

A Rare Case of Spinal Schwannomatosis presenting as Conus-cauda Syndrome

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ABSTRACT

Multiple extra cranial schwannomas are generally associated with neurofibromatosis (NF) syndromes. Presence of multicentric schwannomas in absence of NF2 is a very rare entity. Schwannomatosis is defined as an extremely rare tumor syndrome characterized by multiple schwannomas with no associated evidence of NF1 or NF2 syndromes. Numerous genetic and molecular analyses done for these tumors have established them as distinct clinical syndrome. Very few cases of schwannomatosis have been reported so far in English literature. The authors herein present a case of spinal schwannomatosis presenting as conus-cauda syndrome.

Keywords: Conus-cauda syndrome, Multiple schwannoma, Neurofibromatosis, Schwannomatosis.

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INTRODUCTION

Schwannomas are slow-growing encapsulated benign peripheral nerve tumors. They represent about one-third of all benign primary spinal tumors.¹ Schwannomas tend to occur as solitary lesions; however, multiple localization involving one or more nerves are known.^{2,3} The presence of multiple schwannomas in a patient is suggestive of possible association with neurofibromatosis (NF) syndromes. National Institute of Health (NIH)⁴ has established guidelines for diagnosis of NF1 and NF2 syndromes. NF1 is the commoner form, with an incidence of 1/2,500 births. The gene for NF1 maps to the chromosome band 17q11.1. NF1 is characterized by presence of multiple café au lait spots, axillary/inguinal freckling, multiple subcutaneous nodules, and ocular manifestations.

NF2 with an incidence of 1/33,000 births is much rarer and is associated with significant morbidity and mortality

owing to the frequent localization of the lesions in the brain and in the spinal cord. The mutation in the NF2 gene maps to chromosome band 22q12. The most important finding for the diagnosis of NF2 is the presence of bilateral cerebellopontine angle vestibular schwannomas with positive family history. On review of literature, we have come across case reports of patients with multiple schwannomas without any associated diagnostic features of NF2. The genetic and molecular analysis of these tumors of these patients revealed no mutations of the NF gene, suggesting schwannomatosis to be a distinct clinical and genetic syndrome.^{5,6} Schwannomatosis is defined as an extremely rare tumors syndrome characterized by the presence of multiple schwannomas in the absence of typical signs of NF1 and NF2. We have reported a rare case of spinal schwannomatosis presenting as conus-cauda syndrome.

CASE REPORT

A 60-year-old female presented to department of neurosurgery with complains of low back pain with gradually progressive asymmetric weakness of bilateral lower limbs for the past 1 year associated with complain of urgency, frequency, and urinary incontinence for past 1 month. The patient has been bedridden for the past 2 months and is dependent on others for her activities of daily living. Neurological examination revealed bilateral foot drop with power of 1/5 at hip and knee in right lower limb and a power of 2/5 at hip and knee in left lower limb as per medical research council (MRC) scale. The patient had decreased sensation to 50% of normal, for all modalities (touch/pain/temperature) below L1 dermatome. The muscle tone was increased over bilateral lower limbs with bilaterally absent ankle jerk reflexes and muted plantar reflexes. Sphincter tone was normal. The past medical history was not suggestive of any auditory or visual disturbances. Family history was not suggestive of NF. On general physical examination, there was no evidence of café au lait spots, subcutaneous nodules, or axillary freckling. The ocular fundus examination and the pure tone audiometry (PTA) were normal. An magnetic resonance imaging (MRI) brain scan was done which was normal (Fig. 1). Dorsal lumbar MRI imaging was done which revealed three well-defined smoothly marginated lesions extending from D10 segment till L2-L3 level with significant compression at

