

Case Report

Solitary Neurofibroma Mimicking Giant Cell Tumor of The Upper Cervical Spine: A Case Report

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ABSTRACT

Background: Spinal neurofibromas are benign peripheral nerve sheath tumors typically occurring in the thoracic region. Involvement of the C2 vertebra is particularly rare. Atypical presentations of solitary neurofibromas can pose diagnostic challenges when their radiological features resemble other neoplastic entities.

Case Report: A 35-year-old male presented with a 3-month history of neck pain and no history of trauma. Physical examination revealed mild bulging and localized tenderness in the posterior neck, with restricted neck extension to 30 degrees. Plain radiography showed a round, expansile lytic lesion with well-defined, nonsclerotic borders on the C2 spinous process, while MRI confirmed enhancing solid components and extensive bone involvement, indicative of a giant cell tumor. The histopathology from the core needle biopsy was inconclusive, showing few inflammatory cells and no evidence of malignancy. The lesion was marginally resected from a posterior approach, achieving only subtotal resection to preserve the vertebral artery. Histopathological analysis from the open biopsy confirmed the diagnosis of neurofibroma. At the 3-month follow-up, postoperative imaging showed the residual tumor. Despite this, the patient reported significant neck pain relief.

Discussion: Radiography is insufficient for differentiating spinal tumors; therefore, histopathological biopsy is necessary for an accurate diagnosis. Open biopsy offers higher diagnostic accuracy than core needle biopsy.

Conclusion: Neurofibroma should be considered in differential diagnosis for patients initially suspected of giant cell tumors based on radiological findings.

Keywords: Neurofibroma; Giant cell tumor; Cervical spine; Human and medicine

INTRODUCTION

Neurofibromas are benign peripheral nerve neoplasms arising from various cell types, including Schwann cells and perineural fibroblasts.¹ While commonly associated with neurofibromatosis type I (NF-1), they can occur sporadically at various anatomical sites.² Spinal neurofibromas, constituting only 2-5% of primary spinal neoplasms, are particularly rare.³ Occasional atypical presentations of solitary neurofibromas, especially in the absence of clinical signs of NF-1, can

pose significant diagnostic challenges, particularly when they resemble other neoplastic entities such as giant cell tumors. To the best of our knowledge, no previous cases have been reported in the literature describing a sporadic solitary cervical neurofibroma radiographically resembling a giant cell tumor. Herein, we present the case of a 35-year-old male patient diagnosed with a solitary cervical neurofibroma, initially presenting radiographic features suggestive of a giant cell tumor. This case report adheres to the SCARE 2020 guidelines.⁴



CASE REPORT

A 35-year-old male, presented with a primary complaint of neck pain that had developed three months prior. The pain is exacerbated upon cervical extension, consequently leading to a restriction in the range of neck movement. He had initially sought treatment at local pain clinics and received analgesic medication, but his symptoms worsened over the subsequent months. The patient reported no history of trauma, prior neck surgeries, neurological deficits, or systemic symptoms. During the examination, the patient exhibited a mild bulging in the upper cervical spine with localized tenderness and restricted neck extension to 30 degrees. Neurovascular assessment showed normal findings.

Initial plain radiographs revealed a round expansile lytic lesion with well-defined borders on the posterior tubercle of the C1 vertebra and the spinous process of the C2 vertebra, without periosteal reaction (Figure 1A). Cervical Magnetic Resonance Imaging (MRI) revealed the lesion to measure 3.8 x 4.4 x 4.6 cm (anteroposterior x lateral x craniocaudal) in size, with predominantly solid

components and lobulated cystic areas, appearing hypointense on T1-weighted images, hyperintense on T2-weighted images (Figure 1B-C), increased signal on Short Tau Inversion Recovery (STIR) sequences compared to muscle, and heterogeneous contrast enhancement on Gadolinium-enhanced images. The three-dimensional Computed Tomography (CT) scan reconstruction confirmed significant bone involvement extending into the lamina, left pedicle, left anterior and posterior arches, and the body of the C2 vertebra (Figure 1D-F). Based on imaging findings, a diagnosis of giant cell tumor was highly suspected.

A core biopsy was performed to confirm the suspected diagnosis. However, the histopathology results were inconclusive, consisting of adipose tissue, blood, and a few inflammatory cells, with no evidence of malignancy (Figure 2A). Laboratory tests, including complete blood count and biochemical profile, showed no abnormalities. Given the clinical presentation and imaging findings, marginal excision of the tumor was planned to achieve pain relief, improve cervical range of motion, and halt tumor progression to mitigate potential neurological deficits.

