Case Report

ANNALS OF MEDICINE & SURGERY

OPEN

Rare case of multiple neurofibromas of the scalp and trunk in association with intradural extramedullary spinal tumor: a case report

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Introduction and importance: Neurofibromas are tumors of neural connective tissue composed of Schwann cells and fibroblasts. They can occur anywhere in the body, primarily as a solitary mass or as a component of neurofibromatosis. Only 1/4 of the intradural extramedullary spinal tumors are neurofibromas. While most of the neurofibromas are asymptomatic, the authors report a patient with symptomatic multiple neurofibromas of the scalp and trunk that lacked the salient features of neurofibromatosis-1.

Case presentation: A 63-year-old male from Saptari, Nepal, with multiple insidious swellings in the scalp and trunk region, presented with complaints of weakness of the bilateral lower limb for 6–7 years resulting in difficulties in walking and bleeding from the most significant swelling that was in the occipital area.

Clinical findings and investigations: Masses were present all over the scalp, averaging about 4 × 4 cm, with active bleeding from the two swellings. The authors performed the neurological assessment and histopathological and radiological investigations.

Interventions and outcomes: A confirmed diagnosis of neurofibroma was made with myelography revealing an intradural extramedullary spinal tumor. A laminectomy with total excision of the tumor was performed.

Relevance and impact: Meningiomas and nerve sheath tumors (schwannomas and neurofibromas) can both develop in the intradural extramedullary spinal compartment. Contrast-enhanced MRI is the most sensitive and specific imaging modality to evaluate possible spinal column lesions. Surgical excision, partial or complete, is the hallmark treatment of neurofibroma.

Keywords: case report, intradural extramedullary, laminectomy, nerve sheath tumor (NST), neurofibroma, scalp, trunk

Introduction

Neurofibromas are tumors composed of a mix of Schwann cells, perineurial-like cells, and fibroblasts interspersed with nerve fibers, wire-like strands of collagen, and myxoid matrix[1,2]. Most are solitary (up to 90 percent) and not associated with neurofibromatosis type 1 (NF1). Cutaneous, or dermal, neurofibromas are small, nodular skin and subcutaneous tissue tumors arising from small cutaneous nerves, which may cause local pain or bleeding but do not cause neurologic deficits. In NF1, they are by far the most common form of tumor. Multiple neurofibromas are nearly diagnostic of NF1.

HIGHLIGHTS

- Most of the spinal neurofibromas are asymptomatic, but 2 percent of patients with neurofibromatosis type 1 have neurologic symptoms from spinal lesion.
- Meningiomas and nerve sheath tumors (schwannomas and neurofibromas) can both develop in the intradural extramedullary spinal compartment.
- While most of the neurofibromas are asymptomatic, we report a patient with symptomatic multiple neurofibromas of the scalp and trunk that lacked salient features of neurofibromatosis -1.
- Nerve sheath tumors constitute ~25 percent of tumors arising in the intradural extramedullary space. Approximately 65 percent of intradural NSTs are schwannomas, and most of the remainder are neurofibromas.

Nerve sheath tumors (NSTs) constitute ~25 percent of tumors arising in the intradural extramedullary space[3,4]. Approximately 65 percent of intradural NSTs are schwannomas, and most of the remainder are neurofibromas. Most of the spinal neurofibromas are asymptomatic, but 2 percent of patients with NF1 have neurologic symptoms from spinal lesion[5]. Thus, neurofibroma, not associated with NF1 but having extension to intradural extramedullary space is not much described in the literature, and this fact makes this case reportable. This case report is reported according to the Surgical CAse REport (SCARE) guideline.[6]

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Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article

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Annals of Medicine & Surgery (2023) 85:3054–3057

Received 4 March 2023; Accepted 1 May 2023

Published online 10 May 2023

http://dx.doi.org/10.1097/MS9.0000000000000807

Case presentation

A 63-year-old male from Saptari, Nepal, with multiple insidious swellings involving scalp and trunk region, presented to our out-patient department with complaints of weakness of the bilateral lower limb for 6–7 years and bleeding from the most significant swelling – one in the occipital area. His symptoms included weakness of the bilateral lower limbs with paresthesia and difficulty walking smoothly (due to the weakness). The weakness had progressed over 6–7 years but had no other deficit in neurological status. The swellings were gradually progressive, painless, and firm, with an ulcerated summit with bleeding from three of the multiple swellings. There was no history of scalp trauma or falls from height. The patient did not complain of any visual defect or significant weight loss. There was a positive history of a similar swelling on his grandmother's trunk region; however, the first-degree relatives had no such history.

