Neurofibromatosis-II

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Citation

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Abstract

Neurofibromatosis type II is a phakomatoses with autosomal dominant inheritance characterized by intra-cranial schwannomas, meningiomas and ependymomas with associated peripheral nerve sheath tumors. Incidence is 1 in 30-40,000. Patients present with hearing loss, seizures, gait disturbances. Case of NF-II in a 25-year old girl having bilateral acoustic schwannomas, meningiomas and spinal schwannomas is presented here. Radiographs, C.T. Scan, MRI were performed and presumptive diagnosis of NF-II was considered.

INTRODUCTION

Neurofibromatosis type II (syn Acoustic neurofibromatosis, Bilateral vestibular schwannoma disease, Gardner type neurofibroma, Wishart type neurofibroma) is classified under the phakomatoses though cutaneous manifestations are uncommon in the type II subgroup.[1,2]

It is an autosomal dominant type of inherited disorder showing no racial or sexual predominance. The hallmark of the syndrome is multiple schwannomas – the most typical site being the VIII cranial nerve.

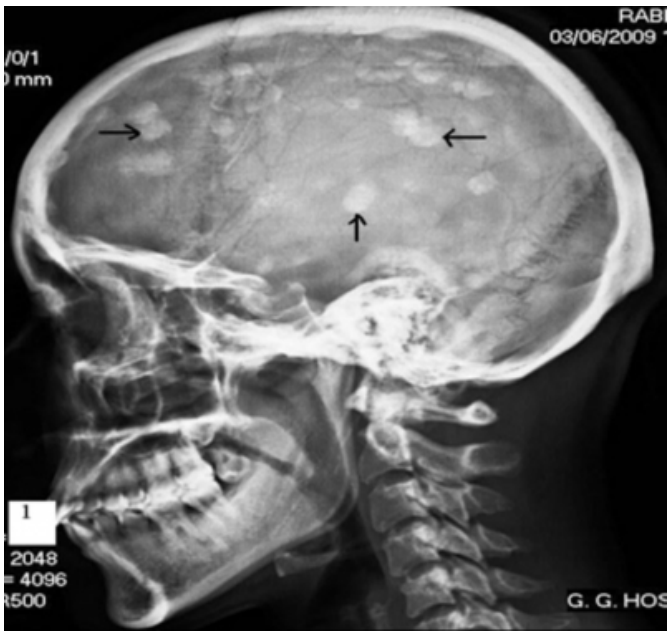
Previously called the central type of neurofibromatosis (as opposed to NF-I – the peripheral type), such distinctions are now considered obsolete as NF-II has peripheral manifestations and vice versa.

CASE REPORT

A 25 years old female patient presented with seizures and long term history of hearing loss and upper backache. Physical examination was unremarkable. Audiometry revealed bilateral sensorineural type of hearing loss.

Figure 1

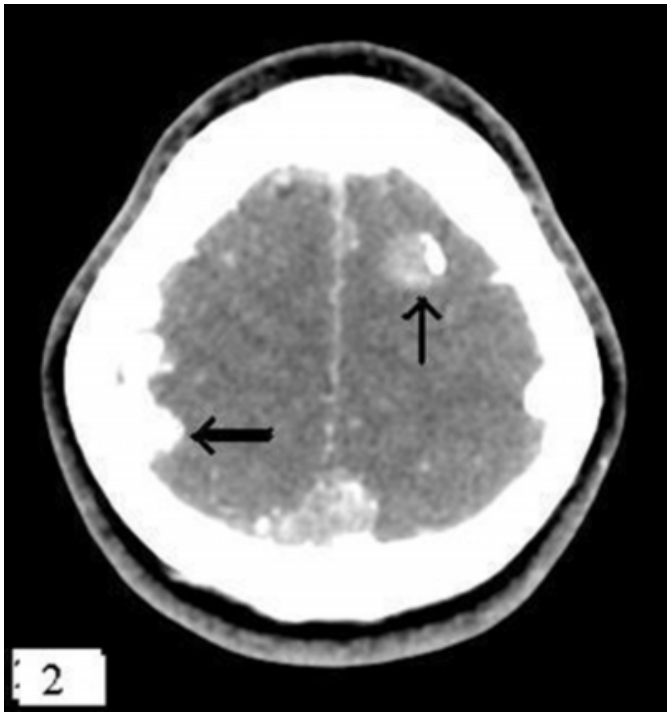
Figure 1: Radiograph of skull (lateral view) shows multiple radio opacities (arrows) on lateral Projection (fig 1).