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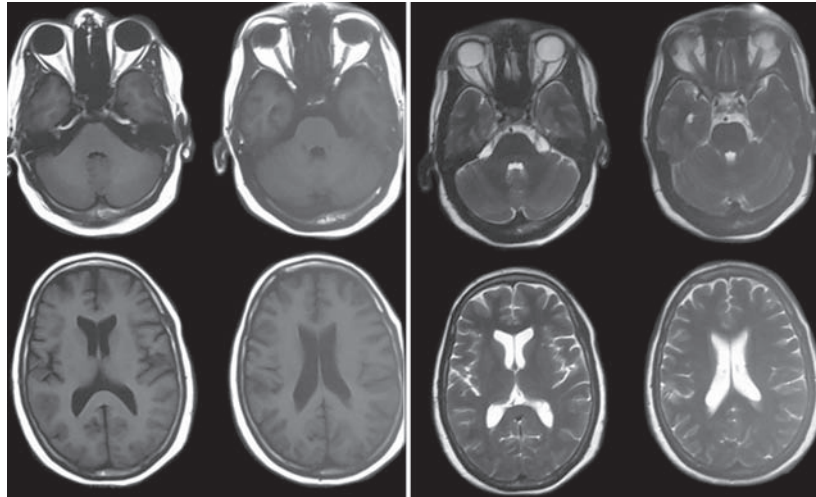
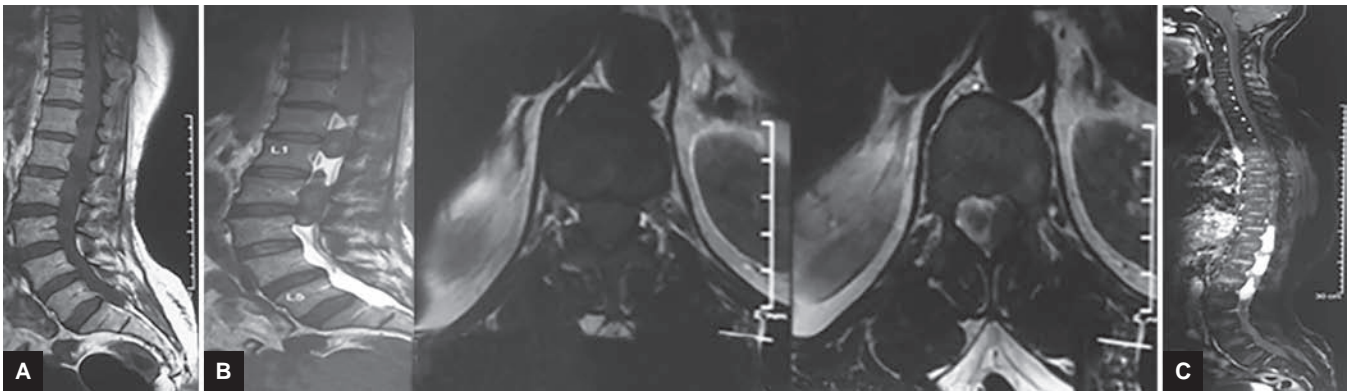


Fig. 1: Magnetic resonance imaging (MRI) brain T1W and T2W images with no evidence of intracranial tumors



Figs 2A to C: (A) Magnetic resonance imaging (MRI) Lumbo sacral spine (LS) T1W image with presence of three very slightly hyperintense well-defined margined intradural lesions at D10-D12, D12-L1, and L2-L3 vertebral body levels; (B) magnetic resonance imaging (MRI) LS spine T2W image with hypointense appearance of the above; and (C) MRI LS spine post contrast images showing significant homogenous post contrast enhancement of the above mentioned lesions

conus medullaris and displacement of the cauda equina nerve roots. Lesions were mildly hyperintense in T1W images (Fig. 2A), hypointense on T2W images (Fig. 2B) and had significant postcontrast enhancement with no obvious extraspinal extension (Fig. 2C). The patient underwent a D10-L3 segment laminectomy with gross total excision of the intradural extramedullary tumors. Tumors were compressing the conus and were densely adherent with the cauda equina roots (Fig. 3). There were two distinct tumors seen at the D10-D12 level and three distinct tumors at D12-L1 and L2-L3 levels (Fig. 4). Pathological examination of the tumors were suggestive of schwannomas (Fig. 5). The patient underwent a postoperative MRI scan after 1 week, which revealed total excision of previously visualized multiple tumors with some localized cerebrospinal fluid collection which was duly managed with ultrasound-guided percutaneous aspiration (Fig. 6). The patient underwent regular physiotherapy in the postoperative period. At 3 months postoperatively the patient has a gradual improvement in the motor power in bilateral lower limbs to 4/5 at hip and