His scalp examination revealed multiple soft to firm, smooth, mobile, noncompressible, nonreducible, nontender, and nontransilluminate nodules with no impulse on coughing. The nodules had ill-defined margins. They were all over the scalp, each averaging about 4 × 4 cm, with the largest one in the occipital area of 6 × 7 cm in dimensions (Fig. 1). The skin over the nodules had bluish discoloration with active bleeding from the two of them. There were no changes in the bone around the margins of the nodules and no diffuse café au lait spots.

Neurological examination revealed decreased sensation below the T5 level, and muscle power assessment showed muscle power of 4/5 in the bilateral lower limb on the MRC scale.

Histopathological examination of the resected specimen confirmed the diagnosis of neurofibroma. Genetic studies could not be done due to unavailability in the institute. An MRI of the spine revealed T11–T12 neurofibroma. (Fig. 2).

Surgery of the scalp neurofibroma had to be done due to profuse bleeding from the ulceration site in the occipital region. Total tumor excision with full-thickness skin graft cover was done. While for the IDEM tumor, the patient was operated on using a posterior approach; laminectomy of T4–T7 vertebrae with total excision of tumor from the intraspinal region and posterior mediastinum was performed. (Fig. 3). The patient followed up regularly in 1 and 3 months, which showed good recovery in terms of patient outcomes and no evidences of recurrence were observed.

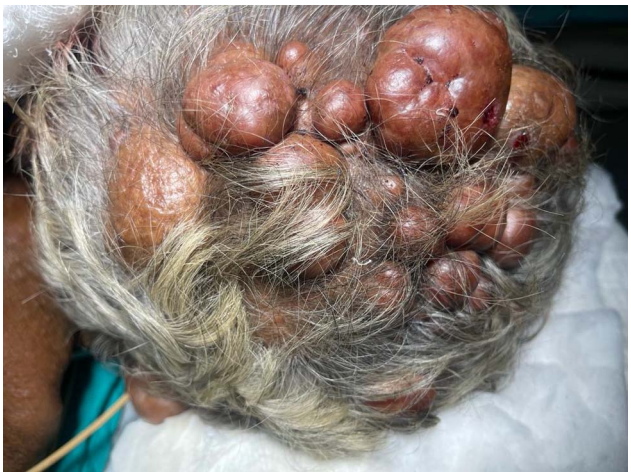

Figure 1. Masses present all over the scalp, each averaging about 4 × 4 cm, with the largest one located in the occipital area of 6 × 7 cm in dimensions.

Discussions

Neurofibromas may occur singly in genetically ordinary people at any point along the peripheral nervous system. Multiple neurofibromas are nearly pathognomonic for NF1. Cutaneous neurofibromas are benign and do not carry a risk of malignant transformation. Still, they often represent a major cosmetic problem in adults but rarely result in neurological dysfunction. Conversely, deep-seated neurofibromas on peripheral nerves and spinal roots frequently lead to neurological disability. Inevitably, adult patients with NF1 have other stigmata of the disorder, with the most common being café au lait spots, skin fold freckling, and Lisch nodules[7,8]. Cases with multiple neurofibromas raise clinical suspicion of the presence of one of the neurofibromatoses.

In our patient, the absence of café-au-lait macules, freckling in the axillary or inguinal region, optic pathway glioma, lisch nodules, and other NF1 stigmata points toward multiple, pathologically proven neurofibromas that do not have other diagnostic features of NF1 or neurofibromatosis type 2.

