

CASE REPORT

INTRACRANIAL AND INTRASPINAL TUMOURS: REVIEW OF MRI FINDINGS IN A RARE NF-2 CASE

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ABSTRACT: Neurofibromatosis type 2 is a rare autosomal dominant syndrome, characterized by multiple intracranial and intraspinal tumours associated with ocular abnormalities. The most common tumor associated with the disease is the vestibule cochlear schwannoma, and as many as 10% of patients with this tumor have neurofibromatosis type 2. Based on clinical and imaging findings the diagnostic of neurofibromatosis type 2 can be made. In this report we aim to report a 24-year-old male who was evaluated for progressive hearing loss, vertigo, ataxic gait and right lower limb weakness. During the workup, cranial CT, Brain and whole Spine MRI was done which showed all the findings in one patient including bilateral vestibulocochlear schwannoma, multiple meningiomas, and intramedullary and extramedullary tumours in spinal cord.

KEYWORDS: Neurofibromatosis type 2, Magnetic resonance imaging, Schwannoma, Meningioma, Intramedullary, Extramedullary.

INTRODUCTION: Neurofibromatosis type 2 (NF2) is a genetic disorder characterised by the development of multiple schwannomas and meningiomas. The disease can be diagnosed when a pathogenic mutation in the NF2 gene is identified or when the criteria proposed by Gutmann et al. for diagnosis of the syndrome are fulfilled (Table 1).^[1] The term MISME has been proposed to the NF 2 syndrome, due to multiple inherited schwannomas (MIS), meningiomas (M) and ependymomas (E). Patients with NF2 may have cutaneous schwannomas that resemble skin tags, but they rarely have café-au-lait spots.^[1] In fact neurofibromatosis type 2 is a misnomer, as the predominant tumours are schwannomas and meningiomas.^[2]

Table 1: Gutmann et al.^[3]

Definite diagnoses of NF2

- I. Bilateral CN VIII schwannomas on MRI or CT scan (no biopsy necessary).
- II. First-degree relative with NF2 with unilateral early-onset CNVIII schwannoma (age <30 y).
- III. or First-degree relative with NF2 with any two of the following:
 - (1) meningioma.
 - (2) Glioma.
 - (3) Schwannoma.
 - (4) juvenile posterior subcapsular lenticular opacity (juvenile cortical cataract).

Presumptive diagnoses of NF2:

- (i) Early onset of unilateral CN VIII schwannomas on MRI or CT scan detected in patients younger than 30 years and one of the following:

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- (1) Meningioma.
- (2) Glioma.
- (3) Schwannoma.
- (4) juvenile posterior subcapsular lenticular opacity.

(ii) Multiple meningiomas (>2) and one of the following:

- (1) unilateral CN VIII schwannoma younger than 30 years of age.
- (2) 1 meningioma, glioma, schwannoma or lens opacity.

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

CASE PRESENTATION: A 24-year-old male presented in out-patient department with complaints of progressive bilateral hearing loss-4months, gait ataxia and vertigo for 2 months. He also had a history of right lower limb weakness and occasional occipital headache since 2 months. There was no history of bowel and bladder involvement and no history of fever. There was no positive family history. Brain Computed tomography (CT) scan, Brain and whole spine MRI showed multiple intracranial and intraspinal masses. Physical examination revealed an alert and well oriented person. There was no evidence of any skin lesion.

MATERIAL & METHOD:

- Brain & MRI brain & spine (Plain & Contrast) has been done at one time.
- Axial CT is performed with 16 slice CT scanner.
- The MR imaging is performed with a 1.5 Tesla SIEMENS Avanto. After the integration of head coil, the examination started in supine position.
- The sequences and the scan parameters;
- Brain: T1W1, T2W1, FLAIR axial, T1W1 Sag, FLAIR Cor, Diffusion, ADC, CISS; T1W1 Post contrast axial, Sag, Cor.
- Spine: T1W1, T2W1, axial, Sag, STIR Sag, Post contrast T1W1.

DISCUSSION: Neurofibromatosis 2 is a rare autosomal dominant neurocutaneous disorder. The term MISME has been proposed to the NF 2 syndrome, due to multiple inherited schwannomas (MIS), meningiomas (M) and ependymomas (E). NF has been divided into two types, defined according to the location of their genetic defect. In NF1, the defect is on chromosome 17, and in NF2, on chromosome 22.^[4,5] The clinical diagnosis may be difficult because of sparse or no skin findings in NF2 unlike characteristic findings in NF1. Presentation of NF2 usually occurs in the second or third decade of life, with a peak in the 20s. It has varied clinical manifestation but usually the first clinical sign in NF2 is a sudden hearing loss due to the development of bilateral or unilateral vestibular schwannomas. Other symptoms are deafness, tinnitus, dizziness, imbalance and Bell's palsy.^[6] At least two thirds of individuals with NF2 develop spinal tumors, which are often the most devastating and difficult to manage.^[5,6,7,8] Several authors have studied series of cases to try to define incidence of the tumors in cases of NF2.