Plain and contrast C.T. study of brain performed on GE Healthcare 16-slice scanner showed two enhancing structures at the cerebello-pontine angle extending into the internal acoustic meatus through the Porus Acousticus with extra-axial rounded enhancing lesions in the falx and over the cerebral convexities with underlying bony hyperosteosis (fig 2).

Figure 2

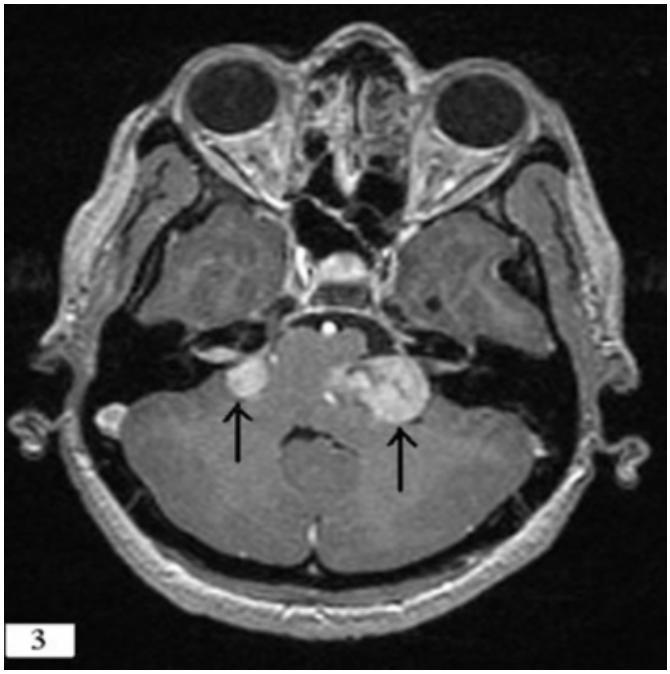
Figure 2: Brain axial contrast C.T. scan shows multiple enhancing lesions (thin arrow) in falx and cerebral convexities with bony hyperosteosis(thick arrow)



MRI study of the brain and spine performed on 1.5T Siemens Magnetom Essenza machine which included T2 weighted axial sequences and 3D contrast enhanced T1 sequences showed enhancing extra-axial mass lesion at the cerebello-pontine angle extending into the internal auditory canal with displacement of the brainstem producing compression effect over the 4th ventricle (fig 3).

Figure 3

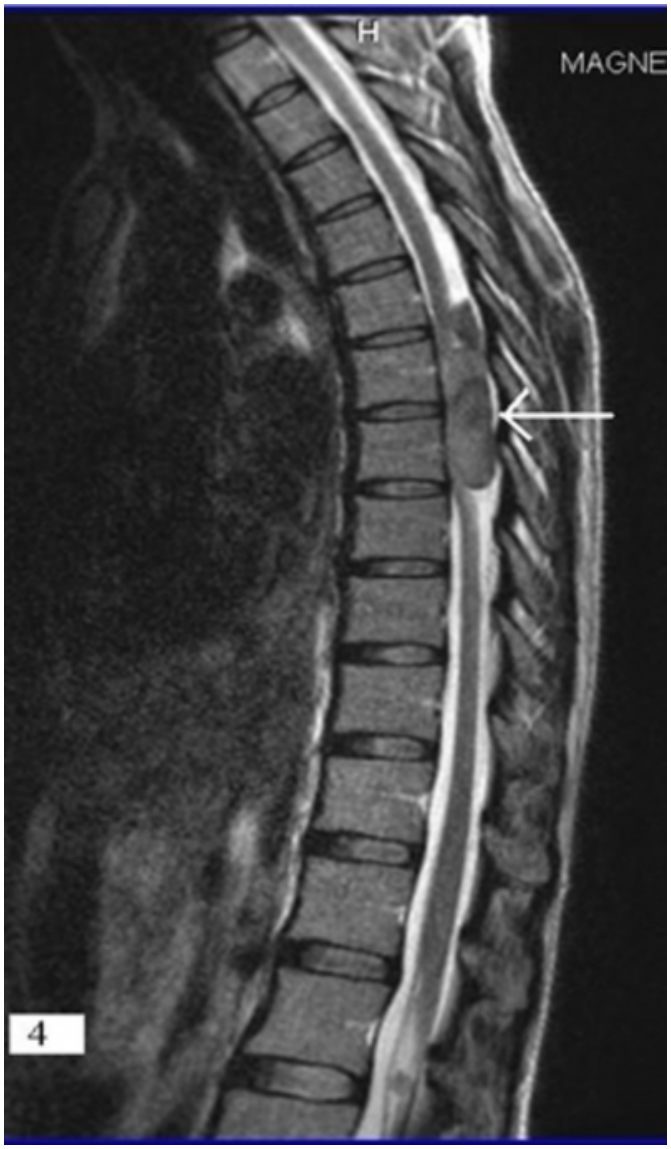
Figure 3 : MRI brain (T1 axial post-contrast) shows enhancing extra-axial lesions at cerebello-pontine angle (arrows).



