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Case Report

Cauda equina myxopapillary ependymoma in von Hippel-Lindau disease: A case report

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ABSTRACT

Background: Patients affected by Von Hippel-Lindau (VHL) are prone to develop central nervous system neoplasms such as hemangioblastomas (HBs). Myxopapillary ependymoma (MPE) is not commonly associated with VHL disease.

Case Description: We present the first case of a VHL patient affected by simultaneous silent cauda equina MPE and a symptomatic conus medullaris HB. The patient was first operated for systemic tumors and followed for asymptomatic HBs. Simple surveillance was maintained until neurological symptoms appeared. Regular follow-up demonstrated objective growth of the cystic conus medullaris tumor while the cauda equina lesion remained stable. Surgery was performed to avoid further neurological worsening. Histopathological examination showed conus medullaris HB and a nearby cauda equina MPE.

Conclusion: Simultaneous spinal HBs and isolated MPE may exceptionally occur in VHL patients.

Keywords: Cauda equine, Conus medullaris, Hemangioblastoma, Myxopapillary ependymoma, Von Hippel-Lindau

INTRODUCTION

Von Hippel-Lindau (VHL) disease is a genetical syndrome[8] related to the mutation of the tumor suppressor gene VHL. Affected patients will develop various neoplasms, including visceral tumors, central nervous system (CNS) hemangioblastomas (HBs), and retinal HB (RH).[8] Progression of CNS HB is known to be the leading cause of VHL-associated morbidity and mortality.[7] HB is the most common VHL-associated tumor and arises all along the neuraxis with a predominant localization in the posterior fossa and spinal cord.[4], Although rarely symptomatic, cauda equina HB is frequent in VHL patients.[9] Potential and most frequent differential diagnoses of cauda equina HB include nerve sheath tumors, myxopapillary ependymomas (MPEs), and meningiomas.[13]

To the best of our knowledge, we describe the first case of a VHL patient affected by a concomitant symptomatic conus medullaris HB and underlying silent cauda equina MPE.

CASE PRESENTATION

An adult patient in his 30s was affected by a familial VHL disease with past surgeries for VHL-associated tumors (i.e., pheochromocytomas, clear cell renal tumors, endolymphatic sac tumors, and pancreatic neuroendocrine tumors) that had asymptomatic CNS tumors diagnosed

during complementary workup. Suspected HBs were identified in the posterior fossa, thoracic spinal cord, conus medullary, and cauda equina. All lesions but one remained stable and silent during radiological monitoring. Objective and exponential growth of the conus medullaris cystic tumor was observed within a few months. The cystic portion grew rapidly while the mural nodule stayed stable in size [Figure 1].

In the meantime, the neurological status rapidly deteriorated from a slight right perineal hypoesthesia to important pains with contractures predominantly on the left side without obvious motor deficit. The patient progressively felt his urinary stream much less powerful, with hypoesthesia spreading to the left perineum, which was not present during past consultations.

Hence, surgery was decided to avoid further neurological worsening and protect sphincter functions. Complete resection of both symptomatic conus medullaris tumors and believed silent underlying cauda equina lesion were our main objective.

Description of the surgical procedure

The patient was placed in the prone position and a median incision was performed from T12 to L2 levels. A laminectomy was completed before the dura's opening was done under the operating microscope. The conus medullaris looked very bulgy and tense, with a refined surface suggesting an underlying cystic cavity. Lower down; a nodular lesion was found on cauda equina spinal roots projecting between the L1 and L2 disc. First, the cystic lesion was opened, allowing access and identification of a small, typical reddish solid nodule that was adherent to both the conus and one of the spinal roots. Neuromonitoring was used to confirm negative motor and sphincter responses before the spinal root was sacrificed. Second, the cauda equina lesion was a little grayish, soft, and very adherent to two spinal roots. The context suggested HB, but the appearance was atypical. One of the roots was successfully separated from the lesion, while the other one was inlaid, could not be dissected, and had to be sacrificed after negative neuromonitoring responses. Complete resection of both lesions was obtained. Careful hemostasis was made before the dura was closed with non-absorbable sutures. The final closure was done in several layers.

Postoperative course

Complete pain relief was obtained immediately after the surgical procedure, with improvement of the urinary stream and other neurological symptoms. Before discharge, the patient had normalized her urinary function, but a slight hypoesthesia in the left perineum area persisted. She regained complete autonomy with normal walking. Immediate and 3 months postoperative magnetic resonance imaging (MRI) confirmed complete resection of both lesions without any complications [Figure 2].

