International Journal of Science and Healthcare Research
Vol.8; Issue: 2; April-June 2023
Website: ijshr.com
Case Report *ISSN: 2455-7587*

Neurofibromatosis 2: A Case Report

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DOI: https://doi.org/10.52403/ijshr.20230265

ABSTRACT

Neurofibromatosis 2 (NF2) is caused by inactivating alterations in the *NF2* gene on chromosome 22q12.2. The 100-kb *NF2* gene is encoded by 17 exons. Patients with NF2 may present with distinctive "plaque-like" cutaneous schwannomas, ophthalmologic findings, or neurologic deficits such as hearing loss or peripheral neuropathies which may prompt further radiologic or genetic evaluation. Café au lait spots may be present in up to 50% of patients but they are typically fewer in number and not associated with freckling as in NF1. This is a case report of a 20 year old female who presented to us with left parapharyngeal schwannoma along with bilateral vestibular schwannoma.

Keywords: NF 2- Neurofibromatosis 2, VS- Vestibular Schwannoma, CECT-Contrast enhanced computed tomography, MRI- Magnetic resonance imaging

INTRODUCTION

Neurofibromatosis type II (NF2) is a tumor predisposition syndrome characterized by the development of distinctive nervous system lesions. NF2 results from loss-of-function alterations in the *NF2* gene on chromosome 22, with resultant dysfunction of its protein product merlin. NF2 is most commonly associated with the development of bilateral vestibular schwannomas; however, patients also have a predisposition to development of other tumors including meningiomas, ependymomas, and peripheral, spinal, and cranial nerve schwannomas.

Vestibular schwannomas (VS) are benign tumors that arise from Schwann cells of the vestibular portion of the vestibulocochlear nerve inside the internal auditory canal (intracanalicular). As they grow, they fill and extend beyond the internal auditory canal into the cerebellopontine (CP) angle (extracanalicular). VS result from genetic abnormalities on chromosome of 22q12 (the neurofibromatosis type 2 (NF2) gene coding for the tumor suppressor Merlin[1]. Approximately, 5% of cases occur as part of the tumor predisposition syndrome NF2, and very rarely as part of the tumor predisposition syndrome schwannomatosis. The remainders are thought to be sporadic due to acquired loss of NF2 gene function [2.]
The most common presenting symptoms are progressive hearing loss (90%) and tinnitus (>60%). Imbalance, dizziness, vertigo, facial paresthesia, and headache secondary to hydro- cephalus can occur with larger VS due to brainstem and trigeminal nerve compression. Up to 12% of patients can present with facial paresthesia due to involvement of the trigeminal nerve, and up to 6% can present with facial nerve palsy; again these symptoms occur in patients with larger VS[3,4]. The lifetime risk of developing a VS is estimated at approximately 1 in 1000, including sporadic and NF2-related tumors [5,6]. The incidence appears to be increasing, thought largely due to the incidental diagnosis of asymptomatic lesions with the increasing use of MRI and, to a much lesser extent, CT[7].
Neurofibromatosis type II (NF2), previously known as "central neurofibromatosis",

demonstrates a predilection for "central" intracranial and spinal lesions, most characteristically vestibular schwannomas
Schwannomas are typically brightly enhancing on post-gadolinium T1-weighted sequences While NF2-associated vestibular schwannomas typically exhibit similar morphologic features as sporadic tumors with a predominance of cellular Antoni A regions which may exhibit palisading nuclei and Verocay bodies and typically a lower proportion hypocellular Antoni B regions.

Case report

A 20 year old female patient presented to us with hoarseness of voice since 5 years and swelling in left oropharynx since 1 year with swelling in left upper neck since 6 months. On examination there is a 3*3 cm swelling in left upper neck 1 cm below the angle of mandible, non mobile, firm in consistency and non tender. Oropharyngeal examination shows a bulge in the left tonsillar fossa along with cleft palate that was present since birth. Bilateral tympanic membrane was normal. Nose and the rest of the examination was normal. BERA test depicts bilateral severe to profound sensory neural hearing loss.

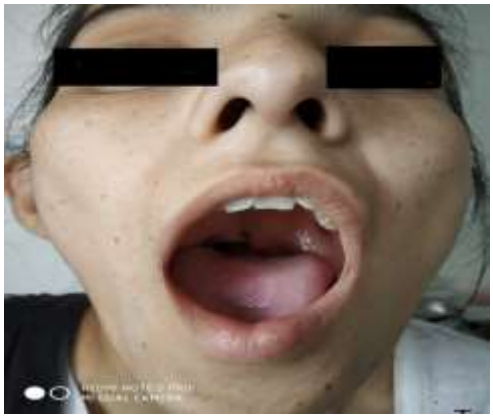

Image 1: clinical photograph of the patient showing left oropharyngeal bulge.

CECT of neck revealed presence of an oblong shaped lesion with predominantly cystic attenuation and few hypodense components within it. Lesion was found to occupy the left parapharyngeal space with extension into the left carotid space and left pharyngeal muscle space. Lesion follows the course of left carotid vessels. there was no surrounding inflammation suggesting a benign etiology.