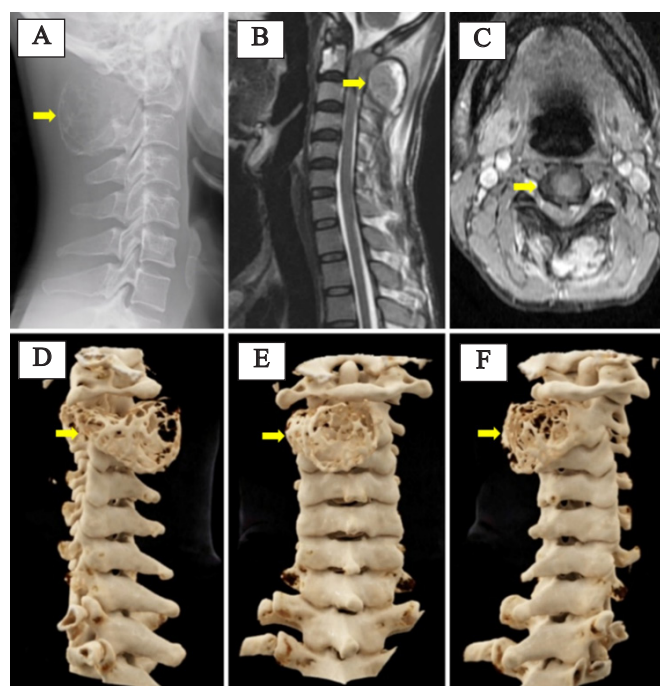


Figure 1. Preoperative imaging of the cervical spine. (A) Lateral plain radiograph demonstrating an expansile lytic lesion involving the spinous processes of C1 and C2 (arrow). (B, C) T2-weighted MRI showing a hyperintense lesion within the C2 vertebra on sagittal (B) and axial (C) views (arrows). (D–F) Three-dimensional CT reconstruction revealing extensive osseous involvement of the lesion (arrows) from the left posterolateral (D), posteroanterior (E), and right posterolateral (F) perspectives.



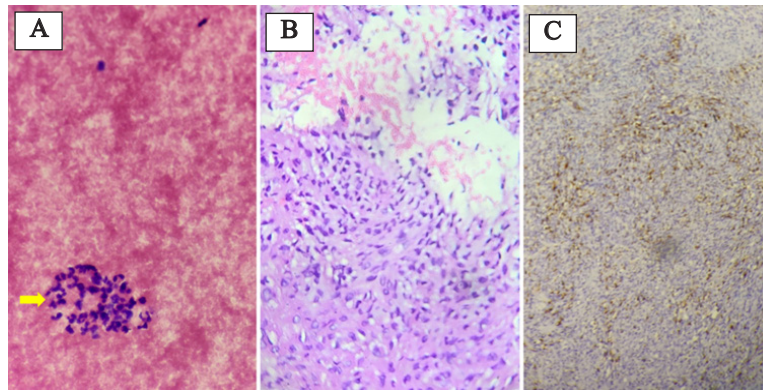


Figure 2. Histopathological examination of the lesion. (A) Core biopsy specimen stained with H&E (×40), revealing the presence of polymorphonuclear (PMN) cells (arrow). (B) Open biopsy specimen stained with H&E (×40), showing tumor cell morphology. (C) Immunohistochemical staining of the surgical specimen for S-100 protein (×40), demonstrating diffuse positivity.

The surgical procedure was conducted through a posterior approach. Following the initial incision and subperiosteal dissection, a tumor located at the spinous process of the C2 vertebra was identified as an unencapsulated, well-defined, grey-tan firm mass. Pedicle screw fixation was performed bilaterally from C1 to C4 vertebrae, excluding the left C2 vertebra due to tumor involvement. Marginal excision of the tumor was attempted; however, due to the tumor's extensive involvement, complete removal was unachievable through the posterior approach alone, as total excision carried a significant risk of injuring the vertebral artery in the transverse foramen. Subsequent laminectomy for decompression at the C3 and C4 vertebrae was performed, followed by posterior spinal fusion to stabilize the spine. The resected mass measured approximately 3.0 x 3.5 x 4.0 cm and was sent for further histopathological examination. The tumor materials were collected during the procedure for further histopathological examination. The patient experienced a blood loss of 1500 ml during surgery. Hemostasis was achieved using a combination of meticulous surgical technique, local hemostatic agents, and intraoperative transfusion of packed red blood cells to maintain hemodynamic stability. The patient was discharged in stable condition seven days after surgery, without any observed complications during the hospitalization period. Initial rehabilitation was initiated one week after discharge.

The final histopathological result showed monomorphic spindle cells with abundant cytoplasm and coarse chromatin within the nuclei, along with nuclear palisading and rare mitotic figures, as observed with Hematoxylin and Eosin (H&E) staining (Figure 2B). Immunohistochemical analysis provided additional support for the diagnosis, demonstrating positivity for S-100 protein focally in approximately 50% of tumor cells (Figure 2C) and negative staining for CD-68, confirming the diagnosis of neurofibroma.

