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(CASE REPORT)

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A case of neurofibromatosis type 2 with classical intracranial findings on MRI

Saumya Soni [1] * and Matendra Singh Yadav [2]

[1] *Department of Radiology, All India institute of medical sciences, Gorakhpur, India.*
[2] *Department of Anesthesiology. Uttar Pradesh University of Medical Sciences, Saifai, India.*

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Abstract

Neurofibromatosis type 2 is an autosomal dominant inherited syndrome, characterized by multiple neoplasms of the central and peripheral nervous system along with cutaneous and ocular abnormality. Up to 10% of patients with vestibulocochlear schwannoma (VIII cranial nerve) may have neurofibromatosis type 2. In the article, we are presenting a case of a 23-year-old pregnant female who presented with a history of bilateral gradual hearing loss. Her magnetic resonance imaging shows bilateral vestibulocochlear schwannoma, multiple meningiomas as well as few other lesions. Based on the imaging findings, diagnosis of neurofibromatosis 2 was made.

Keywords: Vestibular Schwannoma; En-Plaque Meningioma; Microhamartoma; Ubos

1. Introduction

With an incidence of 1 in 32,000 to 41,000, neurofibromatosis type 2 is a relatively rare neurocutaneous syndrome, which have similar incidence in both male and female [1]. Prior to 1987, neurofibromatosis type 1 and type 2 were thought to be one and the same disease. However, it was later shown that the two conditions were caused by distinct chromosome alterations. Neurofibromatosis type 2 shows autosomal dominant mode of transmission, and is associated with mutation at chromosome 22q12. About half of cases are thought to be inherited, and the other half are thought to be caused by sporadic, de novo mutations [2].

We are going to present a case of neurofibromatosis 2, diagnosis of which was made using the patient's medical history, clinical symptoms, and magnetic resonance imaging (MRI) results.

2. Case presentation

A pregnant female patient, age 23, came to the hospital with gradual hearing loss in her bilateral ear that had been present for 3 months. No history of otorrhea, otalgia, or ear trauma was present. There was no family background or contributing past. The person had no history of seizures or other motor or sensory problems.

During general examination, few café-au-lait macule was found on the anterior abdominal and chest walls. Few soft, nodular cutaneous swellings were also noted in the trunk and limbs. Bilateral tympanic membranes and external auditory canals were found to be normal upon local ear examination. Right-sided sensorineural hearing loss was confirmed by audiometric testing.

For further evaluation, MRI brain was advised which showed extra-axial T2 hyperintense, broad-based heterogeneously enhancing mass lesions in bilateral cerebello-pontine angle with beak like extension into bilateral internal auditory meati, characteristic of bilateral vestibular schwannoma. Bilateral vestibular lesions were causing severe compression

* Corresponding author: Saumya Soni

of pons and medulla with partial effacement of fourth ventricle, leading to mild triventricular hydrocephalus and transependymal oedema (Fig 1).

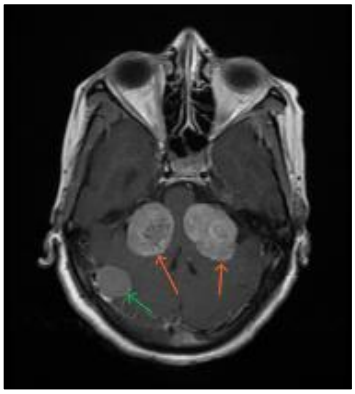

Figure 1 Axial T1+C image of MRI brain showing large heterogeneously enhancing lesions (orange arrow) at bilateral cerebello-pontine angles causing its compression. Also note the presence of a meningioma in posterior fossa (green arrow)

Multiple small extra-axial homogeneously enhancing lesions were also noted in right posterior fossa and along the falx which were characteristic of meningiomas. Contiguous irregular enhancing thickening of falx cerebri suggesting en-plaque meningioma (Fig 2)

