Case Report

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A rare case of schwannomatosis with meningioma: a case report

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Introduction: Schwannomatosis is characterized by multiple schwannomas without vestibular schwannomas or any other stigmata of neurofibromatosis type 2 (NF2). Schwannomatosis is a rare disorder, with a reported incidence ranging from 1 in 40 000 to 1 in 1.7 million. Meningioma is also associated with schwannomatosis in around 5% of cases.

Case presentation: We describe a case of a 20-year-old female presenting with progressive weakness of the right lower limb for 7 months with a tingling sensation and numbness of the same limb for 6 months and was found to have schwannomatosis with multiple spinal and right cerebellopontine angle (CPA) (9th/10th cranial nerve) schwannomas and left anterior cranial fossa meningioma.

Discussion: Schwannomas in schwannomatosis are seen along the cranial, spinal, and peripheral nerves but not along the vestibular nerve, as is characteristically seen in NF2. The occurrence of meningiomas is about 5% in individuals with schwannomatosis, and the patient in our case also had an associated meningioma. The tumor was confirmed to be a schwannoma based on features on an MRI examination and histological examination.

Conclusion: It is of great significance to identify the entire spectrum of the disease in a patient with schwannomatosis, and to differentiate it from related conditions in order to track and surgically manage the patient appropriately based on symptomatology and imaging findings.

Keywords: cerebellopontine angle schwannoma, meningioma, neurofibromatosis-2, schwannoma, schwannomatosis

Introduction

Schwannomas are Schwann cell-derived benign nerve sheath tumors that can develop on peripheral nerves or nerve roots[1,2]. Most of the schwannomas (95%) are solitary and sporadic in nature. The presence of multiple spinal cord tumors in a single patient is quite rare and often seen with neurofibromatosis (NF) and associated disorders[2,3]. Schwannomatosis, a distinct form of NF, is characterized by the development of multiple schwannomas without vestibular schwannomas or any other stigmata associated with neurofibromatosis type 2 (NF2)[3]. NF2 is a rare genetic disorder characterized by the presence of bilateral vestibular schwannomas, benign tumors that affect the vestibular nerve, and this disorder also shows variable involvement with meningiomas, ependymomas, neurofibromas, and spinal and peripheral nerve schwannomas[4]. Patients with schwannomatosis show phenotypic hallmarks of multiple schwannomas without concomitant involvement of cranial nerve VIII. Schwannomatosis is a rare disorder of unknown prevalence with a reported incidence ranging from 1 in 40 000 to 1 in 1.7 million, with a peak incidence reported between the ages of 30 and 60 years without any predilection for race or sex[4]. Meningioma, a benign intracranial tumor, is also associated with schwannomatosis, with an occurrence rate of around 5%[5]. In this case report, we try to understand the entire spectrum of disease in a patient diagnosed with schwannomatosis and appropriate surgical management for the same.

HIGHLIGHTS

- Schwannomatosis, a distinct form of neurofibromatosis, is characterized by the development of multiple schwannomas without vestibular schwannomas or any other stigmata associated with neurofibromatosis type 2 (NF2).
- Meningioma, a benign intracranial tumor, is also seen associated with schwannomatosis, with an occurrence rate of around 5%.
- Patients of schwannomatosis can be either symptomatic or asymptomatic. If the tumors are symptomatic, they should be removed surgically.

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Case presentation

Clinical history

A 20-year-old female with no significant past medical history presented with progressive weakness of the right lower limb for 7 months with tingling and numbness of the same limb for 6 months. The weakness in her limb was insidious in onset, which was initially noticed by slipping of slippers but gradually progressed to the point that she needed support while walking. She had a history of mild back pain for 1 month. Her bowel and bladder habits were normal. She had no history of shortness of breath, difficulty swallowing, blurring of vision, double vision, auditory disturbances, abnormal body movements, or trauma of any kind. She had no family history of malignancy or other tumors.

Examination findings

Her general condition was fair, her vitals were stable, and there were no positive findings on the general examination. The findings of higher mental function examination and other neurological examinations were within normal limits. Other neurological examination findings have been depicted in Table 1.

With this history and examination in mind, we suspected the possibility of a mass compressing the spinal cord clinically. Subsequently, MRI scans of her spine and brain were conducted to identify any apparent pathology.

