Cervicomedullary cavernous malformation of the C1 nerve root: illustrative case

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BACKGROUND Cavernous malformations (CMs) originating from spinal nerve roots are rare but can present with rapidly progressing neurological deficits. Therefore, quick identification and treatment are essential.

OBSERVATIONS The authors present the case of a 52-year-old male presenting with headaches and gait disturbance, found to have a CM of the C1 nerve root. The patient underwent successful suboccipital craniectomy and C1 laminectomy, with complete resection of the lesion, preservation of the cervical spinal nerve roots, and symptomatic improvement postoperatively.

LESSONS This is the first presentation in the literature of a CM originating from the C1 nerve root. The authors show complete resection of the lesion with preservation of the C1 nerve root. Nerve root lesions usually show insidious symptomatic onset. Quick identification and resection are recommended to prevent permanent neurological disability.

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KEYWORDS arteriovenous malformation; surgery; cavernous malformation

Cavernous malformations (CMs) are mulberry-like vascular lesions comprised of irregular, dilated capillaries without intervening neural or glial elements.¹ ² These capillaries are lined by a single layer of endothelium without a muscular layer or internal elastic lamina.³ Although CMs are considered low-flow lesions (in comparison to arteriovenous malformations [AVMs] and other high-flow lesions), the leakiness of the abnormal endothelium can lead to hemorrhage.³ The estimated prevalence of CMs of the central nervous system (CNS) is 0.4%–0.6%.¹ Spinal CMs are rare entities, with those originating from the nerve root even rarer.

Here, we present the case of a patient with a symptomatic spinal CM of the C1 nerve root at the cervicomedullary junction. Imaging characteristics are presented and discussed in the context of the differential diagnosis. The patient underwent gross-total resection of the lesion without complication. He remained symptom-free at follow-up.

Illustrative Case

A 52-year-old male with no significant past medical history presented with 2 weeks of daily positional headaches that woke him from sleep, accompanied by several days of gait disturbance. On physical examination, he was neurologically intact with no long tract signs, and he presented with a Frankel grade E.

Head computed tomography scan showed multiple hyperattenuating intracranial lesions and a 1.5-cm lobular intradural extramedullary mass along the dorsal aspect of the cervical spinal cord at the level of the cervicomedullary junction. On magnetic resonance imaging (MRI), the cervicomedullary mass was noted to be T1 and T2 hyperintense, homogeneously contrast-enhancing, and had a macrolobulated appearance with a peripheral gradient echo (GRE)–positive margin (Fig. 1A–D). The scattered intraparenchymal lesions also demonstrated a GRE signal (Fig. 1E and F). There were no associated short tau inversion recovery signal changes in the cervical cord. Differentials included CMs, ruptured dermoid cysts, and metastases. Metastatic workup showed no evidence of malignancy.

The patient was taken to the operating room for suboccipital craniotomy and C1 laminectomy for resection of the lesion with neuronavigation and neuromonitoring. The lesion was noted to be an approximately 1.5-cm exophytic CM that appeared to be originating...
from a cervical nerve root of C1 (Fig. 2). The left 11th cranial nerve and spinal artery were dissected off the cavernoma. The inferior pole of the CM was dissected first, followed by the lateral and the superior pole. The anterior edge of the CM was dissected last. Complete resection of the lesion was achieved, and the cervical spinal nerves were identified and preserved (Video 1).

**VIDEO 1.** Intraoperative clip demonstrating the successful resection of the C1 CM. Click here to view.

The patient tolerated the procedure well and was neurologically intact in the postoperative period. He was discharged home on postoperative day 2. At his 1-week follow-up, the patient was noted to be doing well with mild neck stiffness. The patient remained neurologically intact with improvement in gait disturbance.

**Histopathological Result**
Pathology revealed clots, blood products, and thin-wall blood vessels of various diameters associated with hemosiderin-laden macrophages, consistent with a diagnosis of CM (Fig. 3).

**Patient Informed Consent**
The necessary patient informed consent was obtained in this study.

**Discussion**

**Observations**
Here, we report the case of a CM originating from the C1 nerve root. To our knowledge, this is the first case ever reported in the literature on this topic. The vast majority of CNS CMs are intracranial and intraparenchymal. A small minority of CMs involve the spine, most commonly the thoracic spine (55.2%), followed by the cervical spine (38%). Only 3 cases of CMs at the cervicomedullary junction have previously been described in the literature. Stone et al. reported the presence of a cavernous angioma situated in the dorsal midline of the upper-most cervical spinal cord. Mocco et al. described an intradural extramedullary CM at the foramen magnum. Wang et al. reported a single cervicomedullary junction lesion in their 137-patient study.
cavernomas only exhibit edema during acute hemorrhage, which typically occurs during the first hemorrhage. Most recently, Henderson et al. reported a C6 ventral root cavernoma, while older case reports have shown lesions between C4 and C7. 

In a meta-analysis including 632 patients with spinal CMs, the mean age of presentation was 39 years, and the male/female ratio was 1.1:1. While our patient had no known family history of vascular malformations, a positive family history of cavernomas was noted in 11.9% of patients with spinal CMs.