Ultrasound, CT, and MRI can be helpful in the noninvasive diagnosis and characterization of NSTs. MRI is the most useful imaging modality to characterize tumor extent and suggest neurogenic origin due to its high contrast resolution and multiplanar capabilities[9].

Surgical excision is the hallmark of the treatment of neurofibroma. Our patient had total excision of the scalp mass with complete wound healing; Because of possible recurrence, yearly follow-up is recommended.

Meningiomas and NSTs can both develop in the intradural extramedullary spinal compartment. Intradural, extramedullary NSTs may be either sporadic or associated with one of three inherited disorders: NF1, neurofibromatosis type 2, and schwannomatosis.

Sporadic NSTs are most common in the fifth to seventh decades and have a similar incidence in males and females[10]. Spinal neurofibromas are most common in the cervical region, with higher recurrence rates and lower rates of gross total resection compared with other tumor types[11].

The specific symptoms of NSTs depend upon the spinal level involved. They usually arise from the dorsal sensory roots and may present with radicular sensory changes. Radiculopathy with motor manifestations is not common, even with the involvement of functional roots in the cervical or lumbar spine[12].

Contrast-enhanced MRI is the most sensitive and specific imaging modality to evaluate possible spinal column lesions. MRI can define the anatomic interface between the tumor and the spinal cord but cannot reliably establish a histopathologic diagnosis[13].

Surgery is the primary treatment modality for large or symptomatic intradural NSTs. The complete surgical resection requires the sacrifice of a nerve root or nerve root fascicles. Although these are typically sensory and their loss is well tolerated, sensory deficits should be anticipated and discussed with the patient preoperatively.