Imaging data

The MRI findings were as below

Findings in the brain

Features suggestive of schwannoma in the right cerebellopontine angle

- An ~$11.0 \times 11.0 \times 10.0$ mm sized well-circumscribed, almost round extra-axial nodular lesion is seen in the right cerebellopontine angle region, adjacent to the medulla. It displays the iso signal in T1; iso-to-intermediate signal in T2 and FLAIR images. Few tiny foci of blooming with high signal intensity were seen in phase SWI (susceptibility-weighted imaging) images, suggestive of hemorrhagic foci. Intense, slightly heterogeneous enhancement is seen in post-contrast images (Fig. 1).

Features suggestive of left anterior cranial fossa meningioma

- Approximately $13.0 \times 7.5 \times 5.0$ mm sized oval extra-axial dural-based lesion is seen in the floor of the anterior cranial fossa superior to the left orbit. It displays iso signals in T1-weighted and T2-weighted images without foci of blooming. Intense, slight heterogeneous enhancement is seen in post-contrast images (Fig. 2).
- Other features of the brain are normal.

Findings in dorso-lumbar spine

Features suggestive of schwannomas in the dorso-lumbar spine

- There is ~$3.9 \times 2.2 \times 1.4$ (CC × TR × AP) (cranio-caudal × transverse or transaxial × antero-posterior) cm sized T1 intermediate to low and T2/fat saturated heterogeneously high signal intensity intradural extramedullary mass noted in the D11, D12 levels. It shows intense heterogenous enhancement in the post-contrast images. The mass is causing anterior displacement and severe compression of the spinal cord at this level. Minimal bilateral D11–D12 neural foraminal extension was noted; however, no extension along the nerve root to the prevertebral region is seen. No associated bony erosion or scalloping was noted.
- There are two homogeneously enhancing T2 intermediate signal intensity nodular lesions measuring 11×8 mm and 9×7 mm noted in the L1–L2 level, which are attached to the cauda equine nerve roots.
- There is ~$7.1 \times 4.0 \times 3.2$ (CC × TR × AP) cm sized heterogeneously enhancing expansile mass displaying T1 iso-to-low and T2 heterogeneously high signal intensity noted in the central spinal canal at L4 to S2 level. The mass is causing smooth erosion and scalloping of the posterior body of the vertebra as well as the posterior osseous component. There is near complete erosion/destruction of the posterior osseous elements at the S1 and S2 levels. The filum terminale and the cauda equine nerve roots are not separately visualized at these levels. Minimal bilateral LS–S1 and S1–S2 neural foraminal extension was noted; however, no extension along the nerve roots to the paravertebral region was seen. Small nodular enhancement is seen in bilateral L3 nerve roots at neural foramina (Fig. 3A, B).

The above-mentioned mass and nodules show mild restriction of diffusion in DWI images.

In summary, magnetic resonance imaging (MRI) of the brain with the whole spine showed ~$11 \times 11 \times 10$ mm well-defined, almost round intensely, and slight heterogenous extra-nodular lesion in the right cerebellopontine angle (CPA) adjacent to the medulla. Approximately $13 \times 7.5 \times 5$ mm oval extra-axial dural-based lesion is seen in the floor of the anterior cranial fossa superior to the left orbit. It displays iso signals in T1-weighted and T2-weighted images without foci of blooming. Intense, slight heterogenous enhancement is seen in post-contrast images. Also,

Table 1

Findings of neurological examination

Neurological examination	Right side	Left side
Motor Function (Power)	Upper limb: 5/5	Upper limb: 5/5
	Lower limb: 4/5 (all muscle groups)	Lower limb: 5/5 (all major muscle groups)
	Ankle dorsiflexion: 5/5	Ankle dorsiflexion: 4/5
	Hip flexion and extension: 5/5	Hip flexion and extension: 4/5
Sensory Examination	Decreased pain and touch sensations (right side, from L2 level) decreased vibration sensation (right foot)	All sensations are intact
Reflexes	Biceps, triceps, supinator jerks: 2+	Biceps, triceps, supinator jerks: 2+
	Knee jerk and ankle jerk: 3+	Knee jerk and ankle jerk: 3+
	Planters upgoing present	Planters upgoing present
	Clonus absent	Clonus absent

