Radiology Case Reports 19 (2024) 3710–3714

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Low back pain with axillary mass in a perimenopausal woman: A case of schwannomatosis mimicking metastasis ☆

Abhikanta Khatiwada[a,*], *Sharada K C*[b], *Aashish Bastakoti*[c], *Bibek Koirala*[a], *Aalok Kumar Yadav*[a]

[a] *Department of Radiology, Tribhuvan University Teaching Hospital, Nepal*
[b] *Department of Internal Medicine, NAIHS, Nepal*
[c] *Department of Surgery, Tribhuvan University Teaching Hospital, Nepal*

ARTICLE INFO

Article history:
Received 10 May 2024
Revised 19 May 2024
Accepted 20 May 2024

Keywords:
Neurofibromatosis
Neurofibromatosis type 2
Nerve sheath tumor
Schwannoma
Intradural extramedullary lesions

ABSTRACT

Schwannomatosis is a rare neurocutaneous syndrome characterized by the presence of multiple schwannomas along the peripheral nerves, distinctly excluding the vestibular nerves. It is recognized as the third principal form of neurofibromatosis, alongside neurofibromatosis types 1 and 2. In this report, we discuss the case of a 45-year-old woman who initially sought medical attention for low back pain and swelling in her left axilla. Her magnetic resonance imaging revealed multiple enhancing intradural extramedullary lesions, along with a mass in the right upper thoracic region and another in the left axilla, raising suspicions of metastasis. However, a comprehensive analysis that aligned imaging results with histopathological findings confirmed the diagnosis of schwannomatosis. This case highlights the importance of differentiating between various conditions that can cause multiple intradural extramedullary masses, such as nerve sheath tumors, meningiomas, and metastasis. The presence of multiple schwannomas suggests a diagnosis of either neurofibromatosis type 2 or schwannomatosis, making the distinction between these two conditions critical for appropriate management.

Introduction

Schwannomatosis is a rare syndrome characterized by multiple schwannomas of the peripheral nerves without involvement of the vestibular nerves [1,2]. It has an incidence ranging from 1:40,000 to 1:170,000 [3], and is considered the third major form of neurofibromatosis, alongside neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF2) [3]. The associated genes, SMARCB1 *and* LZTR1, are the tumor

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

* Corresponding author.
E-mail address: avikant12@gmail.com (A. Khatiwada).

