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Schwannomatosis. A rare case report

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ABSTRACT

Schwannomatosis is characterized by a predisposition to develop multiple schwannomas and rarely meningiomas. People with schwannomatosis are most commonly present between the second and fourth decades of life. The most common feature is localized or diffuse pain or an asymptomatic mass. Schwannomas most commonly involve peripheral and spinal nerves. We report the case of a woman who was initially referred to our department because of suspected neurofibromatosis type 2.

Keywords
schwannomatosis, spine, schwannoma, neurofibromatosis

INTRODUCTION

Schwannomatosis is distinguished by its inherent tendency to manifest multiple schwannomas in affected individuals [1]. It could be familial or sporadic [2]. Differentiating schwannomatosis from both type 1 and particularly type 2 neurofibromatosis is crucial due to their distinct genetic origins, disease progression, and associated impacts.

CASE REPORT

A 47-year-old female patient with a history of hypothyroidism. She was first operated on for uterine fibroids at age 34. Six years later, she underwent surgery for pleural schwannoma. She came to our emergency department because of back pain for 10 years and radicular pain in both legs. Her clinical examination revealed strabismus and multiple achromatic spots. MRI of the spine showed intra dural extra medullary lesion at L3, iso-intense in T1WI, with contrast enhancement after gadolinium injection and hyper-signal in T2WI (Figure 1). Cranial MRI was normal. The patient underwent surgery for spinal decompression and resection of the tumor. Anatomic pathologic examination revealed tumor composed of Schwann cells, which are seen as streams of elongated cells with tapering nuclei. Cellular organization shows loosely arranged cells with areas of vacuolation and

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lipidisation (Antoni B). Mitoses are sparse and the appearances are of a conventional Schwannoma (WHO grade I). (Figure 2). Postoperative evolution showed no back pain. Genetic counseling was performed, and the karyotype showed no gene mutations. There was no family history of neurofibromatosis. The diagnosis of schwannomatosis was made according to Kehrer-Sawatzki criteria (more than two non-intradermal schwannomas and no evidence of bilateral vestibular schwannomas) One year later, the patient is asymptomatic.

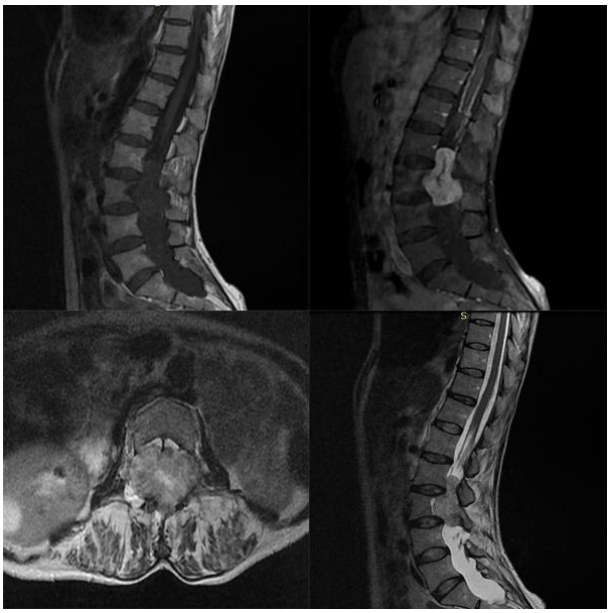

Figure 1. MRI of the spine showed intra dural extra medullary tumor at L3, iso-intense in T1WI, with contrast enhancement after gadolinium injection and hyper-signal in T2WI.

Discussion

The diagnosis of schwannomatosis is determined by either clinical criteria alone or a combination of molecular and clinical criteria [3]. The majority of schwannomatosis cases are considered sporadic, meaning they arise in individuals without a family history of the disorder [2]. While some individuals with sporadic schwannomatosis exhibit mutations in the SMARCB1 or LZTR1 gene, the underlying cause of the condition remains unknown in other cases [4]. Schwannomas mainly affect peripheral nerves (90%) and spinal nerves (75%). Within spinal nerves, the lumbar spine is most commonly involved [5]. Involvement of cranial nerves is rare, but the trigeminal nerve is the most commonly affected cranial nerve in such cases. In symptomatic schwannomas causing pain or neurological deficits, surgical intervention is recommended. The principles of surgical resection of peripheral nerve tumors in such cases are similar to those for removal of sporadic nerve sheath tumors. Although there is a theoretical concern that radiation exposure may increase the risk of malignant transformation, this association has not been demonstrated in individuals with schwannomatosis [6]. The exact role of radiation therapy in this context is not yet clear and requires further investigation.

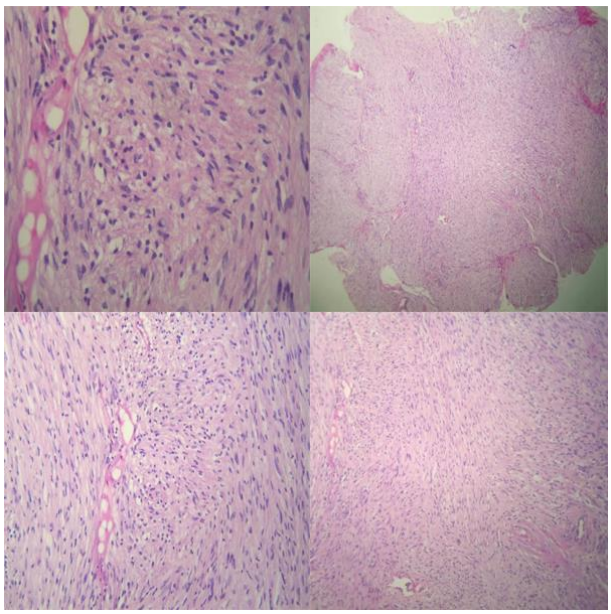

Figure 2. Histological examination reveals a tumor composed of Schwann cells, which are seen as streams of elongated cells with tapering nuclei. Cellular organization shows loosely arranged cells with areas of vacuolation and lipidisation (Antoni B). Mitoses are sparse and the appearances are of a conventional Schwannoma (WHO grade I).

Conclusion

Schwannomatosis can be diagnosed based on clinical or combined molecular and clinical criteria. Surgical intervention is recommended for symptomatic schwannomas causing pain or neurological deficits, following similar principles as sporadic nerve sheath tumors.

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