



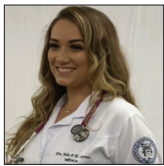
Case Report

Giant dumbbell C2C3 neurofibroma invading prebulbar cistern: Case report and literature review

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ABSTRACT

Background: Neurofibromatosis 1 (NF1) has a broad spectrum of clinical manifestations, most typically involving café-au-lait spots and skin neurofibromas. Only 2% of patients with NF1 have symptomatic spinal tumors.

Case Description: A patient with a previous diagnosis of NF1 presented with cervicalgia, dysphagia/mild dysphonia, gait alteration, and progressive hypoesthesia involving all four limbs. The magnetic resonance documented a giant dumbbell neurofibroma arising between the C2 and C3 levels which extended toward the foramen magnum, causing medullary and bulbar compression. The major challenge of surgical management was the enormous size and location this C2–C3 (5 cm × 4 cm × 5.1 cm) lesion.

Conclusions: Compression of the foramen magnum attributed to a dumbbell giant spinal neurofibroma at the C2C3 level resulting in prebulbar cisterns should be among the differential diagnostic considerations for patients presenting with tetraparesis and underlying NF1.

Keywords: Dumbell neurofibroma, Giant neurofibroma, Neurofibromatosis 1, Spinal tumors

INTRODUCTION

Type 1 neurofibromatosis (NF1) is an autosomal dominant genetic disorder caused by biallelic suppression of the NF1 tumor suppressor gene, located on chromosome 17. It occurs in 1/3,500 individuals and carries a 3%–15% mortality rate.^[3,8] NF1 occurs in all races, and some report a small female preponderance.^[1,2,5] Only 2% of patients with NF1 have symptomatic spinal tumors.

CASE REPORT

A 38-year-old male presented with a 4-year history of cervicalgia, dysphagia/mild dysphonia, gait alteration, and progressive hypoesthesia of all four extremities. The patient had a history of type I NF1 diagnosed 20 years previously. On examination, the patient exhibited cutaneous nodules/café-au-lait spots scoliosis, and a Grade 4 tetraparesis characterized by diffuse hyperreflexia with bilateral Babinski signs [Figure 1].

Radiographic analysis

The cervical magnetic resonance imaging (MRI) demonstrated a solid, nodular, expansive lesion originating at the C2–C3 (5 cm × 4 cm × 5.1 cm) level with extension toward the foramen magnum. It contributed to medullary and bulbar compression [Figure 2]. A secondary lesion was visualized at the C4–C5 level on the right.

Surgical intervention

The surgical approach included a right lateral suboccipital craniectomy, removal of the posterior C1 arch, and C2

laminectomy for resection of the dumbbell C2–C3 lesion. This was accompanied by a right C4–C5 hemilaminectomy for excision of the secondary tumor. Gross total lesion resection of the C2–C3 dumbbell neurofibroma was accomplished. Partial removal of the C4–C5 secondary lesion was performed as this was benign, and it allowed for the preservation of stability [Figure 3]. Somatosensory evoked potential and motor evoked potential monitoring did not change during the procedure. Postoperatively, the patient's tetraparesis progressively improved and hypoesthesia resolved. The postoperative magnetic resonance (MR) scan obtained revealed satisfactory resection of both lesions [Figure 4].



Figure 1: Cutaneous nodules café-au-lait spots.