https://doi.org/10.1016/j.radcr.2024.05.062

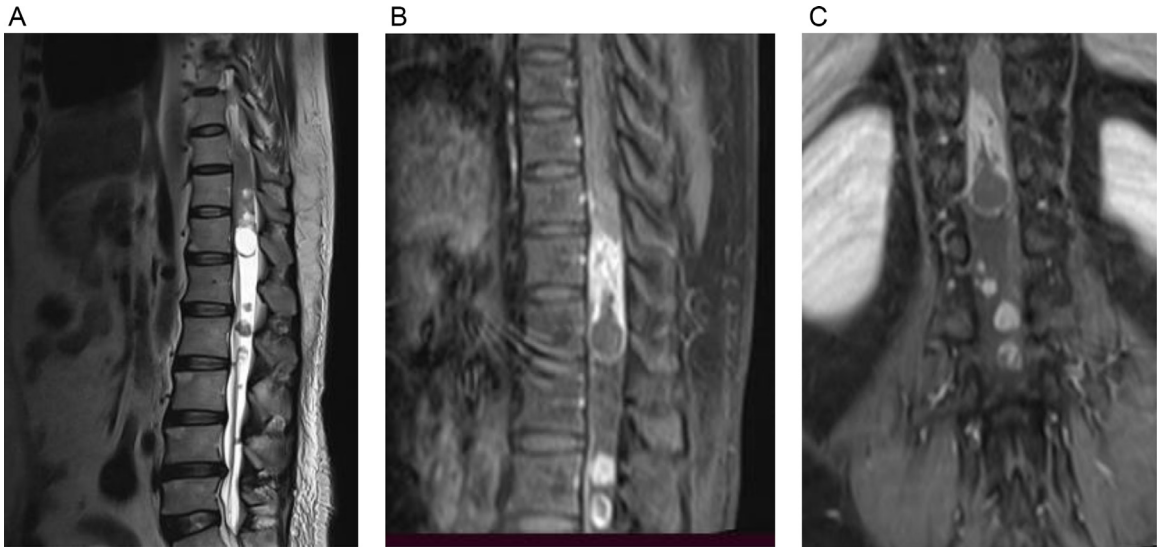

Fig. 1 – (A) T2-weighted sagittal MRI of the thoracolumbar spine showing multiple relatively well-defined heterogenous lesions with intermediate to high signal intensity in intradural extramedullary regions. (B) Post-contrast T1-weighted sagittal MRI of the thoracolumbar spine showing multiple well-defined heterogeneous and intensely enhancing lesions in intradural extramedullary regions. (C) Post-contrast T1-weighted coronal MRI of the thoracolumbar spine showing multiple well-defined heterogeneous and intensely enhancing lesions in intradural extramedullary regions (indicative of multiple schwannomas).

suppressor genes located on the long arm of chromosome 22 [4,5].

Typically, schwannomatosis presents between the ages of 30 to 60 years [6], with pain due to nerve compression [3]. Clinical diagnosis requires one of the following: (a) at least 2 non-intradermal schwannomas, including at least one pathologically confirmed, without evidence of bilateral vestibular schwannomas, or (b) pathologically confirmed schwannoma or intracranial meningioma and an affected first-degree relative [4]. The exclusion criteria include a clinical diagnosis of NF2 or a first-degree relative with NF2 [4].

Here, we present the case of 45-year-old woman who experienced low back pain and swelling in her left axilla. Initial clinical and initial magnetic resonance imaging (MRI) were suspicious for metastasis, but she was eventually diagnosed as schwannomatosis. This case provides valuable insights into differentiating conditions that present with multiple intradural extramedullary lesions and highlights the distinction between schwannomatosis and NF2.

Case report

A 45-year-old woman presented with gradually progressive low back pain and swelling in the left axillary region over the last few months. She had no significant family history. Physical examination revealed firm left swelling in her left axilla. Routine laboratory investigations were unremarkable.

An initial X-ray of the lumbosacral spine revealed little; however, contrast-enhanced MRI of the thoracolumbar spine showed multiple enhancing intradural extramedullary lesions (Fig. 1A-C). Note was made up of partially included mass in right upper thoracic region and mass in the left axilla. Ultrasound of the bilateral breast and axilla revealed a heteroechoic nodular mass in the left axilla, (Fig. 2B) initially raising suspicion of metastasis, prompting further investigation. Neck, chest, abdomen, and pelvic contrast-enhanced computed tomography (CT) scans revealed multiple heterogeneously enhancing soft tissue density masses along the right pleura, left axillary region adjacent to the cervical nerves, and along the bilateral lumbar plexus regions. (Figs. 3A-B) These features were consistent with a nerve sheath tumor. MRI of the brain, including the bilateral cerebellopontine angle (CP) angles, appeared normal. (Figs. 4A and B)

Management included spinal decompression surgery (Fig. 5), after which the patient's symptoms improved significantly. An excisional biopsy was consistent with schwannoma. Imaging findings of multiple non-vestibular schwannomas, were later supported by histopathological reports. (Figs. 6A-C) The patient declined genetic testing due to financial constraints, highlighting challenges in resource limited settings.