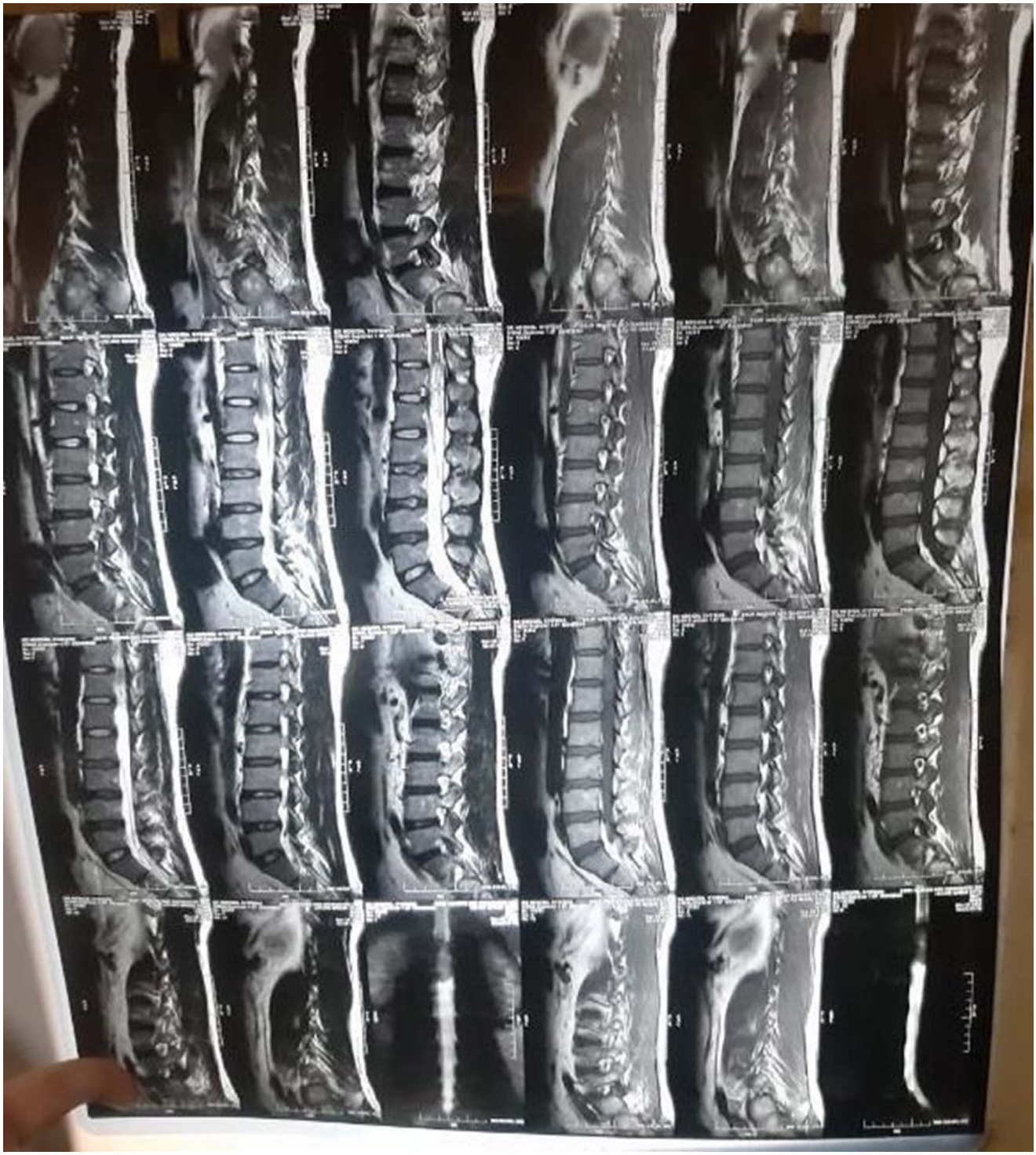

Figure 2. MRI of SPINE revealing T11–T12 neurofibroma.

Resection of intradural NSTs generally uses a posterior or posterolateral approach[14]. Rarely, an anterior approach may be required for midline ventrally located tumors[15]. Minimally invasive approaches that utilize tubular or expandable retractors for transmuscular access have also been described[16]. Intraoperative radiographs are taken to identify the appropriate levels. Following laminectomy, ultrasound may be used over the dura to confirm the location of the tumor. A spinal fluid leak is a rare complication.

Ethical approval

Not required.

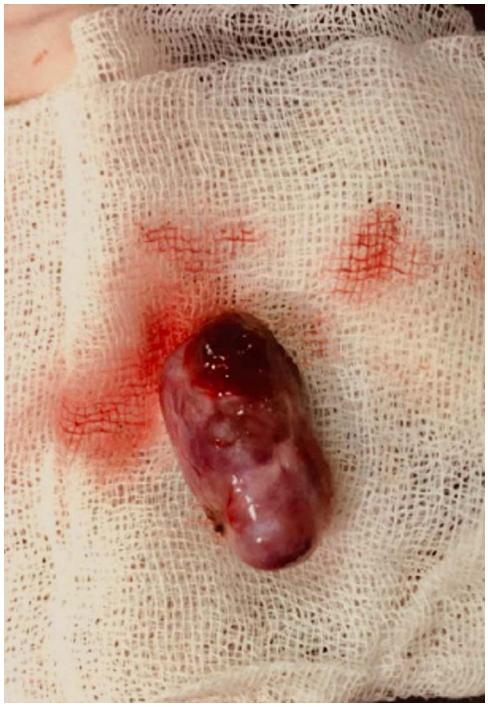

Figure 3. Excised tumor by total excision from intraspinal region.

Consent for publication

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Sources of funding

None.

Author contributions

A.D., A.K.S., N.T.: study concept, data collection, and surgical therapy for the patient; AKS, NT: writing-original draft preparation; A.D., A.K.S., N.T.: editing and writing; A.D., A.K.S., N.T. = senior author and manuscript reviewer. All the authors read and approved the final manuscript.

Conflicts of interest disclosure

None.

Research Registration Unique Identifying Number (UIN)

Not applicable.

Guarantor

Alok Dahal.

Provenance and peer review

Not commissioned, externally peer-reviewed.

Acknowledgments

None.

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