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Case Report

Multiple schwannomas of cauda equina and peripherals: A case report ☆,☆☆

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ABSTRACT

Spinal schwannomas arising from the Schwann cells of the myelin sheath represent the most common intradural extramedullary tumors. However, occurrences of multiple pearly nerve sheath tumors is rare, and such cases affecting the cauda equina are often localized within the spinal canal of the lumbosacral vertebra on 1 nerve fiber. Herein, we present a case of multiple schwannomas involving distinct nerve fibers of the cauda equina. A 37-year-old female with a history of schwannoma presented with multiple space-occupying lesions in the lumbosacral canal in 2022. Due to a subsequent pregnancy (9 months), further examination and treatment were deferred. Lumbar magnetic resonance imaging performed in February 2024 revealed persistent findings of multiple, bead-like nodular masses within the L1–S1 segments, comparable in size and number to those observed in 2022. Contrast-enhanced MRI demonstrated homogenous enhancement throughout the lesions without evidence of bleeding or cystic components. Given these findings, a diagnosis of schwannoma was suspected. In March 2024, the patient was admitted to hospital for further surgical treatment, the pathological examination result of the resected specimen was consistent with the diagnosis of schwannoma. This case highlights the importance of preoperative magnetic resonance imaging for visualizing tumors, defining their relationship with the nerve roots, and guiding surgical planning. Accurate diagnosis by radiologists plays a vital role in optimal patient management in these cases.

Introduction

Spinal schwannomas arising from the Schwann cells of the myelin sheath represent the most common intradural extramedullary tumors. These tumors typically present as completely enveloped, solitary benign lesions with a low predisposition for malignant transformation. As such, occurrences of multiple pearly nerve sheath tumors, often accompanied by peripheral nerve sheath tumors growing in a segmental

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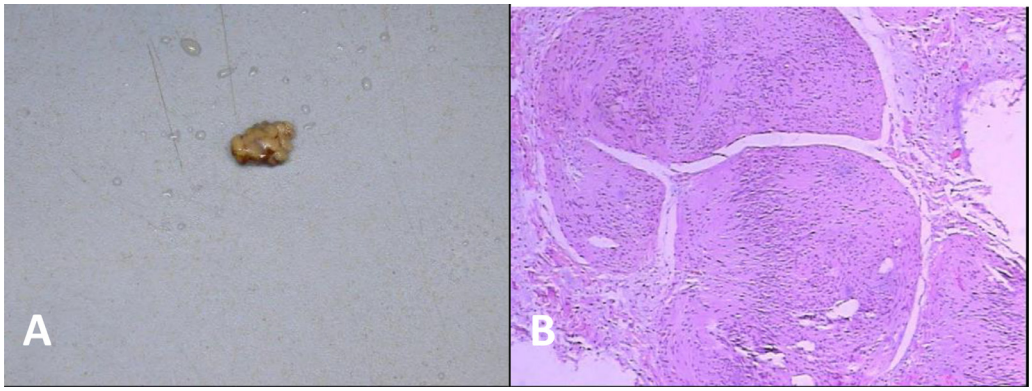

Fig. 1 – Postoperative specimens (A) and pathological images (B) of the right vaginal tumor showed neurinoma.

pattern, is rare. Such cases affecting the cauda equina are often localized within the spinal canal of the lumbosacral vertebra on 1 nerve fiber. Herein, we present a case of multiple schwannomas involving distinct nerve fibers of the cauda equina. Preoperative magnetic resonance imaging proved crucial for visualizing tumors, defining their relationship with the nerve roots, and guiding surgical planning. Accurate diagnosis by radiologists plays a vital role in optimal patient management in these cases.

Case report

A 37-year-old female patient was admitted to our department, after physical examination, for the treatment of multiple space-occupying lesions in the lumbosacral vertebral canal. She reported normal ambulation but with symptoms of chronic constipation. On admission, her neurological examination demonstrated no deficits in bilateral lower limb muscle strength and sensation or sphincter dysfunction. No obvious abnormalities were identified on physical examination. Further history taking revealed a previous history of left perineal subcutaneous tumor resection in 2019, which was confirmed to be a schwannoma (Figs. 1A and B). Currently, there are mobile, palpable, subcutaneous nodules measuring approximately 1 × 1.5 cm in size in the same area, suggestive of postoperative schwannoma recurrence. Her family history was negative for neurofibromatosis.