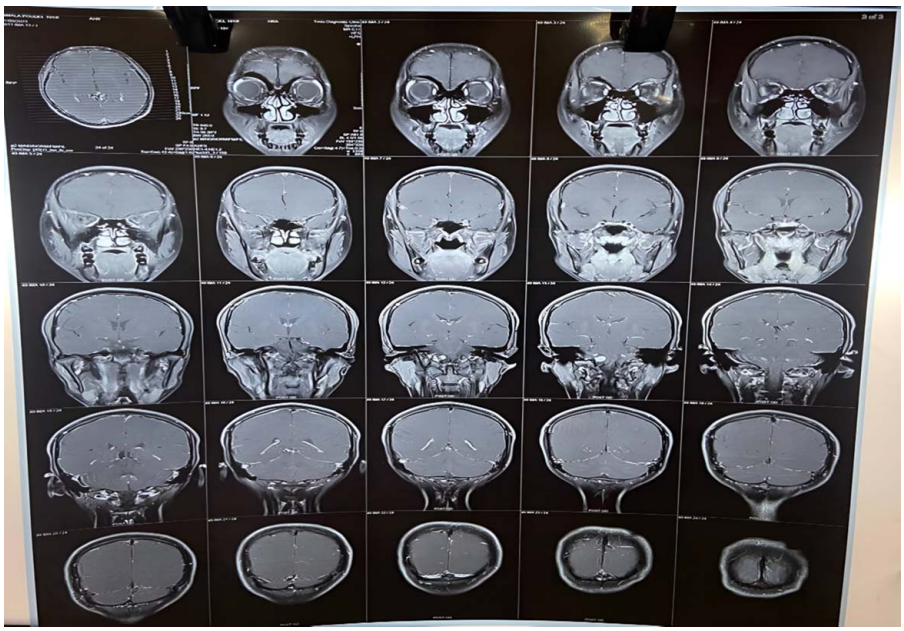

Figure 1. MRI brain showing a well-circumscribed, almost round extra-axial nodular lesion in the right cerebellopontine angle region, adjacent to the medulla.

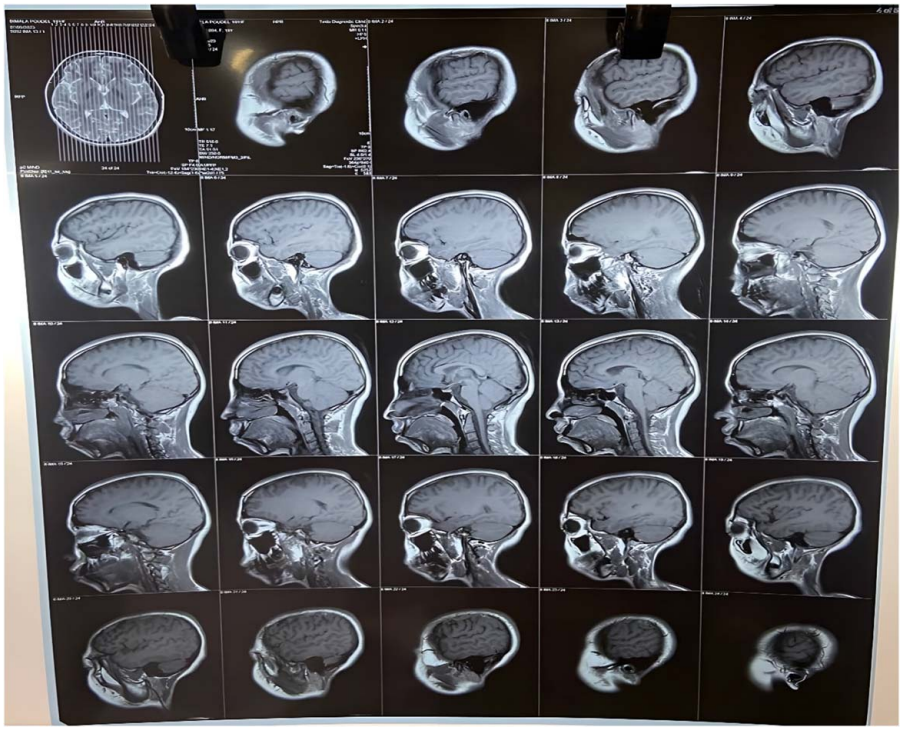

Figure 2. MRI brain showing an oval extra-axial dural-based lesion on the floor of the anterior cranial. Fossa superior to the left orbit.

heterogeneously enhancing expansile intradural extramedullary mass at D11–D12 level causes severe compression of the spinal cord with a normal cervical spine. These features on MRI are highly suggestive of schwannomatosis with multiple spinal and right CPA (9th/10th cranial nerve) schwannomas and left anterior cranial fossa meningioma.