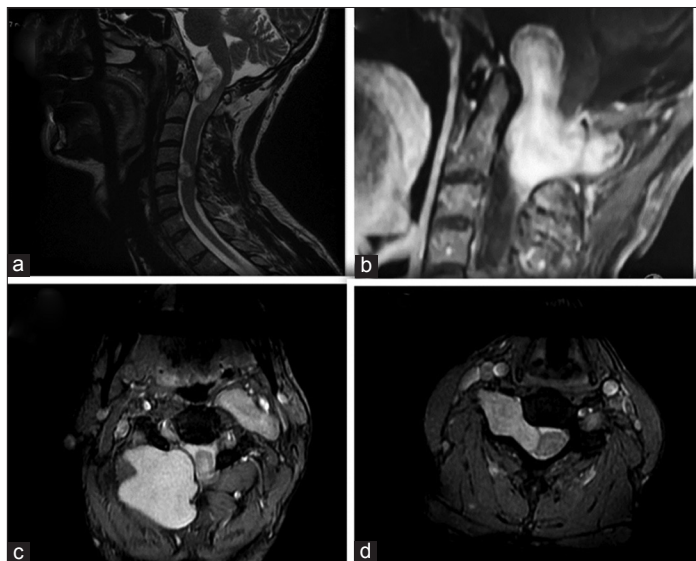


Figure 2: (a) Cervical magnetic resonance imaging – T2 with solid nodular expansive lesion originating from the level of C2–C3 (5 cm × 4 cm × 5.1 cm) with extension toward the foramen magnum, causing medullary and bulbar compression and another lesion from the level of C4–C5. (b) Dumbbell type formation intradural and extramedullary. (c) Represents an extradural component of C2C3 lesion. (d) Anterolateral lesion of smaller size located anterolateral right.

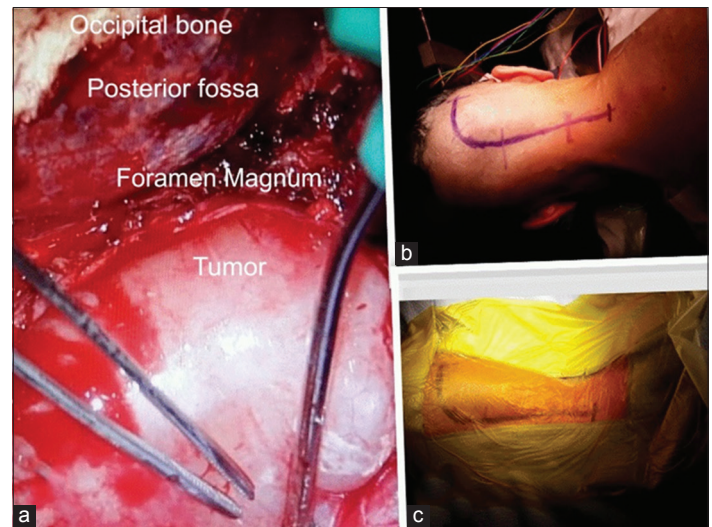


Figure 3: (a) Intraoperative period showing tumoral lesion after removal of the posterior C1 arch and C2 laminectomy. (b) Electrode placement for somatosensory monitoring intraoperatively. (c) Aseptic technique for surgery beginning.

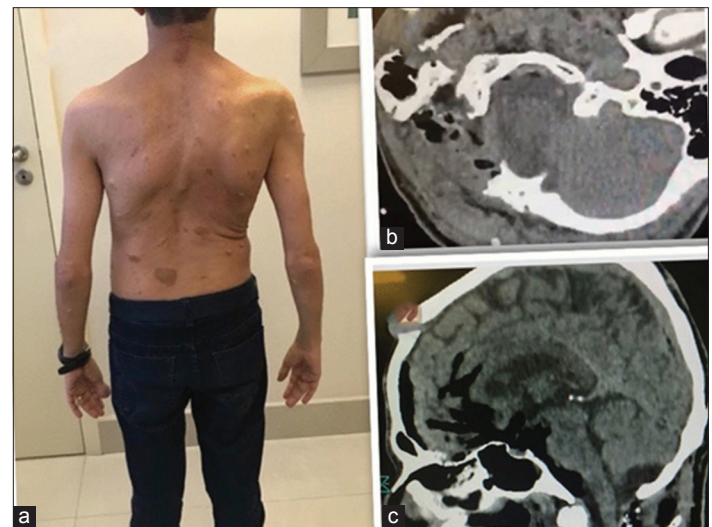


Figure 4: (a) Patient presenting good scarring process after 30 days of the surgery. (b) Postoperative control computed tomography showing lateral suboccipital approach. (c) Noncontrast magnetic resonance imaging T1 showing regression of the brainstem compression.