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Mautner et al.^[5] studied 48 patients with NF2, in which the prevalence of findings were: vestibular schwannomas (CN VIII) in 46 (96%), spinal tumors in 43 (90%), posterior subcapsular cataracts in 30 (63%), meningiomas in 28 (58%), and trigeminal schwannomas in 14 (29%). Our patient also had bilateral vestibular schwannomas, bilateral trigeminal schwannomas, meningiomas and spinal tumors, however cataract was not present in our patient.

Patronas et al.^[7] reported a series of 49 patients with NF2 with spinal MR images, which demonstrated spinal cord and/or canal tumors in 31 (63%). 26 patients (53%) had intramedullary lesions, 27 patients (55%) had intraduralextramedullary tumors, and 22 patients (45%) had at least one tumor of each type. Our patient also had multiple spinal tumours besides bilateral CN VIII schwannomas, bilateral trigeminal schwannomas and meningiomas.

Aoki et al.^[9] reported cranial MR of 11 patients with all patients had acoustic schwannomas, 8 had other cranial nerve tumors (5 multiple and 3 single) and 6 had meningiomas (4 multiple and 2 single). The majority of intramedullary spine tumors in NF2 is ependymomas and arises in either the upper cervical cord or the conus. Meningiomas usually present as intradural extramedullary neoplasms that are very similar to spontaneous meningiomas and most commonly involve the thoracic spine; frequently they are multiple in numbers. Our patient also had bilateral CN VIII schwannomas, bilateral trigeminal schwannomas, multiple intracranial & intraspinal meningiomas, and intramedullary tumors.

Our patient had bilateral CN VIII schwannomas, bilateral trigeminal schwannomas, multiple intracranial & intraspinal meningiomas, two intramedullary tumor (Ependymoma /Astrocytoma), with no histopathological diagnosis. The schwannomas were the cause of hearing loss and vertigo. The multiple meningiomas may be cause of his headache and vertigo. The left parafalcine meningioma was the cause of right lower limb weakness. Although he had no family history, the image findings of bilateral acoustic schwannoma, multiple intracranial meningiomas, intramedullary and extramedullary tumor, suggested the diagnosis of NF2.

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Figure 1: CT axial sections showing bilateral vestibular schwannomas. Bone window CT base of skull axial section showing widening of internal auditory meatus (yellow arrows).



Figure 1

Figure 2: Vestibular schwannomas. Axial and coronal contrast enhanced T1-weighted MR images showing bilateral enhancing solid masses in the cerebellopontine angles, compressing the pons (a, b, c). In addition right cavernous sinus meningioma is seen (yellow arrow) (b). SWI image & phase image shows punctate hemorrhage in b/l vestibular schwannomas (d, e). CISS image shows better demonstration of intracannicular extension of the tumour (f).

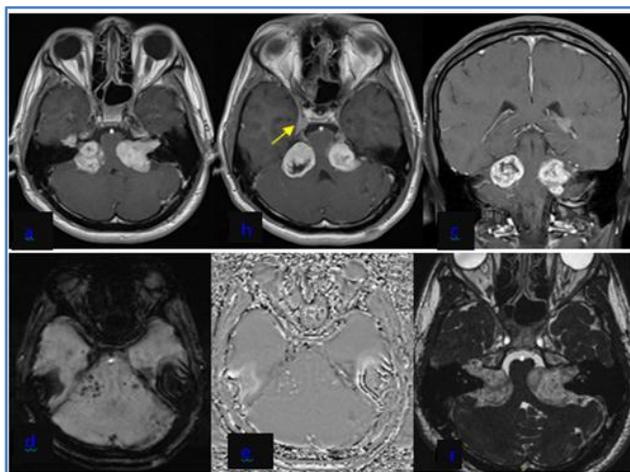


Figure 2

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Figure 3: Axial postcontrast & CISS images showing Right trigeminal schwannoma (white arrow) (a, d); Left trigeminal schwannoma (block arrow) (b, e); and right cavernous sinus meningioma (yellow arrow)(c, f).

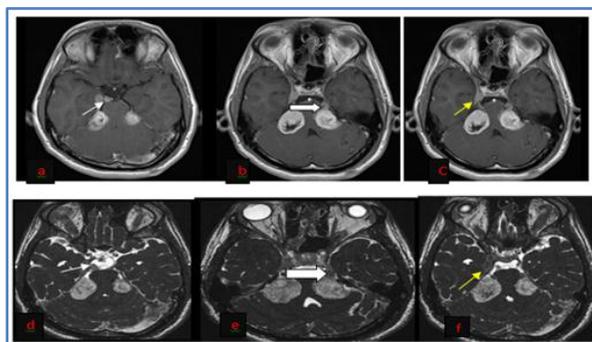


Figure 3

Figure 4: CT axial sections showing parafalcine and posterior falx meningioma.

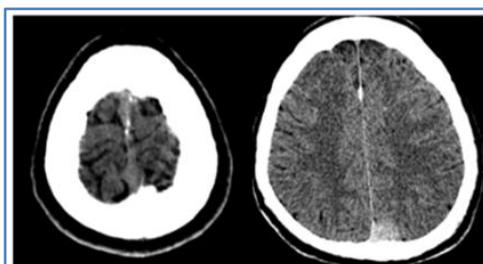


Figure 4

Figure 5: Multiple Meningiomas: Axial & Saggital contrast enhanced T1-weighted MR images showing showing parafalcine plaque & globose meningioma (a); meningioma along anterior & posterior falx (b, c); cerebral convexity frontal and parietal meningioma (d).