Discussion

When encountering multiple intradural extramedullary masses, it is crucial to differentiate between possible nerve sheath tumors, meningiomas, and leptomeningeal metastasis [7]. Meningiomas typically shows homogenous enhancement

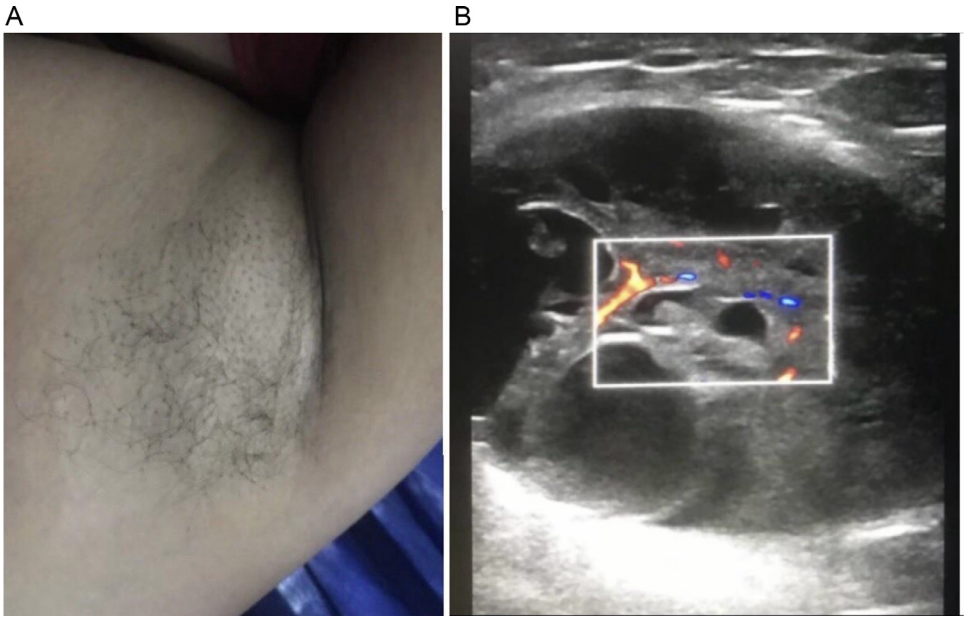

Fig. 2 – (A) Swelling over the left axilla. (B) Ultrasound examination of the left axilla with color Doppler showing well-defined oval, solid-cystic heteroechoic mass with notable internal vascularity within the solid components.