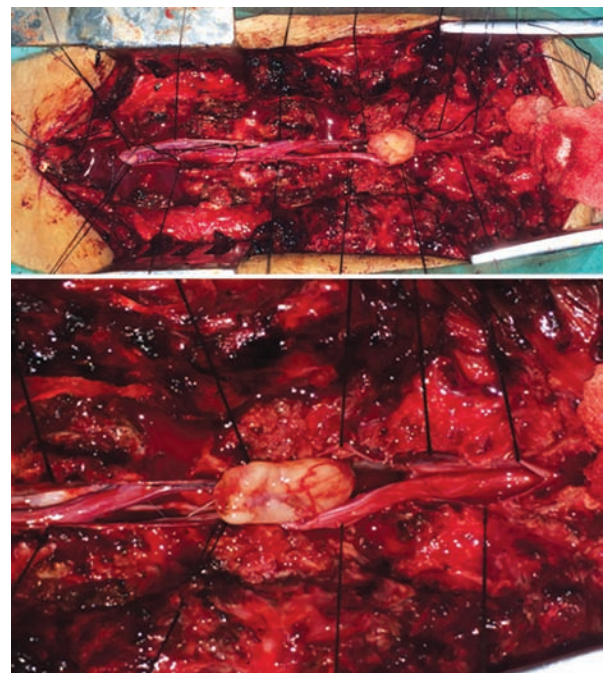


Fig. 3: Intraoperative images of the conus and cauda equina lesions following laminectomy and durotomy, tumors being delivered from the cauda equina roots

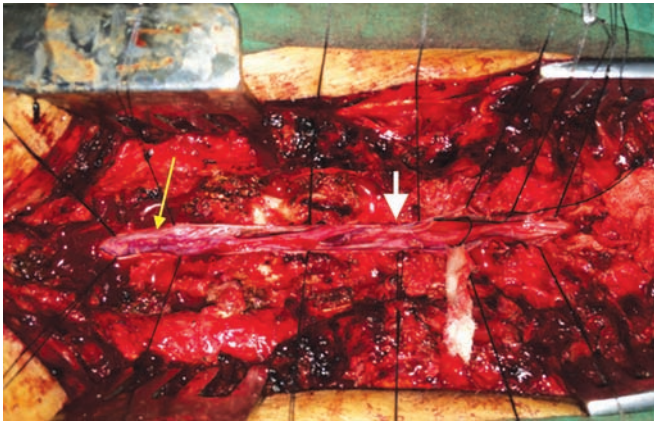


Fig. 4: The conus and cauda equina roots visualized after resection of all the tumors; yellow arrow showing the tortuous veins over conus and the white arrowhead at cauda equina roots



Fig. 5: Resected schwannoma specimens with scale for size

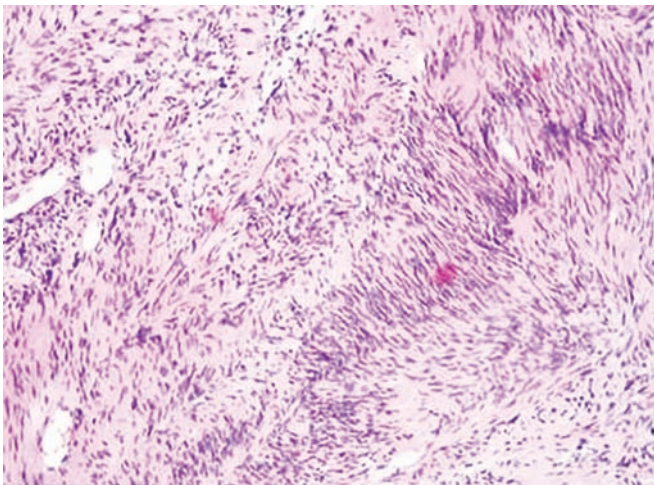


Fig. 6: Histopathology image at low power showing crowded spindle cells with palisading nuclei (Antoni A pattern) and loosely arranged spindle cells in abundant myxomatous areas (Antoni B pattern)

knee and 2/5 at level of ankle. The patient also reports an improvement in sensation to 80% of normal. However, there is no improvement in bladder symptoms, and she was taught clean intermittent self-catheterization.