Abdominal magnetic resonance imaging (MRI) performed in January 2022 confirmed the presence of multiple, well-defined, bead-like nodules of varying sizes within the lumbosacral canal (Fig. 2A). These nodules exhibited homogenous signal intensity without evidence of bleeding, necrosis, or cystic changes. Furthermore, the surrounding structures were unremarkable, and the terminal demarcations were unclear. Due to a subsequent pregnancy (9 months), further examination and treatment were deferred. Lumbar MRI performed in February 2024 revealed persistent findings of multiple, bead-like, nodular masses within the L1–S1 segments (Figs. 2B–E), comparable in size and number to those seen in 2022. Contrast-enhanced MRI demonstrated homogenous enhancement throughout the lesions without evidence of bleeding or cystic components. The margins were well-defined, and the adjacent structures were unremarkable. Normal and contrast-enhanced scans of the head, cervical spine, and thoracic spine showed no definite abnormalities. Given these findings, a diagnosis of schwannoma was suspected. Subsequent genetic testing for NF2, SMARCB1, and LZTR1 was negative.

In March 2024, the patient underwent intramural tumor resection, nerve root decompression, and cerebrospinal fluid leak repair. Multiple schwannomas originating from distinct nerve roots were visualized following left laminectomy of several vertebrae. The 2 largest lesions, approximately 1.5 × 1.0 × 1.0 cm and 1.0 × 1.0 × 0.8 cm, were localized at the L1 and S1–S2 segments, respectively. These tumors adhered tightly to the surrounding nerve roots, significantly compressing the L5–S1 nerve root during surgery. Surgical resection encompassed the entire L1–L2 and L5–S1 segments due to significant mass effect (Fig. 2F). The pathological examination result of the resected specimen was consistent with the diagnosis of schwannoma (Fig. 3).

Postoperatively, the patient displayed a favorable recovery with no deficits in lower limb sensation, pain, urinary dysfunction, or bowel movement changes (e.g., constipation). No evidence of cerebrospinal fluid leakage was detected at the incision site.

Discussion

Schwannomatosis was historically considered a mild variant of NF2. However, genetic studies in 2003 revealed this condition to be a distinct entity, representing the third major form of neurofibromatosis [1]. Unlike NF1 and NF2, the causative gene for schwannomatosis remains undetermined. Although a subset of patients harbor germline mutations in *SMARCB1* and *LZTR1* [2-4], these mutations are not identified in all cases. Specifically, constitutional mutations in the *SMARCB1* gene have only been reported in 40%–50% of familial cases and 8%–10% of sporadic cases.

Diagnostic criteria for polyneurofibromatosis have undergone revisions from previously established guidelines by Jacoby et al. in 1997 [5]. Current criteria define schwannomato-

