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RECURRENCE OF SPINAL SCHWANNOMAS IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1: A CASE REPORT

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Recurrence of spinal schwannomas is common in neurofibromatosis type 2

The occurrence of spinal schwannoma in neurofibromatosis type 1 is a rare phenomenon

Further studies is necessary to establish the association between spinal schwannoma and neurofibromatosis type 1

ABSTRACT

Introduction and importance

Spinal schwannomas are benign tumors usually attached to peripheral nerves, consisting of a clonal population of Schwann cells. Neurofibromatosis type 1 is an autosomal dominant neurocutaneous disorder that predominantly affects the skin, bone and nervous system. Neurofibromatosis type 1 is a clinically and genetically distinct from neurofibromatosis type 2. This case report highlights the rare association between spinal schwannoma and neurofibromatosis type 1.

Case presentation

The patient with a past medical history of spinal schwannoma, operated 1 year back, presented with back pain, weakness of lower limbs and urge incontinence. On examination, she had cutaneous features suggestive of neurofibromatosis type 1 and there was impairment of all sensory modalities below hip region. MRI revealed spinal schwannoma at D9 level for which laminectomy with removal of schwannoma was performed.

Clinical discussion

The occurrence and recurrence of spinal schwannoma in neurofibromatosis type 2 is common finding. But such association has not been established between spinal schwannoma and neurofibromatosis type 1. In this case, the recurrence of spinal schwannoma has been linked to neurofibromatosis type 1 in the absence of other well-defined etiologies.

Conclusion

The occurrence of spinal schwannoma can be genetic or sporadic. The recurrence is usually associated with familial tumor syndrome. The available literature has not established association between neurofibromatosis type 1 and spinal schwannoma, thus, emphasizing the need of more focused studies .

Key words: Neurofibromatosis type 1, spinal, schwannoma

Introduction

Schwannomas are benign, well-circumscribed tumors usually attached to peripheral nerves, consisting of a clonal population of Schwann cells.[1] Spinal schwannomas account for about 25% of primary intradural spinal cord tumors in adults.[2] In the vast majority of cases, schwannomas are solitary and sporadic, while multiple schwannomas are associated with neurofibromatosis type 2 (NF2) and schwannomatosis. Neurofibromatosis type 1 (NF1) is an autosomal dominant neurocutaneous disorder that predominantly affects the skin, bone and nervous system. NF1 is distinct on clinical and genetic grounds from neurofibromatosis type 2, a rare disorder characterized by bilateral vestibular schwannomas and other benign nervous system tumors[3]. Gold standard treatment for symptomatic spinal schwannomas is complete surgical resection, which stops symptoms progression, helps recovery in most patients, and decreases the rate of recurrence. Radiotherapy can be considered as second-choice treatment in patients who are not good candidates for surgery, or for recurrent tumors[4]. The recurrence rate for the spinal schwannomas ranges from 4.08 to 9.4% and depends on size, extent of tumor and genetic associations rather than the location of the tumor[5][6]. This case has been reported in line with the SCARE 2023 criteria[7].

Case presentation

A 50 year female presented to OPD with complains of back pain and weakness of lower limbs for 3 months. Pain was insidious in onset, gradually progressing and later radiating to bilateral lower limbs. It was associated with weakness of lower limbs in the form of difficulty in walking and inability to stand for prolonged period of time that had increased to an extent that she had been unable to walk without support. She had urge incontinence as she could not control her bladder and bowel once she had sensation. There is also a past history of being operated 1 year back after she was diagnosed with spinal schwannoma; she had undergone laminectomy with total resection for the spinal schwannoma extending from L5-S1.

On examination, there was skin patch over her left-hand measuring 2cm, axillary freckling along with multiple fibromas over her back; suggestive of NF type 1. Genetic studies were not performed. The diagnostic criteria utilized to make diagnosis is NIH criteria for neurofibromatosis 1[8]. Her higher mental function was intact, cranial nerves were intact. There was no motor or sensory deficit over both upper limbs. Power of right lower and left lower limb were 3/5 and 4/5 respectively. There was sensory impairement of all modalities below hip region (right >>left).

Her recent MRI revealed single right sided intradural extramedullary, homogenously enhancing, well marginated mass at D9 level (Fig 1). Other laboratory parameters were within normal range.

She underwent bilateral laminectomies of D9, following the durotomy. A well defined solid yellowish lesion was visualized adjacent to the D9 vertebrae; it was then completely resected (Fig 2). Neuromonitoring potentials remained unchanged. Tumor was sent for histopathology and was later confirmed as benign spinal schwannoma. The patient recovered post operatively with gradual increase in strength of lower limbs and was discharged on 3rd post-operative day.

Discussion

Recurrence is defined as reappearance after definite surgical excision or symptomatic increase in size of a residual tumor on follow up imaging studies. Local recurrence means reappearance in the same place as the original tumor or very close to it. Regional recurrence means reappearance into lymph nodes or tissues near the original tumor. Distant recurrence means reappearance in organs or tissues far from the original tumor[9].

