CASE REPORT

Neurofibromatosis type 2: A challenging case report and multidisciplinary management approach

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Abstract

Neurofibromatosis type 2 (NF2) is a genetic disorder characterised by multiple benign tumours in the nervous system. We present the case of a 16-year-old female with NF2, seen on April 29, 2023, at Nishtar Hospital, Multan, exhibiting symptoms such as swollen left eye, chronic headache, decreased vision, bilateral hearing loss, tinnitus, and gait issues. Diagnostic evaluation, including Magnetic Resonance Imaging (MRI), confirmed NF2 diagnosis based on National Institutes of Health (NIH) and Manchester criteria. Surgical resection is the primary treatment, but the patient refused and opted for herbal treatment due to personal and socioeconomic reasons. The report highlights the challenges in NF2 management and the importance of patient preferences.

Keywords: Neurofibromatosis; Neurological Disorders; Radiology.

DOI: https://doi.org/10.47391/JPMA.21415

Introduction

Neurofibromatosis type 2 (NF2) is a rare genetic disorder characterised by the development of multiple benign tumours in the central and peripheral nervous system. It is caused by mutations in the NF2 gene, resulting in the loss of function of the tumour suppressor protein known as merlin.[1] NF2 affects approximately one in 25,000 individuals worldwide, making it a relatively uncommon condition.

The diagnosis of NF2 is based on clinical criteria, including the National Institutes of Health (NIH) criteria and the Manchester criteria.[2,3] The NIH criteria include bilateral vestibular schwannomas, unilateral vestibular schwannoma with a family history of NF2, or a combination of specific tumour types. The Manchester criteria also include bilateral vestibular schwannomas as a major criterion for the diagnosis of NF2.[4]

Early diagnosis is crucial for appropriate management and treatment planning. The management of NF2 involves a multidisciplinary approach, including surgical interventions, radiation therapy, and pharmacological approaches. However, the overall management is tailored to individual patients' condition and may vary, based on the extent and location of the tumour.[5,6]

Case Report

A 16-year-old female presented with a swollen and outwardly displaced left eye, chronic headache, pressing eye pain, significant hearing loss in both ears, and gait and balance issues, on April 29, 2023, at Nishtar Hospital, Multan. Physical examination revealed a visibly swollen left eye with decreased visual acuity. Cranial nerve examination showed bilateral sensorineural hearing loss. Central nervous system examination revealed gait ataxia and impaired coordination. Laboratory investigations, including tumour and inflammatory markers and metabolic panels, were within normal limits; genetic testing was refused by the patient due to it being locally unavailable and extensive cost. MRI of the brain and orbits revealed ring enhancing vestibular schwannomas along bilateral cerebellopontine angles, extending into the internal auditory meatus on both sides. Additionally, multiple meningiomas were observed along the interhemispheric fissure and in the left parasellar area (1.5-2.5cm) (Figure 1). A left intraconal orbital lesion (2.5cm) with significant enhancement encasing the optic nerve was identified, resulting in mild proptosis (Figure 2). These findings fulfilled the diagnostic criteria for neurofibromatosis type 2 (NF2) according to both the National Institutes of Health (NIH) and Manchester

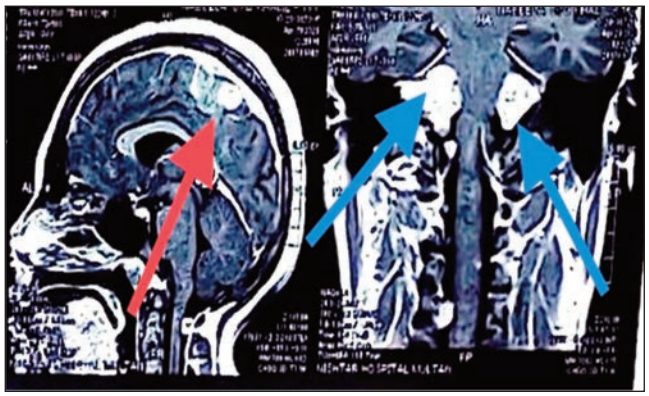

Figure-1: Interhemispheric Meningioma: Red arrow; Bilateral Cerebellopontine angle schwannomas: Blue arrows; T2W MRI.