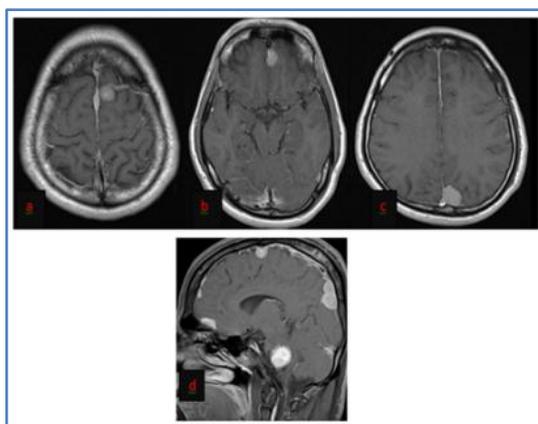


Figure 5

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Figure 6: Axial NECT and postcontrast MRI images showing hyperdense intra ventricular meningioma in left trigone(a); DW image shows restricted diffusion of left trigonal meningioma (yellow arrow)(b); showing enhancement on post contrast study(c).

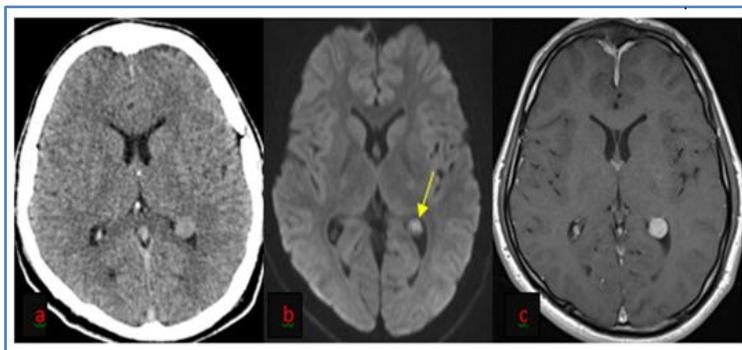


Figure 6

Figure 7: T1W postcontrast image showing enhancing intradural meningiomas (white arrow) (a,b); enhancing lesion within the dorsal cord, ependymoma/astrocytoma (yellow arrow) (c); T2W Sagittal image and T1W postcontrast image of dorsal lumbar spine showing a focal hyperintense intramedullary enhancing mass (Ependymoma or Astrocytoma) in conus at D12 –L1 level (yellow arrow)(d,e). In addition multiple T2 hypointense nodular enhancing masses along conus and filum terminale at D12-L1 and L3 levels suggestive of meningiomas (arrowheads).

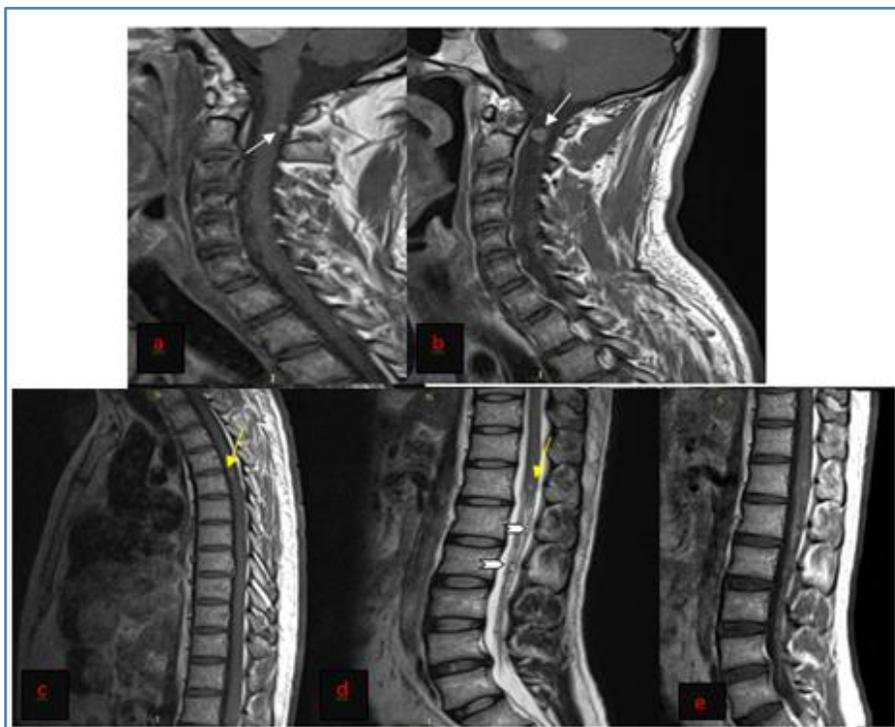


Figure 7

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Abbreviation:

NF: Neurofibromatosis

CT: computed tomography

MRI: Magnetic Resonance Imaging

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