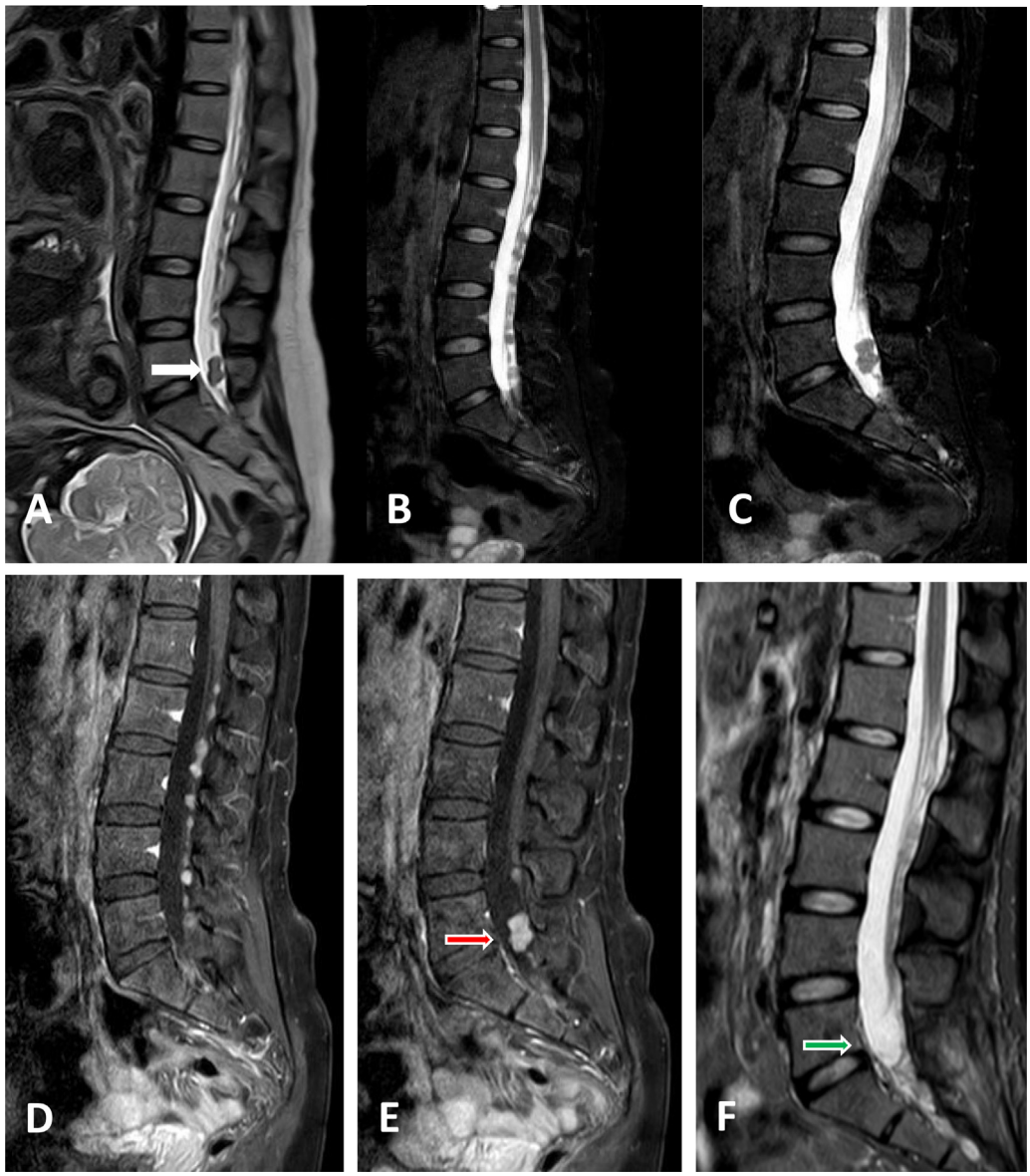

Fig. 2 – Magnetic resonance (MR) images showing of the abdomen and lumbar spine. (A) Abdominal MR images (January 2022). Multiple lesions are observed within the spinal canal, with the 2 largest lesions being adjacent to each other (indicated by the white arrow). (B–E) Plain and contrast-enhanced MR images of the lumbar spine (January 2024). Multiple nodules are observed within the spinal canal, with significant enhancement of the lesions in the spinal canal on contrast imaging (indicated by the red arrow). (F) A subsequent MR imaging scan, performed on July 9, 2024, showing that the 2 largest lesions have been surgically removed (green arrow denotes the excised area).

sis as the presence of multiple schwannomas without fulfilling the diagnostic criteria for NF2 and lacking a somatic *NF2* gene mutation. In previous studies, individuals with 2 or more pathologically confirmed nondermal schwannomas and no evidence of vestibular neuroma on MRI after 30 years of age have been diagnosed as cases of schwannomatosis [6,7]. Following these criteria, the present case fulfills the diagnosis of sporadic schwannomatosis without mutations in the *SMARCB1* or *LZTR1* genes.

Multiple schwannomas in the present case involved the conus and cauda equina. This is consistent with previous reports indicating a higher prevalence of spinal schwannomas in the lower thoracic and lumbar regions [8]. Owing to the large space of the lumbar canal, these tumors are often asymptomatic at an early stage, leading to delayed diagnosis [9,10]. Moreover, plain radiography and computed tomography are usually ineffective at detecting schwannomas, except in cases with aggressive growth and spinal invasion. Physical examination in this case revealed relatively small lesions that did not cause significant symptoms of motor dysfunction or paresthesia. Thus, we strongly advocate for MRI as the preferred initial imaging modality for patients presenting with back and limb pain or sensory disturbances. Typical MRI findings of cauda equina schwannomas include circular,