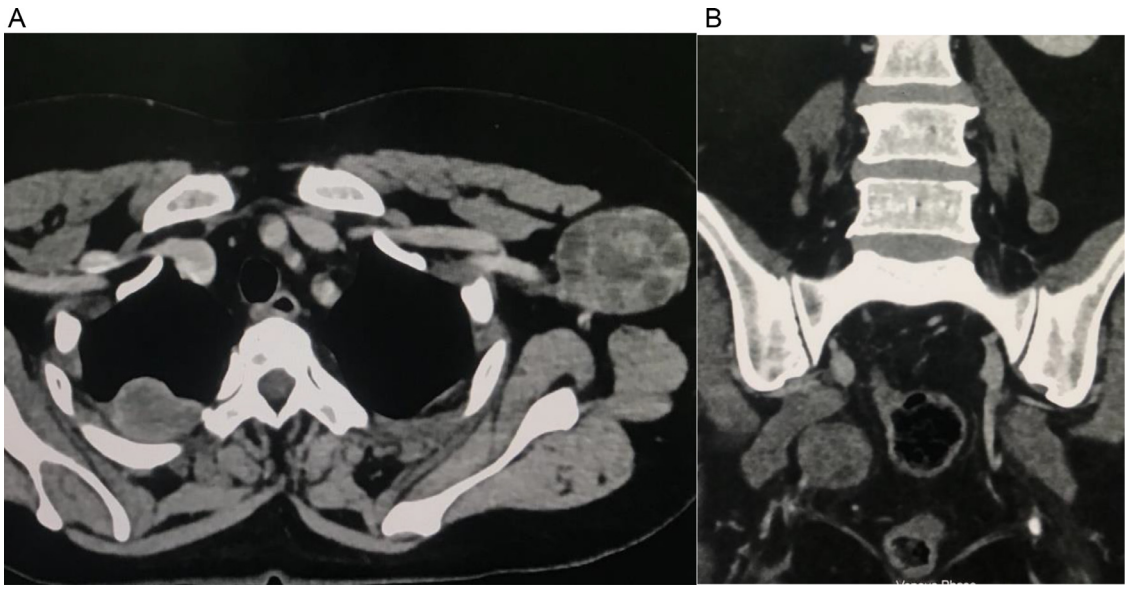

Fig. 3 – (A) Chest contrast-enhanced CT scan axial image showing well-defined heterogeneously enhancing soft tissue density masses along the right pleura and left axillary region adjacent to the cervical nerves. (B) Abdomen and pelvic contrast-enhanced CT scan coronal view showing multiple well-defined heterogeneously enhancing soft tissue density masses along the bilateral lumbar plexus region.

with a dural tail sign [7]. In cases of leptomeningeal metastasis, multifocal enhancing tumor nodules appear at the margins of the spinal cord and/or nerve roots, particularly the cauda equina, with a characteristic sugar coating [8]. The diagnosis is limited to nerve sheath tumors when distribution is along the nerves in both intradural and extradural locations. Extradural neural foraminal extension with a dumbbell appearance is a characteristic feature of nerve sheath tumors [7]. Schwannomas are usually eccentric to the nerves, encapsulated (70%) and multiple schwannomas

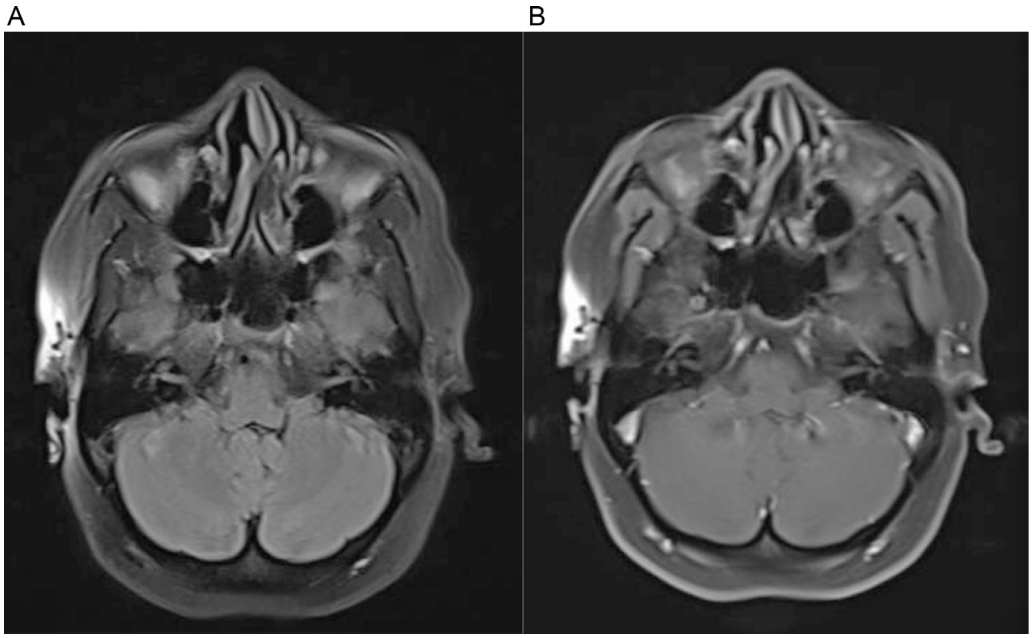

Fig. 4 – (A) MRI of the brain, FLAIR axial image showing normal bilateral CP angle. (B) Post-contrast T1-weighted MRI axial image showing no obvious enhancing lesions in the bilateral CP angle regions.