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Submission completed: 16-07-2024 **1st Revision received:** 19-11-2024
Acceptance: 26-03-2025 **Last Revision received:** 25-03-2025

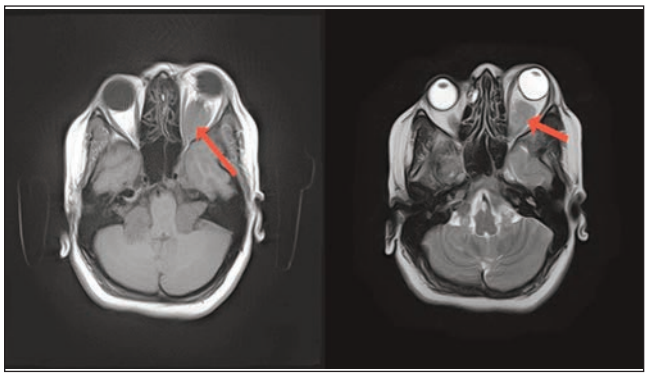

Figure-2: Left intraconal lesion showing significant enhancement, T1W MRI and T2W MRI, red arrows.

criteria .[2,3] Referral for an MRI of the spine was made to rule out spinal lesions. Ophthalmology and otorhinolaryngology consultations were requested to evaluate the left eye's condition and assess the extent of hearing loss. A CT scan was suggested as an adjunct to confirm the findings but was refused by the patient owing to lack of funds. Alternative treatment with herbal medications was chosen by the patient due to personal preference and socioeconomic constraints. The healthcare team employed several strategies to mitigate the patient's hesitation towards evidence-based treatment, such as building trust through open communication and addressing her specific concerns about potential side effects and costs. They explored potential compromises, such as combining conventional treatments with complementary therapies when appropriate. Involving family members in the decision-making process and providing comprehensive and understandable information were also considered. Despite these efforts, the team was unable to convince the patient to pursue evidence-based treatment.

Discussion

The MRI findings in our patient correlate with the diagnostic criteria of NF2 according to both the National Institutes of Health (NIH) and Manchester criteria.[5] According to the NIH criteria, NF2 can be diagnosed when either bilateral vestibular schwannomas are present or a unilateral vestibular schwannoma is accompanied by any two of the following: meningioma, glioma, schwannoma, or juvenile posterior subcapsular lenticular opacity. In the present case, the patient had bilateral vestibular schwannomas along with multiple meningiomas, fulfilling the NIH criteria. Similarly, the Manchester criteria for NF2 diagnosis state that either bilateral vestibular schwannomas are present or a unilateral vestibular schwannoma is accompanied by any two of the following: meningioma, glioma, schwannoma, neurofibroma, or juvenile posterior subcapsular lenticular opacity. In this patient, the presence of bilateral vestibular schwannomas, along with multiple meningiomas, met the Manchester criteria for NF2 diagnosis.

NF2 is known to have an autosomal dominant inheritance pattern, but in some cases, it can occur sporadically without a family history of the disease.[5] Our patient had a negative family history of NF2, suggesting a de novo mutation or incomplete penetrance.

The management of NF2 involves a multidisciplinary approach considering the location, size, and growth patterns of the tumours, as well as the patient's age, health status, and preferences. Surgical resection remains the mainstay of treatment for symptomatic or progressive tumours, aiming to alleviate symptoms and prevent complications. A comprehensive management plan for NF2 involves multidisciplinary care, including surgical interventions, symptom management, tumour surveillance, genetic counselling, and psychological support. Regular follow-up, tailored treatment, and collaboration among specialists are crucial for optimal patient outcomes.[6]

However, the patient's preference and socioeconomic constraints led to the decision of alternative treatment with herbal medications by a local non-qualified person. The healthcare approach for patients with NF2 should focus on providing accurate information, counselling patients regarding the risks and limitations of alternative treatments, and emphasising the importance of evidence-based medical care. Challenges in managing NF2 in resource-limited settings, such as Pakistan, further complicate patient care. Studies from Pakistan have highlighted the clinical features of NF2 and emphasised the challenges faced due to limited resources and access to specialised care.[7]

Conclusion

The case report illustrates the clinical challenges encountered in managing NF2 and emphasises the significance of diagnostic criteria, such as the NIH and Manchester criteria, in confirming the diagnosis. Pakistani studies have already contributed to our understanding of NF2 in the local population, shedding light on clinical features, genetic characteristics, and management challenges.[7,8] In a low-resource settings, such as the one in which the patient was diagnosed, the physicians prioritised providing the patient with access to basic diagnostic tools along with genetic counselling and educating the patient about their condition. However, the patient's own preferences and socioeconomic constraints lead the patient to seek alternative treatments.

Informed Consent: Appropriate informed medical consent was taken from the patient regarding the publishing of this case report.

Disclaimer: None.

Conflict of interest: None.

Funding disclosure: None.

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Author Contribution:

MU: Wrote the Abstract, introduction and conclusion portions, references portions of the case report and final approval.
SMSH: Collected the radiological imagery pertinent to the patient, wrote the legends, edited the figures and wrote the case report portion.
MB & SAM: Wrote the discussion portion.
RC: Wrote the discussion portion and checked the article for possible grammatical errors before submission.