DISCUSSION

Schwannomatosis is a syndrome characterized by multiple schwannomas in the absence of bilateral vestibular schwannomas, with no diagnostic characteristic of NF1 or NF2. Schwannomatosis has no constitutional mutations of NF1 (17q11.1) or NF2 (22q12) specific genes.⁷ Schwannomatosis was first described in 1973 as NF type 3.⁸ The characteristic of this syndrome was the presence of multiple skin and spinal's schwannomas in the absence of vestibular involvement or other relevant features of NF1 and NF2. Few cases of patients with multiple schwannomas without vestibular lesions have been reported in

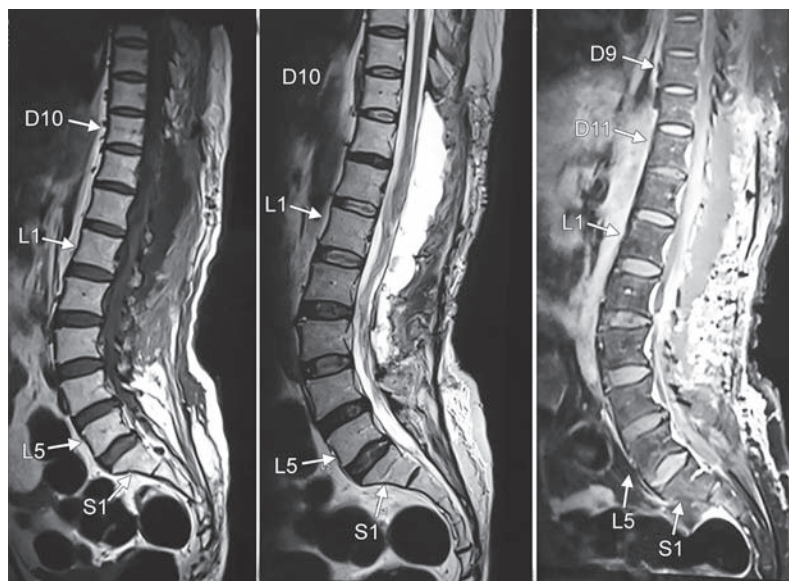


Fig. 7: Postoperative MRI LS spine images T1W, T2W and postcontrast images showing complete excision of the tumors with area of cerebrospinal intensity fluid collection

literature, suggesting the existence of a distinct syndrome from the NF. Daras et al² first reported multiple spinal intradural schwannomas in the absence of von Recklinghausen's disease. A subsequent study, conducted by Altinors et al,⁹ reported a case with three schwannomas at the spinal region, and one intracranial schwannoma without evidence of NF, and defined them as "craniospinal schwannomatosis" to emphasize that the schwannomas were not solely confined to the spine. Kayaoglu et al¹⁰ were the first to report multiple spinal intradural schwannomas in the absence of von Recklinghausen's disease at the cauda equina region; however, the lesions were small and did not cause severe compression of the cauda equina nerve roots unlike the case presented here.

There is no NIH diagnostic criteria for schwannomatosis at present. Jacoby et al⁵ have proposed that presence of two or more schwannomas, in the absence of radiological evidence of vestibular lesions in patients older than 18 years, is indicative of schwannomatosis.

Michael et al¹¹ proposed modifications thus increasing the specificity of diagnosis of schwannomatosis. The important criteria for schwannomatosis are age more than 30 years with multiple schwannomas, no diagnostic criteria of NF2, a normal MRI brain scan, no family history of NF2, and no known constitutional NF2 mutations. Another important criterion is the presence of first degree relative with a confirmed diagnosis of schwannomatosis with one or more pathologically confirmed nonvestibular schwannomas. In case of presence of first-degree relative, there is no limit of age for the diagnosis. MacCollin et al¹² have analyzed alterations in the NF2 locus in patients with schwannomatosis, revealing a typical truncating mutations at NF2 gene, with loss of heterozygosity of chromosome 22. The authors recommend that the symptomatic lesions should undergo gross total excision of tumor as soon as possible with preservation of the roots. The use of intraoperative neurophysiological monitoring is useful in preventing neurological damage. Asymptomatic lesions may be followed at regular intervals with serial MRI scans for development of additional tumors.

CONCLUSION

Schwannomatosis is a distinct syndrome from NF. The presence of multiple schwannomas should entail

a complete imaging of the craniospinal axis owing to multicentric nature of pathology. Symptomatic lesions should be taken up for surgery as early as possible with preservation of the roots. Serial MRIs are mandatory to detect additional tumors keeping with the metachronous nature of these tumors.

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