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Spinal Schwannomatosis: Case Report of a Rare Condition

Spinal Schwannomatosis: Ender Görülen Bir Olgu Sunumu

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ABSTRACT

Schwannomatosis is a rare tumour syndrome characterized by more than one schwannoma without any sign of neurofibromatosis (NF). A 22-year-old male patient was admitted with weakness in his extremities. Neurological examination revealed a tetraparesis syndrome below the C6 level. Magnetic resonance (MR) imaging showed a demarcated mass with strong enhancement at the C4-7 levels. The patient improved rapidly after removal of the tumour. The histological diagnosis was schwannoma. Two years later, the same patient was admitted with the complaint of severe low back and leg pains. The neurological examination was normal except bilateral positive straight leg raising test and dysesthesia at the L3,4 and 5 dermatomes. MR showed a well-circumscribed lesion at L4-5. Laminectomy and tumour excision relieved his complaints. The histological diagnosis was schwannoma. A detailed clinical examination and MR scanning of the central nervous syndrome excluded NF2. In conclusion, although schwannomatosis is a benign condition, symptomatic tumours should be resected surgically.

KEY WORDS: Neurofibromatosis, Schwannoma, Schwannomatosis, Spinal schwannomatosis

ÖZ

Schwannomatosis herhangi bir nörofibromatosis bulgusu olmadan birden fazla schwannoma sahip olmayla karakterize nadir görülen bir sendromdur. 22 yaşındaki erkek hasta kol ve bacaklarında kuvvetsizlik yakınmaları ile başvurdu. Nörolojik muayenesinde C6 altında tetraparezi saptandı. Manyetik rezonans (MR) görüntülemede C4-C7 arasında kuvvetli boyanma gösteren bir lezyon saptandı. Hasta lezyon çıkartılması ameliyatından sonra hızla düzeldi. Histopatolojik tanı schwannom olarak bildirildi. Aynı hasta iki yıl sonra bel ve bacaklarında şiddetli ağrı yakınmaları ile tekrar başvurdu. Muayenede bacak germe testi iki taraflı müspetti ve L3, L4 ve L5 segmentlerinde disestezi mevcuttu. MR'da L4-5 düzeyinde iyi sınırlı bir kezyon görüldü. Laminektomi ve tümör eksizyonunu takiben hastanın yakınmaları geçti. Histopatolojik tanı schwannom olarak bildirildi. Ayrıntılı muayene ve santral sinir sisteminin MR ile tetkiki yapılarak NF2 olasılığı dışlandı. Sonuç olarak schwannomatosiz bening bir durum olmasına rağmen semptomatik tümörler çıkartılmalıdır.

ANAHTAR SÖZCÜKLER: Nörofibromatozis, Schwannoma, Schwannomatozis, Spinal schwannomatozis

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INTRODUCTION

Schwannomas are slowly grooving, encapsulated benign peripheral nerve tumours derived from Schwann cells that surround the nerve axons (3). Schwannomatosis is a rare tumour syndrome that is based on clinical criteria of multiple schwannomas without any sign of neurofibromatosis (NF) (6). Schwannomatosis may be classified as "definite" or "possible" or "segmental" according to proposed diagnostic criteria (7). Definite schwannomatosis includes: having two or more nonintradermal schwannomas, being older than 30 years, having no evidence of vestibular schwannomas and having no known constitutional NF2 mutations. For possible schwannomatosis, the patient must have no symptoms of eighth nerve dysfunction, must be older than 45 years old, must have two or more nonintradermal schwannomas and not have a known constitutional NF mutation. When the patient has schwannomatosis limited to one limb or five or fewer contiguous segments of the spine, it becomes segmental schwannomatosis meeting criteria for either definite or possible schwannomatosis.

NF2 is the main disease in the differential diagnosis of schwannomatosis (5). High-resolution magnetic resonance (MR) imaging of central nervous system, detailed ophthalmologic examination and a constitutional NF2 mutation test is mandatory to exclude NF2 (2). The clinical presentation and tumour histology may be helpful in the diagnosis. In this paper, a case of spinal schwannomatosis in a 22-year-old male is presented.

CASE REPORT

A 22-year-old man was admitted with a one-week history of tetraparesis. Neurological examination showed tetraparesis below C6, T4-6 hypoesthesia, aesthesis below T7, and diminished anal reflexes with urinary and fecal incontinence. Magnetic resonance (MR) imaging disclosed a well-circumscribed, oval shaped mass with contrast enhancement (Figure 1) at C4-7 levels. Laminectomy and excision of the intradural extramedullary tumour was performed. After an unremarkable postoperative period, the patient showed rapid improvement and was discharged on advice of rehabilitation. The histopathology of the tumour was reported as schwannoma (Figure 2). The patient was then lost to follow-up. Approximately 2 years after his operation, he was admitted again with complaints of severe low back and leg pains. His neurological examination revealed bilateral positive straight leg raising test and dysesthesia at the L3,4 and 5 dermatomes. The tetraparesis found at the previous admission had disappeared. MR examination showed a demarcated, spontaneous high intensity at T1-weighted images and heterogeneous low intensity at the T2-weighted lesion (Figure 1). A single level lumbar laminectomy and excision of the tumour was performed. The