T2 weighted sagittal study of spine showed intra-dural extra-medullary mass lesion dorsal to the thoracic part of spinal cord, pushing it anteriorly (fig 4)

Figure 4

Figure 4 : T2W sagittal scan of spine showing intra-dural extra-medullary mass lesion, dorsal to the thoracic spinal cord (arrow)



Presumptive diagnosis of NF-II was considered after applying National Institute of Health consensus criteria for NF-II.

DISCUSSION

One of the rarer varieties of phakomatoses, NF II has an incidence of about 1 in 30 to 40 thousand and a prevalence of about 1 in 2,10,000. Inherited as an autosomal dominant disease, the genetic defect has been identified as deletions from chromosome 22. Persons with this abnormal chromosome 22 are predisposed to schwannoma, meningiomas, ependymomas, intracranial non-tumoral calcification.[2,3,4]

Cutaneous manifestations like neurofibromas and schwannomas are uncommon in NF-II as compared to NF-I and they rarely have café-au-lait spots.

Most classical site of schwannoma associated with this syndrome is the VIIIth cranial nerve, followed by the Vth cranial nerve.

Mostly presenting in the 2nd to 3rd decade, the typical clinical features are related to symptoms related to the effects of an intracranial SOL and the disturbed anatomy of the VIIIth cranial nerve - like hearing loss, imbalance and seizures.[5] In females, an exacerbation in the clinical severity is noted during pregnancy.[6] The vestibular schwannomas associated with this are invasive (as opposed to sporadic vestibular schwawnnomas).[7,8]

Patients of NF-II may also show ocular pathologies in the form of early onset juvenile posterior sub-capsular cataract[9,10,11,12,13], retinal and choroidal hamartomas, optic nerve sheath meningiomas all of which may also affect patient's vision.

NATIONAL INSTITUTE OF HEALTH CONSENSUS CRITERIA FOR NF-II

DEFINITE DIAGNOSIS OF NF2

Bilateral CN VIII schwannomas on MRI or CT scan (no biopsy necessary)

First-degree relative with NF2 and either unilateral early-onset CN VIII schwannoma (age <30 y) or any 2 of the following:

Meningioma

Glioma

Schwannoma

Juvenile posterior subcapsular lenticular opacity (juvenile cortical cataract)

PRESUMPTIVE DIAGNOSIS OF NFII

Early onset of unilateral CN VIII schwannomas on MRI or CT scan detected in patients younger than 30 years and 1 of the following:

Meningioma

Glioma

Schwannoma

Juvenile posterior subcapsular lenticular opacity

Multiple meningiomas (>2) and unilateral CN VIII schwannoma or 1 of the following:

Glioma

Schwannoma

Juvenile posterior subcapsular lenticular opacity

Plain radiographs of the skull – AP and lateral projections may show multiple scattered radio-opacities due to underlying hyperosteosis of the skull vault. Widened internal acoustic meatus maybe noted in typical cases in the Rhese view.

On C.T. is noted enlargement/deformity of internal acoustic canal in more than 70% cases.[15, 16]. Prominent enhancement is noted following contrast administration. The lesion often appears heterogenous with cystic, haemorrhagic changes.[17,5,18,19] There maybe noted involvement of more than one cranial nerve.

The meningiomas are multiple, dural based masses, associated with underlying hyperosteosis. Can present in the lateral ventricles (as opposed to sporadic meningiomas). Striking feature is the younger age of presentation and multiplicity, both of which pronounce a bad prognosis. The enhancement pattern is typically less than that of schwannomas.

Often associated are spinal tumours like schwannomas and meningiomas amongst extramedullary ones and ependymomas amongst intramedullary ones. However, intramedullary schwannomas do occur. Schwannomas are noted mostly dorsally to the spinal cord, compressing the cord anteriorly and can extend from a site of origin in the nerve root.

On MRI, T2 weighted sequences show bilateral acoustic schwannomas at the cerebello-pontine angle extending into the internal auditory canal producing ice cream cone appearance and associated cystic and haemorrhagic changes. Post contrast T1 images show intense enhancement.

Meningiomas appear as dural-based, extra-axial lesions with underlying hyperosteosis. They are iso-intense to cortex on T1WI and iso-to-hyperintense to the cortex on T2WI. Post-contrast study shows moderate enhancement.

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