]

Our present case was presented with bilateral vestibular schwannomas, right and trigeminal mass multiple intracranial meningiomas, left optic nerve sheath meningioma, posterior fossa pilocytic astrocytoma, and two intramedullary cervical ependymomas. Though he had no family history, the presence of bilateral schwannomas is diagnostic for the NF-II syndrome, without the need for a biopsy.

The management of neurofibromatosis is by a multidisciplinary approach comprising surgery and radiotherapy.[6] The schwannoma patients can be treated with bevacizumab, which produces tumor regression.[8] Another option for small tumors is stereotactic radiotherapy. The intracranial meningiomas may be resected surgically except that of optic nerve sheath.[6]



Figs 6A to C: (A and B) Sagittal T1 and T2W imaging evaluation of cervical spine revealed two intramedullary lesions opposite to C2 and C3 vertebral bodies, appearing isointense to hypointense on T1W and isointense to hyperintense on T2W images. Adjacent cord edema is seen extending from C1-2 to C5-6 level; (C) Sagittal T1 + C FS image showing uptake of contrast in above-mentioned lesions

There is quite variability of disease progression in neurofibromatosis.[9] Due to various cranial and spinal neoplasms, there may be long-term morbidity and reduced life expectancy in NF-II patients. MRI scans are frequently required for management.[9,10]

Conclusion

The improved understanding of the clinical manifestations of neurofibromatosis in addition to better precision in neuroimaging and genetic tests has dramatically improved prompt diagnosis of patients. Early and correct diagnosis of the disease may be beneficial in preventing disabilities even in the next generation.

Orcid

Sumanjeet Kaur https://orcid.org/0000-0003-3290-2909

References

1. Campian J, Gutmann DH. CNS tumors in neurofibromatosis. J Clin Oncol 2017;35(21):2378–2385. DOI: 10.1200/JCO.2016.71.7199.
2. Hekmatnia A, Ghazavi A, Marashi Shooshtari MJ, et al. Imaging review of neurofibromatosis: helpful aspects for early detection. Iran J Radiol 2011;8(2):63–74. PMID: 23329919.
3. Borofsky S, Levy LM. Neurofibromatosis: types 1 and 2. Am J Neuroradiol 2013;34(12):2250–2251. DOI: 10.3174/ajnr.A3534.
4. Rodriguez D, Poussaint TY. Neuroimaging findings in neurofibromatosis type 1 and 2. Neuroimaging Clin N Am 2004;14(2):149–170. DOI: 10.1016/j.nic.2004.03.004.
5. Gangadhar K, Kumar S, Bhatia L, et al. A complete constellation of nervous system lesions of NF2: imaging evaluation. Case Rep Radiol 2012;2012:353179. DOI: 10.1155/2012/353179.
6. Swetha P, Omojola MF. Intracranial tumors in neurofibromatosis type 2: a review of magnetic resonance findings. J Radiol Radiat Ther 2014;2(1):1026.
7. Spilberg G, Marchiori E, Gasparetto EL, et al. Magnetic resonance findings of neurofibromatosis type 2: a case report. Cases J 2009;2(1):6720. DOI: 10.4076/1757-1626-2-6720.
8. Kollmann P, Mautner VF, Koeppen J, et al. MRI-based volumetric measurements of vestibular schwannomas in patients with neurofibromatosis type 2: comparison of three different software tools. Sci Rep 2020;10(1). DOI: 10.1038/s41598-020-68489-y.
9. Asthagiri AR, Parry DM, Butman JA, et al. Neurofibromatosis type 2. Lancet 2009;373(9679):1974–1986. DOI: 10.1016/s0140-6736(09)60259-2.
10. Rennie ATM, Side L, Kerr RSC, et al. Intramedullary tumors in patients with neurofibromatosis type 2: MRI features associated with a favorable prognosis. Clin Radiol 2008;63(2):193–200. DOI: 10.1016/j.crad.2007.08.003.