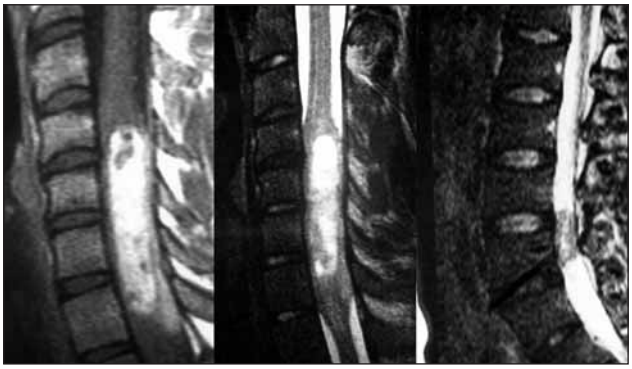

Figure 1: *A well-circumscribed, oval shaped large mass with contrast enhancement at C4-7 levels on sagittal T1-weighted (left) and T2-weighted (middle) MRI images. A small, well-circumscribed, heterogeneous hypo-intense mass at L3-4 levels on sagittal T2-weighted MRI images (right).*

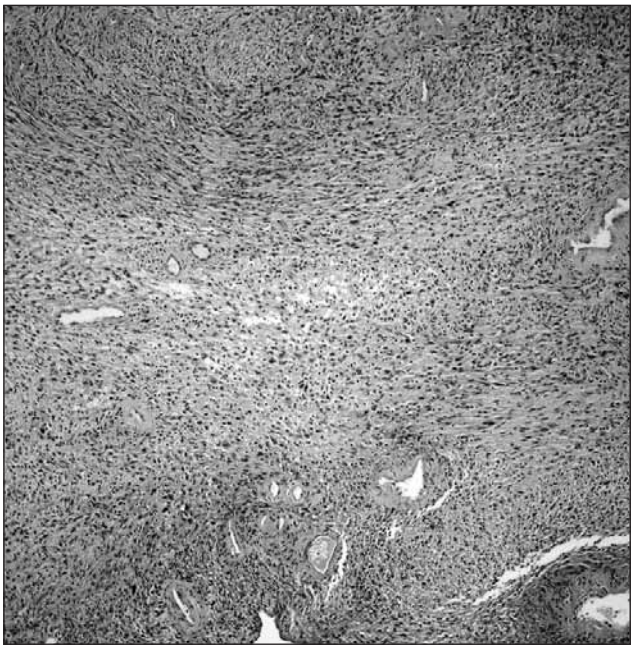

Figure 2: *Schwannoma, showing compact fascicles of elongated tumour cells with nuclear palisading and hyalinized vessels (HEx100).*

postoperative period was unremarkable. The histological examination of the tumour revealed schwannoma. After this diagnosis, a detailed family history and ophthalmologic examination and high-resolution assessment of all the nervous system was performed. No positive findings suggesting NF2 was detected. The patient is now symptom free for a 4-year follow period.

DISCUSSION

Schwannomatosis is a term used for the patients with more than one schwannoma and constitutes a third form of neurofibromatosis (7). There have been some reports that delineate patients having multiple schwannomas without vestibular schwannomas (5,6,8). The presence of multiple schwannomas in a patient implies a number of genetic predispositions including neurofibromatosis (6). Neurofibromatosis is a disease with a group of genetic disorders where the two principal forms, NF1 and NF2, are distinctive (3). NF2 is also known as central neurofibromatosis and its clinic features involve bilateral vestibular schwannoma as well as meningiomas and ependymomas. An overlap in the presentation and phenotype of the disease can be seen in patients with schwannomatosis and NF2 (7), but they are distinct entities. Molecular studies of NF2 patients have revealed an autosomal germline mutation in the NF2 gene at 22q12 (10), but schwannomatosis has a nongermline genetic linkage to chromosome 22 (4).

While the incidence curve of schwannomas plateaus from the third through the sixth decade (3), multiple schwannomatosis generally becomes symptomatic during the third decade and associated with pain and show no sex predilection (1,6,7,9). Patients usually have a single spinal or peripheral schwannoma and only a small part of them have schwannomatosis (7). In a population-based study made in Finland, the annual incidence of schwannomatosis was found to be about 1/1,700,000, which can be accepted to be as common as NF (1).

In most of these patients with schwannomatosis, the tumours are located in a single extremity or a segment of the spine (7). According to the diagnostic criteria defined by MacCollin and et al (7), the presented patient can be accepted as a possible schwannomatosis. He is younger than 30 years old, has two non-intradermal schwannomas, two histological confirmations and no evidence of vestibular tumour on a high quality MR scan. The case is not accepted as segmental, as he would need to have tumours limited to one limb or five or fewer contiguous segments of the spine for segmental schwannomatosis (7,8). He has spinal schwannomas located at C3-7 and L3-4 segments that are further than 5 spinal segments from each other. Pain and progressive motor deficit or sings of spinal cord compression due to a mass effect are the main symptoms (7,8). In the presented case, the leading symptoms were progressive motor deficit and tetraparesis, during the first admission; and low back and leg pain during the second admission.

MRI is the gold standard for detecting these tumours. They are seen as well-defined, rounded or oval lesions. The differential diagnosis of schwannomatosis includes NF2 and the Carney complex (5,7).

Surgery is indicated for the symptomatic lesions in schwannomatosis. Symptomatic schwannomas should be resected (6,9,10). In the presented case, both schwannomas were removed surgically as they became symptomatic. Before the first admission, the patient presented with tetraparesis, and removal of the tumour not only improved his neurological status but also provided a definite histological diagnosis. The second time, the patient suffered from severe low back and leg pain resistant to medical treatment; and again the excision of the tumour provided relief of his symptoms and also provided a histological diagnosis. In the previously reported surgical series, the percentage requiring schwannoma resection was about 2.4 to 5% (1,6,9).

In conclusion, schwannomatosis and NF2 are two different pathologies and accurate diagnosis is important. It is also essential to examine a patient with a schwannoma for a possible diagnosis of schwannomatosis. If the tumours are symptomatic, they should be removed surgically.

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