The patient underwent laminectomy and tumor excision with laminoplasty. The operative findings of the patient showed a fleshy yellowish-white, soft tumor with vascularity and thick walls on the T11–T12 level with a tumor anterior and right of the spinal cord; at L2 level with a single 2 cm × 2 cm, globular tumor to the left of the cord; and at the L4–S2 level with a tumor

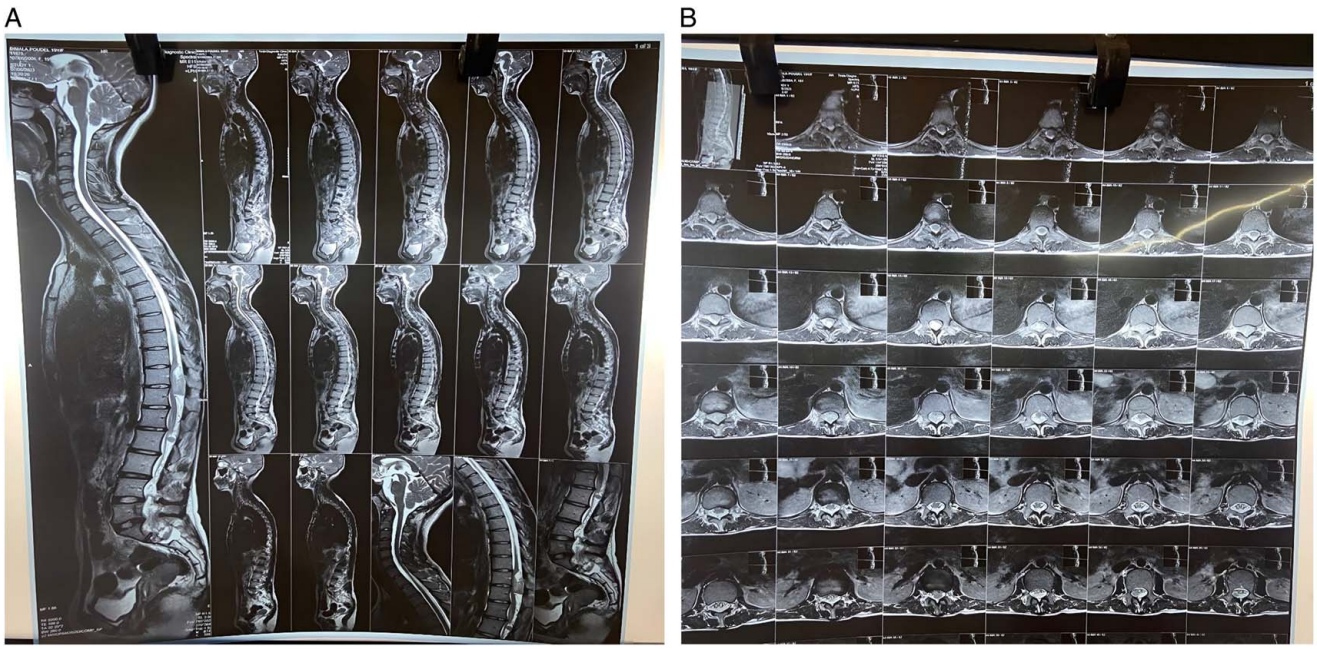

Figure 3. A, B: showing heterogeneously enhancing expansile intradural mass at L4–S2 level with smooth erosion/scalloping of the vertebrae, heterogeneously enhancing intradural extramedullary mass at D11–D12 level causing compression of the cord and enhancing intradural nodular lesions at L1–L2 level and bilateral L3–L4 neural foramina.

adherent to the dura with thinning of the dura, diffuse with bony scalloping. Both the CPA tumor and the meningioma were excised in the same setting via craniotomy, after which the patient was shifted to the ICU. Her postoperative ICU stay was for a week, which was uneventful. The patient was discharged from the hospital 9 days after the surgery with GCS (Glasgow Coma Scale) 15, and power was normal in all four limbs. The patient has been kept on regular follow-up at the neurosurgery outpatient clinic. There was a gradual improvement in the power and sensory functions of the affected limb, and the patient made a full recovery without any postoperative complications. No recurrence of spinal tumors has been noted.