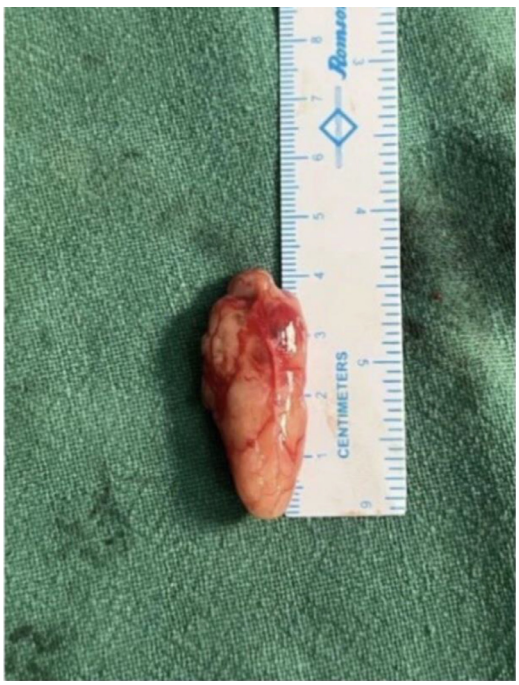

Fig. 5 – (A) Surgically excised specimen of the mass at T11-T12 level, which was responsible for spinal cord compression.

are associated with either NF2 or schwannomatosis [9], whereas neurofibromas are usually centrally located, mostly non-encapsulated (30% capsulated) and associated with NF1 [9].

Differentiating between NF2 and schwannomatosis is essential as these conditions differ significantly in management and prognosis [3]. NF2 patients typically present at a younger age (17 to 24 years), often with neurological deficits and bilateral vestibular schwannomas and are associated with multiple inherited schwannomas, meningiomas and ependymomas [3]. The genetic basis for NF2 involves the NF2 gene (tumor suppressor gene) present on chromosome 22q, which codes for the merlin protein.

MRI is the preferred imaging modality for evaluating nerve sheath tumors [3]. with CT scan and ultrasound also useful depending on the case. Typical imaging findings include multiple, discrete, well-defined lesions along the course of peripheral nerves [1]. The lesions in schwannomatosis are eccentric to the nerve of origin [10]. On T1-weighted MRI, these lesions exhibit low to iso signal intensity, whereas on T2-weighted images, they show high signal intensity [1]. The lesions exhibit heterogenous intense enhancement in post contrast MRI images [1]. In CT scans, these lesions are hypo to isodense (compared to skeletal muscle) and shows variable degree of enhancement [11]. Pathologically, the multiple schwannomas associated with this condition closely resemble sporadic/isolated schwannomas [3].

The mainstay of treatment is conservative management, with surgery usually reserved for cases with symptomatic spinal cord compression [12,13]. Patients with schwannomatosis generally have a normal life expectancy, unlike those with NF2 [1,3].

This encounter emphasizes key radiological aspects across different imaging modalities crucial in differentiating schwannomatosis from other conditions with similar presentations. Correlation with histopathological examinations is invaluable in confirming the diagnosis.