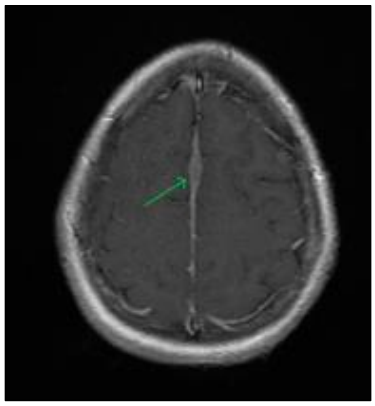

Figure 2 Axial T1+C image of MRI brain at high fronto-parietal region, irregular enhancing thickening along the falx cerebri - characteristic of en-plaque meningioma (green arrow)

Ill-defined T2/FLAIR hyperintense foci were noted in bilateral cerebral hemisphere, predominantly in right frontal lobe - These lesions may represent both microhamartomas or UBOs (Fig 3)

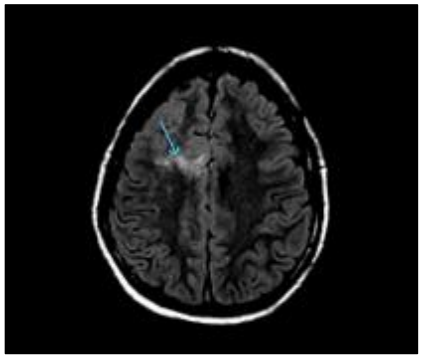

Figure 3 Axial FLAIR image of MRI brain at centrum semiovale level, showing irregular hyperintense foci in right anterior centrum semiovale, likely to represent microhamartomas or UBOs (sky-blue arrow)

Biopsy was done for the nodular lesions on trunk and were confirmed to be neurofibroma.

Patient was referred for the opinion of a neurosurgeon after being diagnosed with neurofibromatosis type 2 (NF2). The patient was offered surgery to treat vestibular schwannomas and to relieve obstructive hydrocephalus.

3. Discussion

Neurofibromatosis type 2 (NF2), a neurocutaneous disease is characterised by multiple tumours of the central and peripheral nervous systems. With an incidence of 1 in 60,000, it is extremely uncommon. The NF2 gene/Merlin/Schwanomin gene, which is found at band 12.2 (22q12.2) on the long (q) arm of chromosome 22, is linked to NF2. Similar to some members of the ERM (ezrin, radixin, and moesin) family of proteins, which are known for their function in connecting cytoskeletal components with cell membrane proteins, the Schwanomin gene encodes the cytoskeletal protein neurofibromin 2. Neurofibromin 2 interacts with cell surface proteins, controls ion transport, and alters cytoskeletal dynamics. This gene is fundamentally expressed by Schwann cells, meningeal cells, nerve cells, and lens cells (3).

The diagnostic criteria for NF2, which were first put up in 1988, are for either bilateral vestibular schwannomas or a family history of unilateral vestibular schwannoma together with any one of the following: posterior subcapsular opacities, meningioma, glioma, neurofibroma, or schwannoma. Other diagnostic criteria, such the Manchester criteria or the National Neurofibromatosis Foundation criteria, have been proposed, and these criteria have undergone numerous revisions and adjustments (4).

Bilateral schwannomas of the superior vestibular branch of the eighth cranial nerves are the disease's most characteristic feature. In addition to these, meningiomas, spinal canal tumours, and schwannomas of the other cranial nerves are present in the condition. The abbreviation, which stands for Multiple Intracranial Schwannomas, Meningiomas, and Ependymomas, is also known as MISME (5).

With a peak in the 20s, NF2 is typically diagnosed in the second or third decade of life. Although the clinical manifestation of NF2 varies, symptoms resulting from cranial nerve (CN) VIII schwannomas, such as hearing loss, tinnitus, balance impairment, and weakening in CN VII distribution, account for about 30 to 45 percent of patient diagnoses. This is because CN VIII schwannomas exhibit symptoms at a comparatively small size. Compression or stretching of the cochlear nerve, compression of the blood supply to the nerve or cochlea, or bleeding into the nerve or cochlea are the ways in which the tumour produces symptoms [5]. On rare instances, vestibular schwannomas can present with CSF rhinorrhoea, especially when concurrent aberrant arachnoid granulations are present (6).