At the 3-month follow-up, no neurological deficits or surgical complications were observed. Plain radiograph evaluation revealed an inhomogeneous opacity with well-defined, regular margins projecting on the C2 vertebra, suggesting the presence of a residual lesion without any signs of recurrence or expansion into adjacent tissues (Figure 3A-B). Three-dimensional CT scan reconstruction further demonstrated lytic expansion and cortical thinning within the affected anatomical structures (Figure 3C-E). Additionally, MRI findings reveal an extramedullary residual mass with solid components involving the C2 vertebral body, left pedicle, left transverse process, left lamina, and spinous process. The mass shows hypointensity on T1-weighted images and hyperintensity on T2-weighted images relative to muscle tissue. Post-contrast imaging demonstrates homogeneous strong contrast enhancement. Despite these findings, the patient reported improved relief from neck pain.



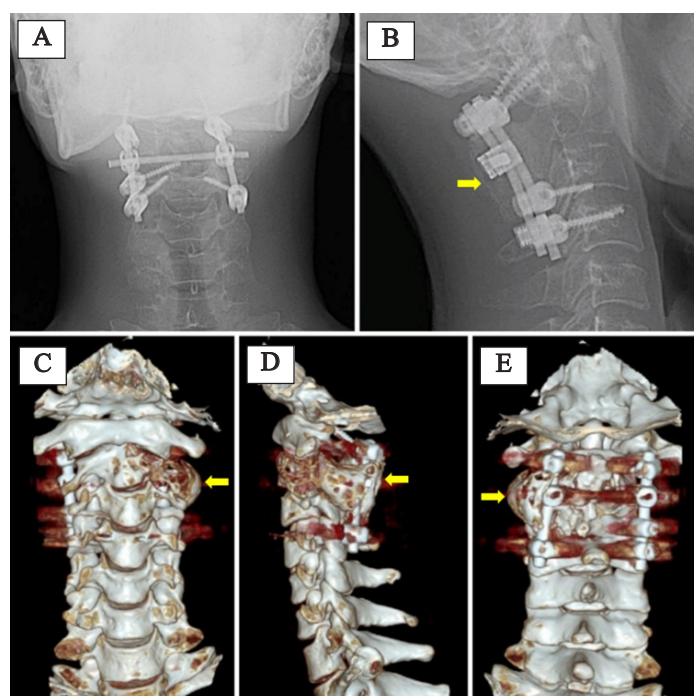


Figure 3. Three-month postoperative imaging of the cervical spine. (A) Anteroposterior plain radiograph showing alignment of the cervical vertebrae post-tumor resection. (B) Lateral plain radiograph demonstrating residual lesion with regular margins at the C2 level. (C–E) Three-dimensional CT reconstructions illustrating lytic expansion and cortical thinning associated with the residual mass (arrows), viewed from the anterior (C), lateral (D), and posterior (E) perspectives.

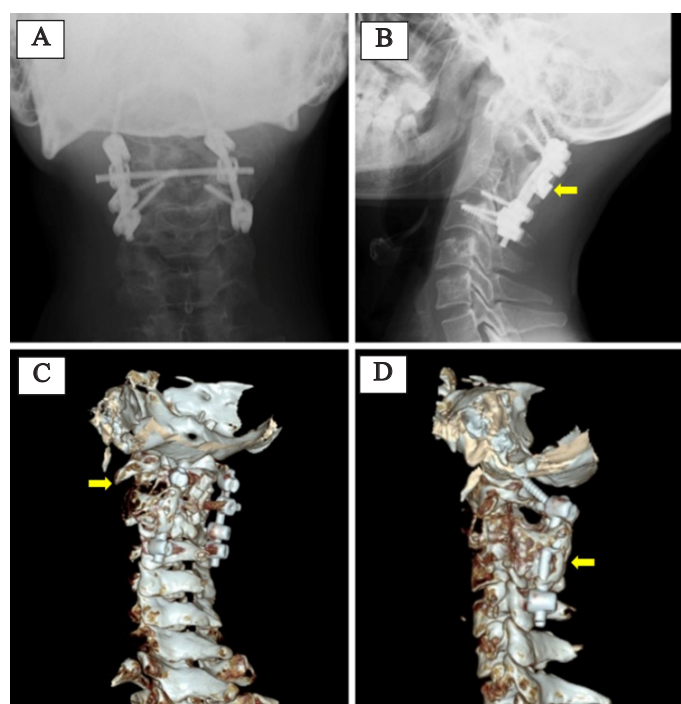


Figure 4. Nine-month postoperative imaging of the cervical spine. (A) Anteroposterior plain radiograph showing maintained alignment and intact posterior instrumentation from C1 to C4. (B) Lateral plain radiograph demonstrating a residual lesion at the C2 level with no evidence of interval growth. (C–D) Three-dimensional CT reconstructions showing a stable residual lytic lesion with cortical thinning without signs of progression or new involvement, viewed from the posterior oblique (C) and lateral (D) perspectives.

and was able to perform cervical extension up to 10° without discomfort. The limited range of motion at this stage was attributed to the use of a cervical

brace, which was maintained until the end of the third postoperative month. The patient expressed satisfaction with the overall outcome.