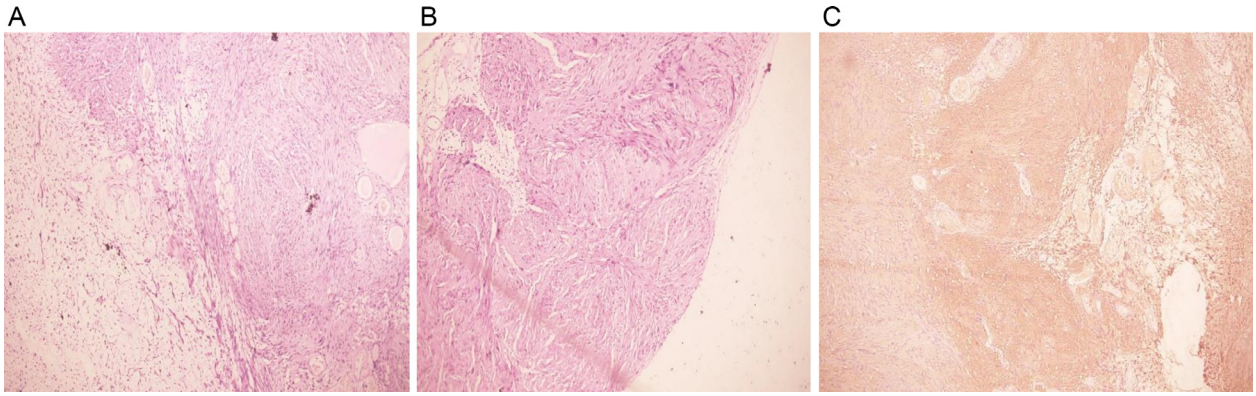

Fig. 6 – (A) Histopathological examination [Haematoxylin and Eosin (H&E) stain, 400 times magnification] of the cut section of the excised specimen revealing hypercellular Antoni A as well as hypocellular Antoni B areas. The tumor cells are spindle-shaped showing mild to moderate pleomorphism. (B) Histopathological examination with H&E stain (200 times magnification) showing Verocay bodies. (C) Immunohistochemistry of the same specimen revealed strong and diffusely positive staining for S-100 (200 times magnification).

Patient consent

Written informed consent was obtained from the patients for publication of this case report and any accompanying images.

Acknowledgement

Dr. Ashutosh Ghimire, Department of Radiology, TUTH. Dr. Reeya Shrestha, Department of Pathology, TUTH.

REFERENCES

[1] MacCollin M, Chiocca EA, Evans DG, Friedman JM, Horvitz R, Jaramillo D, et al. Diagnostic criteria for schwannomatosis. Neurology 2005;64(11):1838–45.
[2] Sestini R, Bacci C, Provenzano A, Genuardi M, Papi L. Evidence of a four-hit mechanism involving SMARCB1 and NF2 in schwannomatosis-associated schwannomas. Hum Mutat 2008;29(2):227–31.
[3] Koontz NA, Wiens AL, Agarwal A, Hingtgen CM, Emerson RE, Mosier KM. Schwannomatosis: the overlooked neurofibromatosis? Am J Roentgenol 2013;200(6):W646–53.
[4] Kehrer-Sawatzki H, Farschtschi S, Mautner VF, Cooper DN. The molecular pathogenesis of schwannomatosis, a paradigm for the co-involvement of multiple tumour suppressor genes in tumorigenesis. Hum Genet 2017;136(2):129–48.
[5] Plotkin SR, Messiaen L, Legius E, Pancza P, Avery RA, Blakeley JO, et al. Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation. Genet Med 2022;24(9):1967–77.
[6] Antinheimo J, Sankila R, Carpén O, Pukkala E, Sainio M, Jääskeläinen J. Population-based analysis of sporadic and type 2 neurofibromatosis-associated meningiomas and schwannomas. Neurology 2000;54(1):71–6.
[7] Koeller KK, Shih RY. Intradural extramedullary spinal neoplasms: radiologic-pathologic correlation. RadioGraphics 2019;39(2):468–90.
[8] Osborn AG. Diagnostic neuroradiology. St. Louis: Mosby; 1994. p. 936.
[9] Beaman FD, Kransdorf MJ, Menke DM. Schwannoma: radiologic-pathologic correlation. RadioGraphics 2004;24(5):1477–81.
[10] Lin J, Martel W. Cross-sectional imaging of peripheral nerve sheath tumors: characteristic signs on CT, MR imaging, and sonography. AJR Am J Roentgenol 2001;176(1):75–82.
[11] Weber AL, Montandon C, Robson CD. Neurogenic tumors of the neck. Radiol Clin North Am 2000;38(5):1077–90.
[12] Westhout FD, Mathews M, Paré LS, Armstrong WB, Tully P, Linskey ME. Recognizing schwannomatosis and distinguishing it from neurofibromatosis type 1 or 2. J Spinal Disord Tech 2007;20(4):329–32.
[13] Mrugala MM, Batchelor TT, Plotkin SR. Peripheral and cranial nerve sheath tumors. Curr Opin Neurol 2005;18(5):604–10.