Ependymomas make up the majority of intramedullary spine tumours in NF2, and they can develop in the conus or upper cervical cord. Most often affecting the thoracic spine, meningiomas manifest as intradural extramedullary neoplasms that resemble spontaneous meningiomas. Often, they are numerous in number. In a series of 49 individuals with NF2 who had spinal MR imaging, Patronas et al. [7] found that 31 (63%), had spinal cord and/or canal tumours. 22 patients (45%) had at least one tumour of each kind, 26 patients (53%) had intramedullary lesions, and 27 patients (55%) had intradural extramedullary tumours. In patients with neurofibromatosis, vascular complications like aneurysm and moyamoya syndrome may also be present which can predispose to intracranial haemorrhage (8)

Our patient had bilateral vestibular schwannomas, multiple meningiomas, unidentified bright objects (UBOs) and microhamartomas, these all findings are characteristic of neurofibromatosis type II. The schwannomas were the cause of his bilateral hearing loss. Although he had no family history, finding of bilateral schwannomas is diagnostic for the syndrome, without the need of a biopsy. Considering the ongoing pregnancy and presence of CSF flow obstruction, the patient was referred to higher centre for careful management of intracranial lesions in critical care set up (9).

List of abbreviations

- NF- Neurofibromatosis
- UBOs- Unidentified bright objects

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

References

[1] Evans D.G. Neurofibromatosis type 2 (NF2): a clinical and molecular review. Orphanet J Rare Dis. 2009;4:16. doi: 10.1186/1750-1172-4-16. [DOI] [PMC free article] [PubMed] [Google Scholar]

[2] Seth, N., Varshney, D. & Verma, S. Giant sacral schwannoma in a neurofibromatosis type 2 patient. Egypt J Radiol Nucl Med 54, 125 (2023). https://doi.org/10.1186/s43055-023-01071-4

[3] Tsukita S., Yonemura S. ERM (ezrin/radixin/moesin) family: from cytoskeleton to signal transduction. Curr Opin Cell Biol. 1997;9(1):70–75. doi: 10.1016/s0955-0674(97)80154-8.

[4] Baser M.E., Friedman J.M., Wallace A.J., Ramsden R.T., Joe H., Evans D.G. Evaluation of clinical diagnostic criteria for neurofibromatosis 2. Neurology. 2002; 59:1759–1765. doi: 10.1212/01.wnl. 0000035638. 74084.f4.

[5] Mautner VF, Lindenau M, Baser ME, Hazim W, Tatagiba M, Haase W. The neuroimaging and clinical spectrum of neurofibromatosis 2. Neurosurgery. 1996; 38:880–885. doi: 10.1097/00006123-199605000-00004.

[6] Dr. Namdev Seth, Dr. Mayank Srivastava, Dr. Saumya Verma. Vestibular schwannoma clinically presenting with CSF rhinorrhea: A case report. J Case Rep Sci Images 2024;6(2):56-60. DOI: 10.22271/27080056.2024.v6.i2b.94

[7] 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; 218:434–442. doi: 10.1148/radiology.218.2. r01fe40434.

[8] Kanani J, Sheikh M I. Exploring Nontraumatic Brain Hemorrhage in Sudden and Unexpected Deaths: A Novel Autopsy-Based Investigation. Asian Journal of Neurosurgery (efirst). doi:10.1055/s-0044-1800811

[9] Joshna K, Seth N, Abhishek HN, Thakur A and Akshaya NS The Pattern and Outcome of Obstetric Patients Getting Admitted in Surgical Critical Care Unit of Rural Tertiary Care Unit. Anaesth Critic Care Med J 2023, 8(1): 000219. DOI: 10.23880/accmj-16000219