Spinal schwannoma is solid, encapsulated, mostly benign and intradural- extramedullary tumor. They may occur spontaneously, or in the context of a familial tumor syndrome such as neurofibromatosis type 2 (NF2), schwannomatosis and Carney's complex[1]. However, the occurrence of spinal schwannomas in neurofibromatosis type 1 is not well established and such phenomenon is rare finding in itself. But as in other causes of spinal schwannomas, gross total resection (GTR) with preservation of neurological functions is the best treatment to relieve patients' complaints and to reduce the recurrence rate[6].

Analyzing our case, the patient developed spinal schwannomas at two different locations at different period of time and at both instances, she underwent laminectomy with removal of schwannoma with good postoperative recovery. Recurrence is common in schwannoma if residual tumor is left during excision but at the same site of initial lesion[10]. There should be high degree of suspicion to consider the recurrence of spinal schwannoma, especially if the patient has been previously operated for the same condition. Chordomas, Giant cell tumors, Meningioma and Chondroblastoma are part of the differential diagnoses for the spinal schwannomas and each should be carefully excluded to reach a diagnosis of spinal schwannoma. Laboratory tests combined with MRI have a good sensitivity and specificity for spinal schwannoma. Laminectomy has normally been used as a standard approach for intradural spinal tumors but this procedure is associated with spinal instability and deformity. Laminoplasty was developed to overcome this complications[11]. However, studies have not demonstrated laminoplasty to have any better functional outcome compared to laminectomy in the resection of intradural lesions[12][13].

Since, schwannoma is a slow growing, benign neoplasm, the expected interval between two episodes can be years but the patient developed two different neoplasms within a period of 1 year; the cause of which cannot be linked to other than neurofibromatosis type 1. The neurofibromatosis type 1 is typically associated with occurrence of plexiform neurofibromas and

less commonly central nervous system gliomas[14]. There is no literature describing the recurrence of spinal schwannomas in neurofibromatosis type 1. Thereby, the spinal schwannoma is a rare occurrence in neurofibromatosis type 1 and needs further study to establish a link between these two entities.

Thus, though there is well established association between spinal schwannomas and neurofibromatosis type 2; the diagnosis should be considered in a patient with neurofibromatosis type 1 whenever the patient develops symptoms or the symptoms worsens followed by proper investigation to identify/ rule out the diagnosis.

Conclusion

The occurrence of spinal schwannomas in a patient with neurofibromatosis type 1 is a rare phenomenon and in the absence of well-established association between spinal schwannoma and neurofibromatosis type 1, it becomes difficult for the clinicians to consider the diagnosis especially when there is recurrence following the initial surgery. Thus, further study is needed between neurofibromatosis type 1 and spinal schwannomas to establish an association between them so that it would help the clinicians to consider the diagnosis when the patient presents with such symptoms.

Conflict of interest

No conflict of interest

Source of funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Ethical approval

Ethical approval is not required for case report from ethics committee at our institution.

Consent

Written informed consent was obtained from the patient for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Research Registration

1. Name of the registry: not applicable

2. Unique identifying number of the study: not applicable

3. Hyperlink to your specific registration (must be publicly accessible and will be checked): not applicable

Provence and peer review

Not commissioned, externally peer-reviewed

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Figure 1: MRI revealed intradural extramedullary lesion extending from D9 to D10 level and compressing the cord (marked by arrow).

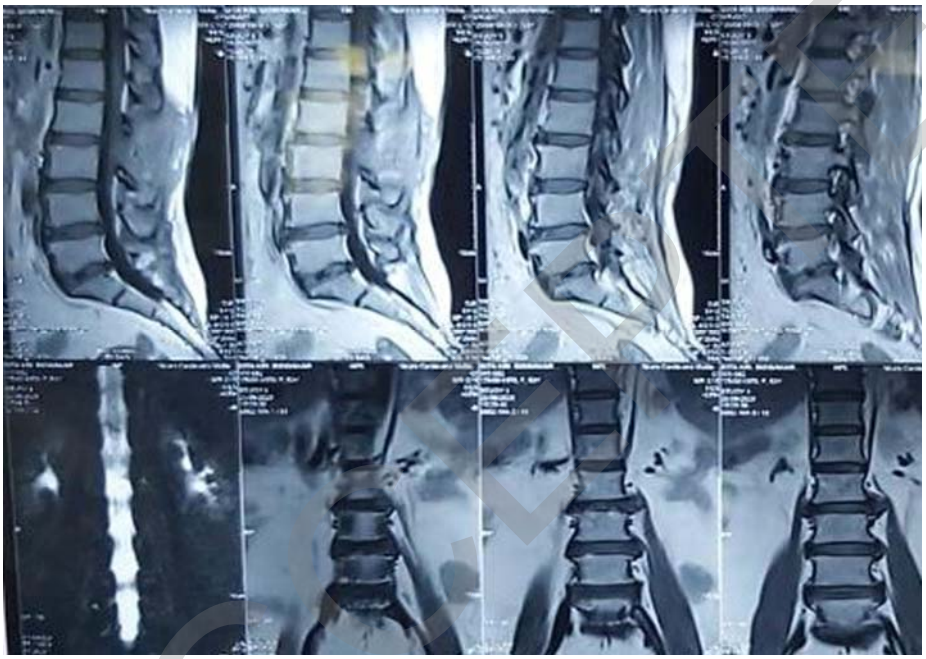

Figure 2: Postoperative tumor (schwannoma) after excision.