The immunohistochemistry documented an NF1, for example, negative epithelial membrane antigen (EMA), CD34 positive, KI-67 (MIB-1) negative, positive S100 Protein, and positive neurofilament.

DISCUSSION

Genetic transmission or spontaneous mutation

NF1 may either be genetically inherited (e.g., autosomal dominant) or, more frequently, may occur as a new mutation. The NF1 gene encodes a protein called neurofibromin, which is a tumor suppressor. Changes in expression of the NF1 gene contribute to tumorigenesis and the formation of neurofibromas. They develop around the peripheral nerves, often manifesting as encapsulated masses, and are localized in the dermal and/or subcutaneous tissues. Others may present as plexiform neurofibromas involving peripheral nerve roots.^[4]

Varied phenotypes

NF1 tumors present with varied clinical phenotypes characterized by; café-au-lait spots (95% single/multiple skin lesions) and neurofibromas.^[3,6] Plexiform neurofibromas occur in 30% of patients who present with multiple neurofibromas, diffusely involving peripheral nerves. Of the paraspinal neurofibromas, 72% are intradural and extramedullary and 14% are only extradural, while 13% are dumbbell lesions.^[3]

Clinical presentation

With NF1, spinal tumors occur 40% of the time. Notably, only 2% are symptomatic, often correlated with progressive quadriparesis, along with neck pain and urinary incontinence. Here, the patient had MR-documented medullary compression invading the prebulbar cistern attributed to the C2C3 lesion, plus a secondary C4–C5 tumor contributing to right-sided cord compression.

MR findings of neurofibromatosis 1 lesions

NF1 findings on MRI, as in this case, include iso or hyperintensity on T1, hyperintensity on T2, and homogeneous enhancement with gadolinium of T1 images.^[4]

Pathological/histological and immunohistochemistry findings with neurofibromatosis 1 tumors

Microscopically, NF1 lesions are comprised of Schwann cells, fibroblasts, and perineural cells which typically permeate between/separate the axons from the nerves. The immunohistochemistry commonly shows; S100 positive and EMA negative. Here, the pathological and immunohistochemical results confirmed the diagnosis of NF1; positive immunohistochemistry

for CD34, S100 protein, and Ki67, EMA negative, and positive neurofilaments in the intervening axons.

Surgical approach and risks

There are multiple surgical options for dealing with NF1 lesions depending on their spinal location and size. George and Lot proposed a classification system for extramedullary foramen magnum tumors.^[7] The anterior lesions were bilateral or unilateral; the lateral lesions were located between midline and dentate ligament and the posterior lesions behind dentate ligament.^[7] Here, one of the biggest challenges with cervical tumors is the relationship with the vertebral artery (VA) that can be displaced and compressed by the tumor. Therefore, preoperative angiography may be warranted. George and Lot noted the lateral approach (anterolateral and posterolateral) provided the best results versus a standard posterior approach due to the better clinical results and complete lesion resection. The standard posterior approach is most effective when there the tumor is not adherent to the cord.

Purely extradural tumors anterior to the VA necessitate an anterolateral approach, while most cervical dumbbell warrants a posterolateral approach.

Here, we chose to perform a lateral suboccipital craniectomy, which facilitated identification and dissection away from the VA. The posterior C1 arch was removed, and the C2 laminectomy was performed to achieve complete removal of the C2–C3 lesion. This was followed by a right-sided C4–C5 hemilaminectomy for partial resection of the second C4–C5 lesion (e.g., only partial facet removal to preserve stability).

CONCLUSIONS

In a patient with NF1 who presented with dysphagia/dysphonia and tetraparesis, we removed a giant C2–C3 plexiform/dumbbell neurofibroma extending into the foramen magnum/prebulbar cistern along with a secondary right-sided C5 lesion.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understanding that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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