Discussion

Schwannomas are benign, encapsulated, slow-growing nerve sheath tumors consisting of Schwann cells, representing about one-third of all benign primary spinal tumors[6]. The presence of multiple spinal cord tumors in a single patient is extremely rare and most often seen in NF and associated disorders, while considering the fact that most of the schwannomas are typically solitary[2,3]. Schwannomatosis is a term used for patients with multiple schwannomas and is often considered a third form of NF[6,7]. Schwannomas in schwannomatosis are seen along the cranial, spinal, and peripheral nerves but not along the vestibular nerve, as is characteristically seen in NF2. Similarities in the presentation and phenotype of the disease can be seen in patients with schwannomatosis and NF2, which are distinct entities[7], making it essential to differentiate between the two. The presence of bilateral vestibular schwannomas with variable involvement of meningioma, ependymoma, neurofibromas, and spinal and peripheral nerve schwannomas is characteristic of NF2. The presence of vestibular schwannomas rules out the possibility of schwannomatosis, and this marks the significance of high-quality radiological investigations in making the distinction between the two[4].

Schwannomatosis was first reported in 1973 as NF type 3[6]. Schwannomatosis is a rare disorder with an estimated incidence of 1 in 40 000 to 1 in 1 700 000 characterized by the occurrence of multiple schwannomas and much less commonly meningiomas. The occurrence of meningiomas is about 5% in individuals with schwannomatosis and has been reported in individuals with SMARCB1 pathogenic variants[8,9]. The patient in our case also had an associated meningioma. However, genetic analysis was not possible at our center to identify the form of the associated mutation in this case[4].

The incidence of schwannomatosis in reported literature is quite limited, and the spectrum of features and diagnostic criteria is still not well understood[6]. According to the diagnostic criteria defined by Kehrer-Sawatzki *et al*.[9], the reported patient can be considered a possible case of schwannomatosis. She has more than two non-intradermal schwannomas with histological confirmations and no evidence of vestibular schwannomas on a high-quality MRI examination and detailed study of the internal auditory canal. In addition to this, ophthalmological and general physical examinations did not reveal any findings suggestive of NF, and there was no family history of NF or schwannomatosis. The lack of vestibular schwannomas also helped us exclude the diagnosis of NF2.

Among the patients with schwannomatosis, one-third are found to have tumors in an anatomically limited distribution, such as a single limb or less than 5 contiguous segments of the spine, known as segmental schwannoma, which has an occurrence rate of 1 in 120 000[10]. In our case, the spinal schwannomas are located at T11–T12, L2, and L4–S2, with each covering less than five contiguous segments of the spine. In addition to this,

the patient has a right CPA (9th/10th cranial nerve) schwannoma and a left anterior cranial fossa meningioma.

The major symptoms associated with the disease include pain, progressive motor deficits, or signs of spinal cord compression due to the mass effect[7]. The patient in this case presented with the leading symptoms of progressive weakness, including tingling and numbness in the right lower limb. The tumor was confirmed to be schwannoma based on features on MRI examination and histological examination[4], wherein the presence of multiple discrete, well-defined, round-to-oval lesions situated along the course of the peripheral nerve with T1 iso-to-low and T2 heterogeneously high signal intensity suggests schwannomatosis and the presence of two distinct histological patterns termed 'Antoni A' and 'Antoni B' regions suggests schwannomas.

Patients with schwannomatosis can be either symptomatic or asymptomatic; asymptomatic patients are managed conservatively, while surgery is indicated for symptomatic lesions secondary to schwannoma and due to spinal cord compression[7,11]. The presented case is of a symptomatic patient, so the tumors were removed surgically by the operative procedure 'laminectomy and tumor excision with laminoplasty.' In addition to this, craniotomy and excision of both the CPA tumor and meningioma were done in the same setting.

Conclusion

Schwannomatosis and NF2 are two different pathologies, and an accurate diagnosis is important. It is also essential to examine a patient with schwannoma for a possible diagnosis of schwannomatosis. Schwannomas are typically slow-growing tumors whose clinical manifestations depend upon the level of the spinal cord involved. Symptoms can progress rapidly if spinal cord compression occurs. Contrast-enhanced MRI is the most sensitive and specific imaging modality to evaluate possible spinal column lesions. Patients with schwannomatosis can be either symptomatic or asymptomatic. If the tumors are symptomatic, they should be removed surgically.

Ethical approval

Ethical approval is not required for the case report in our institution, Tribhuvan University Teaching Hospital.

Consent

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

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Author contribution

S.N., A.K., S.P., and K.K.Y.: wrote the original manuscript, reviewed, and edited the original manuscript; G.B., P.Y., A.A., S. K.K., S.P., and A.G.: reviewed and edited the original manuscript.

Conflicts of interest disclosure

The authors declare no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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Data availability statement

All the relevant data have been included in the manuscript itself.

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