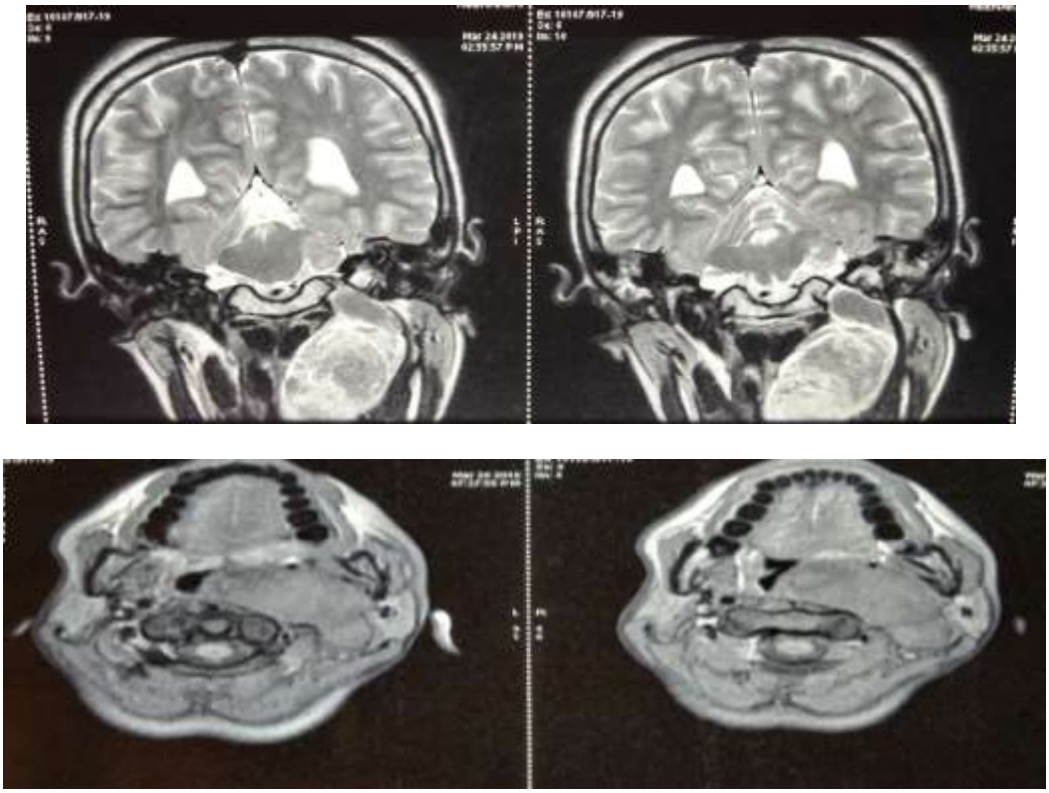

Image 2: MRI images showing lesion in the left parapharyngeal space with extension into carotid space and oropharynx.

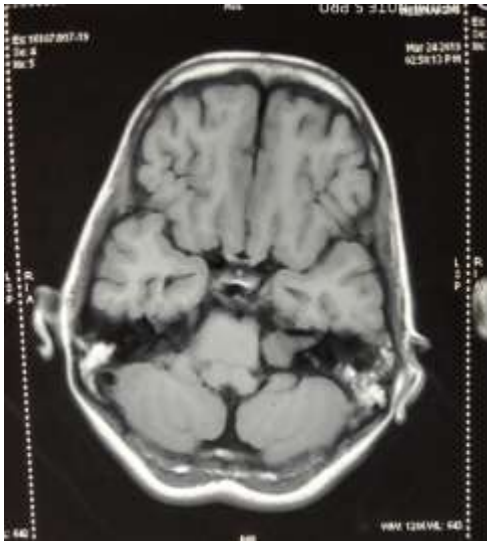

Image 3: MRI image showing left CP angle schwannoma (ice cream cone lesion).

MRI reveals a dumbbell shaped heterogenous circumscribed mass centred in posterior styloid compartment of left parapharyngeal space with extension into carotid space and oropharynx with widening of stylomandibular tunnel with anterior displacement of carotid vessels and lateral displacement of deep lobe of parotid findings consistent with left parapharyngeal schwannoma.

Bilateral cerebellopontine angles show bilateral vestibular schwannoma, left larger than right.

Fine needle aspiration cytology of the left oropharyngeal swelling reveals schwannoma.

Patient underwent excision of left parapharyngeal mass, biopsy report of the mass shows a biphasic tumor with hypercellular (Antoni A) and hypocellular (Antoni B) areas in collagen rich matrix consistent with schwannoma. The hypercellular areas show nuclear palisading around fibrillary processes with Varocay bodies.

Post operative after excision of the lesion the patient had left vagal and left hypoglossal nerve palsy.

Patient also underwent neurosurgery for excision of the left vestibular schwannoma.

DISCUSSION

There are various treatment options for patients with VS and NF2 .Observation 'wait and watch' Small tumors with no mass effect and no growth or slow growth , in patients with small tumors and serviceable hearing. Bevacizumab is used in tumor growing by at least 4 mm/year (or 60% by volume).

Surgery is done in large tumors with brainstem compression, and in rapidly growing tumors with a high risk of developing brainstem compression, deterioration in facial nerve function, deterioration in serviceable hearing.

Radiosurgery/ radiotherapy is used for small tumors without mass effect but demonstrable growth, in elderly patients with mild phenotype and in patient with high risk surgical candidate[8].

CONCLUSION

Patients with VS secondary to NF2 require a different management strategy from those patients with sporadic VS, with the goal of management being preservation of function and quality of life. When tumors are detected, the risks of treatment versus the risks of observation have to be carefully balanced. Strategies for managing patients with NF2 and VS are observation, surgery, SRS and stereotactic radiotherapy, and targeted therapies, particularly the vascular endothelial growth factor (VEGF) inhibitor bevacizumab.

Declaration by Authors

Acknowledgement: None

Source of Funding: None

Conflict of Interest: The authors declare no conflict of interest.

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How to cite this article: Richa Mehra, Varun Sachdeva. Neurofibromatosis 2: a case report. *International Journal of Science & Healthcare Research.* 2023; 8(2): 495-498. DOI: *https://doi.org/10.52403/ijshr.20230265*

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