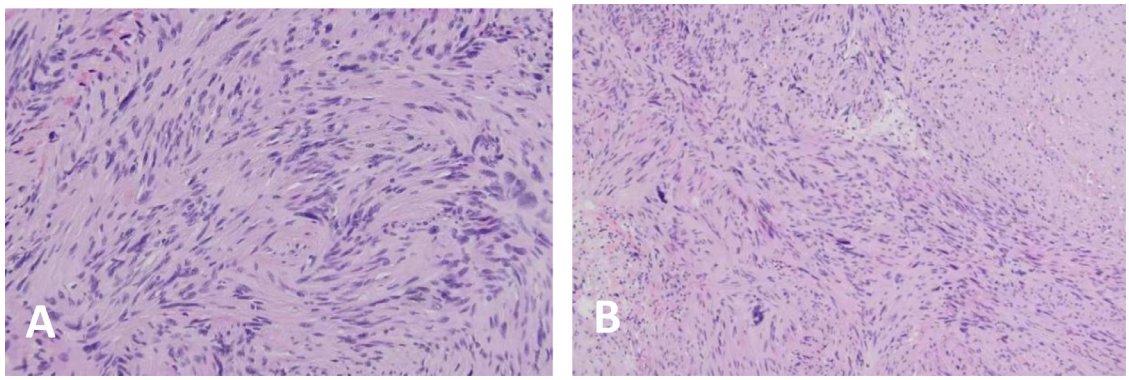

Fig. 3 – Postoperative pathological images of the resected specimen. The results of the pathological examination of the resected specimen were consistent with the diagnosis of schwannoma.

oval, or irregular dumbbell-shaped lesions with well-defined capsules. Contrast-enhanced MRI further delineates the solid component based on significant intensity as compared to the nonenhancing cystic component [10].

The “string of beads” presentation in spinal schwannomas is uncommon. This presentation was observed in the current case, with the largest "bead" located in the L5–S1 segments. To the best of our knowledge, only a few reports have documented this presentation in the cauda equina [11,12]. Although these studies described multiple tumors adhering to a single nerve fiber to form strings of bead-like configuration [11–15], pathological findings our case revealed the involvement of distinct nerve roots. Understanding the potential distribution patterns and MRI characteristics of these tumors can aid clinicians and radiologists in the early recognition and accurate diagnosis of schwannomatosis with multiple lesions.

The majority of multiple schwannomatosis cases are sporadic, with unclear findings with regard to genetic risk. Familial schwannomatosis, accounting for approximately 15%–25% of cases [4], follows an autosomal dominant inheritance pattern, conferring a 50% genetic probability for inheritance. However, incomplete penetrance, as described by Piotrowski et al. [3], suggests that some cases with the predisposing gene mutation may not develop tumors. Genetic testing and close monitoring are therefore recommended for both patients with multiple schwannomatosis and their family members.

Schwannomatosis with multiple nodules typically exhibits static characteristics and is primarily located in the cauda equina. Ozawa et al. [16] reported variable growth patterns in intraspinal schwannomas, including prolonged dormancy, sustained slow growth, initial slow growth, and rapid progression. However, nodules >5 mm in diameter are more likely to develop into larger tumors [17]. Surgical intervention becomes increasingly challenging with tumor growth, and existing symptoms, such as urinary dysfunction, might not show significant improvement postoperatively [18]. Consequently, surgical resection is indicated for symptomatic or rapidly growing tumors, whereas small and asymptomatic tumors can be monitored with annual clinical evaluations [19,20].

Although spinal schwannomas are generally curable with surgical resection, some patients may require multiple surgeries due to the development of new schwannomas. Given that surgical difficulty increases with tumor growth, delayed surgery carries potential risks. Seppälä et al. [21] correlated successful postoperative outcomes in patients with intravertebral schwannomas with preserved preoperative neurological function. Early intervention before significant neurological impairment is therefore crucial to achieve optimal results. Furthermore, delayed surgical treatment can also increase the risk of malignant transformation. However, Merket et al. [22] cautioned against the overdiagnosis of malignancy in multiple schwannomatosis, emphasizing that other malignant tumors may be more likely.

Conclusion

The present case involves the highest number of tumors reported in a single patient with multiple nerve sheath tumors in the cauda equina. It is unique in that the tumors were present on multiple nerve roots. In addition, the case-specific genetic test was negative, ruling out a genetic cause. This case highlights the importance of preoperative MRI for visualizing tumors, defining their relationship with the nerve roots, and guiding surgical planning. Accurate preoperative imaging diagnosis plays a crucial role in determining the optimal surgical timing and approach.

Patient consent

This patient consents to the reporting of images and other clinical information about his or her case in a medical journal.

Ethics approval

The case report was conducted in accordance with the principles of the Declaration of Helsinki and was approved by the

institutional review board of [The Third People's Hospital of Zigong, Sichuan Province, China].

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