At the 9-month postoperative follow-up, plain radiographs demonstrated maintained cervical alignment and stable posterior instrumentation (Figure 4A-B), while contrast-enhanced three-dimensional CT imaging showed no evidence of tumor recurrence or progression of the residual lesion (Figure 4C-D). The patient remained asymptomatic, and cervical extension had improved to 55 degrees without pain. A subsequent follow-up is scheduled one year after this evaluation to monitor for any delayed recurrence.

DISCUSSION

Neurofibromas most commonly occur in the skin, with spinal neurofibromas being relatively rare, constituting only 2-5% of primary spinal neoplasms.^{3,5} While spinal neurofibromas most frequently show a location in the thoracic region, involvement of the C2 vertebra is particularly uncommon. Up to 60% of individuals with NF1 may develop spinal neurofibromas.⁶ However, this patient did not exhibit the hallmark clinical features of NF1, which include multiple café-au-lait macules, intertriginous freckling, multiple cutaneous neurofibromas, subcutaneous or deep nodular neurofibromas, plexiform neurofibromas, or characteristic ocular signs.⁷ Furthermore, there is no family history of neurofibromatosis, suggesting that the patient's condition is likely sporadic.

The misleading factor that led to the initial misdiagnosis of a giant cell tumor was the patient's radiographic images. Although cervical spine involvement by giant cell tumors is rare, occurring in only 0.4% to 1.0% of cases, the patient was initially misdiagnosed due to specific imaging features.⁸ A giant cell tumor typically shows purely lytic features with well-defined but non-sclerotic margins, as observed in our patient.⁹ While radiography is generally reliable for diagnosing giant cell tumors in the appendicular skeleton, its diagnostic accuracy decreases in the spine, where biopsy and histopathological examination remain essential to distinguish giant cell tumors

from other spinal pathologies.¹⁰

Tissue biopsy is considered the gold standard for diagnosing neurofibromas.² Neurofibromas may exhibit variability in cellular composition across different regions of the tumor, which small core biopsies may inadvertently capture areas dominated by inflammatory polymorphonuclear cells rather than the characteristic cells such as Schwann cells and perineural fibroblasts. Open biopsy provides a larger and more comprehensive tissue sample, and may enhance the ability to accurately identify specific tumor characteristics. However, the diagnosis does not significantly impact the treatment plan, which prioritized symptom control, structural stabilization, and histopathological confirmation.

One regrettable aspect of this case is the missed opportunity to explore the potential benefit of a combined posteroanterior approach to achieve total resection, potentially decreasing the recurrence rate. In this patient, the decision to pursue a posterior-only approach was influenced by the predominant posterior location of the mass. However, to preserve the integrity of the vertebral artery within the transverse foramen, only subtotal resection was feasible, prioritizing neurovascular preservation over complete tumor removal. Consequently, postoperative imaging revealed a residual tumor. The patient remained asymptomatic and could extend his neck without pain, successfully achieving the surgical goals. No adjuvant chemotherapy was given, as the tumor was benign with no malignant features. Given the subtotal resection, regular clinical and radiological follow-up was planned to monitor for recurrence. At the 9-month evaluation, there was no sign of progression, and the next follow-up is scheduled at 21 months.

CONCLUSION

Solitary cervical neurofibroma is rarely encountered, occasionally exhibiting radiographic features that mimic giant cell tumors, as observed in this



case. Therefore, despite its rarity, neurofibroma should be considered in the differential diagnosis of patients suspected to have a giant cell tumor. A core needle biopsy may be insufficient for a definitive diagnosis of neurofibroma, whereas an open biopsy provides greater diagnostic accuracy.

ACKNOWLEDGEMENTS

The authors would like to thank the patient and their family for providing consent and allowing us to share this case. We also express our gratitude to the medical team at Dr. Moewardi Regional General Hospital for their support in the management and documentation of this case.

FUNDING

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

CONFLICT OF INTEREST

The authors declare that they have no competing interests.

ETHICAL APPROVAL

Ethical approval was not required for this case report in accordance with the institutional policy.

INFORMED CONSENT

Written informed consent for the publication of this case report and any accompanying images was obtained from the patient.

DATA AVAILABILITY STATEMENT

All data generated or analyzed during this study are included in this published article. Additional data are available from the corresponding author on reasonable request.

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