While intracranial CMs tend to present with symptoms like seizures or focal neurological deficits, spinal CMs can present with a more rapid or severe progression of neurological impairment. Commonly, the initial presentation of spinal CMs involves motor (60.5%) or sensory (57.8%) symptoms, although pain or bowel/bladder symptoms have also been reported. About 45.4% of patients with spinal CMs exhibit an acute step-wise clinical deterioration suggestive of a hemorrhagic event into the surrounding parenchyma, while 54.6% of patients have a slowly progressive onset of symptoms, consistent with internal microhemorrhages and/or gradual enlargement of the CM. The bleeding rate for spinal CMs is as high as 66%. Of note, at least 2 of the 3 cases of cervicomedullary CMs described in the literature presented with headache, as with our patient. This may be suggestive of obstructive symptoms or cerebrospinal fluid outflow impairment at the level of the foramen magnum even in the absence of radiographic ventriculomegaly.

The annual rate of hemorrhage of spinal CMs is thought to be approximately 1.6%-4.1%. CMs closely associated with spinal nerve rootlets, as was seen in our patient, are thought to be at higher risk of hemorrhage due to tethering. Patients may present with acute subarachnoid hemorrhage, given the communication of these lesions with the subarachnoid space.

**Imaging**

On MRI, CMs can appear heterogeneous (“popcorn appearance”) on T1- and T2-weighted sequences depending on the stage of hemorrhage. GRE is highly sensitive to CMs due to hemosiderin deposition. Contrast enhancement is generally minimal.

In this case, the cervicomedullary lesion was unusual on MRI in its marked T1 and T2 hyperintensity and strong contrast enhancement, with GRE signal only along its periphery likely representing a rim of hemorrhagic products. The appearances of the scattered intracranial lesions were more classic for CMs, with GRE signal and mixed T1 and T2 intensity. The nonclassical appearance of the cervicomedullary lesion on MRI led to some diagnostic ambiguity, with a ruptured dermoid cyst (typically T1 hyperintense due to lipid content, with scattered cyst contents in the sulci and subarachnoid space) remaining on the differential. Chemical meningitis from cyst rupture may also cause a subacute headache consistent with our patient’s presentation.

A hemorrhagic metastasis may be similar to a CM; however, tumor hemorrhages are usually smaller than the tumor itself, and repeat hemorrhages are rare. Unlike brainstem tumors with prolonged edema, cavernomas only exhibit edema during acute hemorrhage, which typically resolves within days.

Differentiating CMs from multiple sclerosis on MRI is challenging due to similarities in inactive lesions. Both conditions can present with progressive or recurrent neurological decline. However, a key distinction is that CMs exhibit symptomatology localized to a specific neuraxis location, whereas multiple sclerosis lesions are more widespread. Additionally, demyelinating lesions in multiple sclerosis consistently respond to steroids and show rapid changes on serial imaging. AVMs are typically high-flow lesions, whereas CMs are low flow; however, a thrombosed low-flow AVM cannot be completely excluded. Hematomas from AVMs lack the characteristic “popcorn” feature. Ependymomas and hemangioblastomas are also associated with hemorrhages, but they rarely present with a mix of subacute and chronic bleeding, and contrast enhancement is expected.

In patients with spinal CMs, it is important to consider imaging for intracranial lesions as well. It has been reported that as many as 16.5%-47% of patients with spinal CMs have concurrent intracerebral CMs, as our patient did.

**Management**

In general, early gross-total resection of spinal CMs is recommended due to the risk of hemorrhage and progressive neurological deterioration, particularly for patients whose clinical presentation follows a more acute stepwise pattern of decline. In retrospective comparisons, rates of improved neurological outcomes are higher among patients who undergo surgery versus those who are managed conservatively with observation (51% vs 30%).

For spinal CMs, electromyography monitoring with motor evoked potentials and somatosensory evoked potentials is an important operative adjunct. Sharp dissection around the periphery of the CM, coagulation with fine-tipped bipolar electrocautery, and careful mobilization of the lesion from the spinal cord are recommended to avoid injury to the cord parenchyma. A gliotic plane can often be identified between the CM and the spinal cord, facilitating circumferential delivery of the lesion; a cottonoid can be used to further define this plane and aid in resection.

In cases in which the CM is closely associated with the spinal nerve rootlets, it may be necessary to sacrifice the spinal nerve rootlet. However, in this case, a plane could be found allowing mobilization of the cavernoma off of the spinal nerve, allowing for preservation of the nerve.

**Lessons**

In this case report, we describe the successful resection of a spinal CM at the cervicomedullary junction originating from the C1 nerve root. Spinal CMs are rare but can present with a more rapid onset of neurological deficits compared to their intracranial counterparts. Cervicomedullary CMs, in particular, may present with a headache due to obstruction of the foramen magnum. In patients with spinal CMs, especially those with acutely presenting symptoms, resection is recommended to prevent further hemorrhage and the development of permanent deficits. Brain imaging to identify concurrent intracranial CMs is also recommended.

**References**


Disclosures
The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

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Conception and design: Chang, Zhang, Marianayagam, Kumar, Fatemi. Acquisition of data: Chang, Yuan, Zhang, Marianayagam, Kumar. Analysis and interpretation of data: Zhang, Marianayagam, Kumar. Drafting the article: Zhang, Yuan, Marianayagam, Kumar. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Chang. Statistical analysis: Zhang. Administrative/technical/material support: Park. Study supervision: Chang.

Supplemental Information
Videos

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