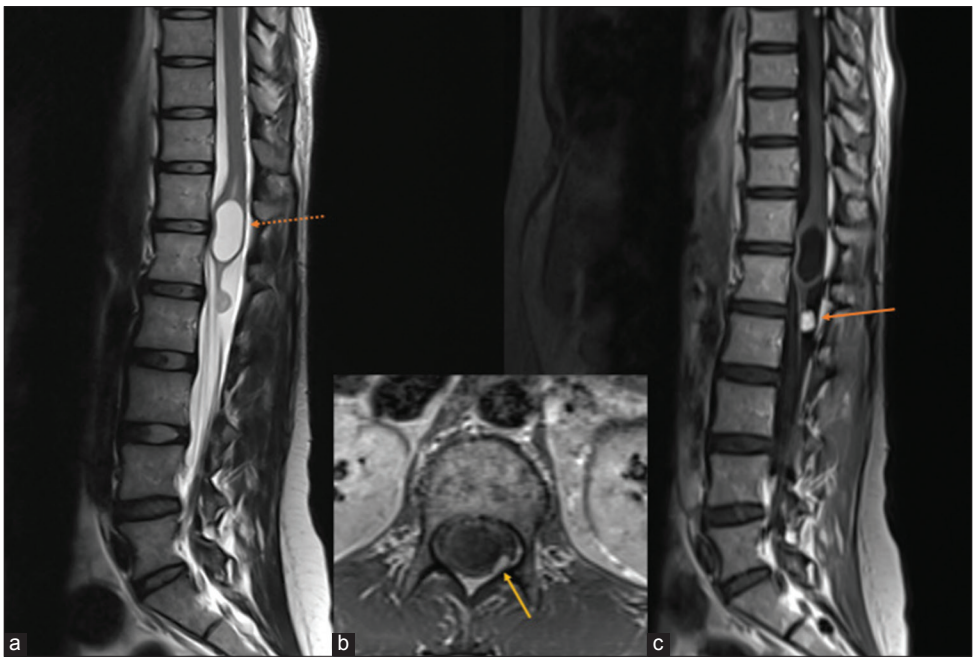

Figure 1: (a) The orange dotted arrow exhibits a cystic conus medullaris tumor on a sagittal T2 magnetic resonance imaging (MRI) between T12 and L1 with an underlying nodular lesion. (b) Axial T1 gadolinium MRI between T12 and L1 shows a small posterior left mural nodule around the cystic cavity, advocating for a possible spinal hemangioblastoma (yellow arrow). (c) Sagittal T1 gadolinium MRI shows a homogeneous contrast enhancement of the cauda equina tumor (orange arrow) between L1 and L2.

Laboratory results

Microscopic examination of hematoxylin and eosin of the sample of the conus medullaris tumor showed proliferation made of a double contingent of stromal cells with regularly rounded nuclei, clear micro vacuolated cytoplasm, and a vascular contingent made of a multitude of small capillary cavity [Figure 3a]. The immunohistochemical study confirmed the diagnosis of HB (the World Health Organization [WHO] grade 1) with positive immunoreactivity for PS100, epidermal growth factor receptor, alpha inhibin [Figure 3b], brachyury (cytoplasmic expression) toward the stromal component, and positivity of vessels toward CD34 [Figure 3c].

The cauda equina lesion specimen showed the proliferation of cells with ovoid nuclei and often bipolar brush-like extensions with frequent perivascular rosettes and a myxoid proliferation. Glial fibrillary acidic protein (GFAP) diffuse positivity confirmed diagnoses of an MPE (the WHO grade 2). Oligodendrocyte transcription factor (OLIG2) was negative. There was no dot on the Epithelila membrane antigen(EMA). CD34 marked some vascular structures [Figure 3d].

DISCUSSION

Intradural, intramedullary conus medullaris HBs are rare lesions, even in VHL patients.[10] A recent case report[2] advocated conus medullaris surgery, using intraoperative monitoring microsurgery technique, as safe and feasible for symptomatic HB, allowing significant neurological improvement. In this case, we performed surgery for a symptomatic conus medullaris HB and a believed silent cauda equina MPE, with substantial symptom improvement. Sphincter function disturbance was related, in our opinion, to the progressive growth of the conus medullaris lesion and was the determining factor in our surgical decision. The typical radiological appearance of this lesion, comprising a mural nodule and a cyst, suggested the highly probable diagnosis of HB. On the other hand, the intensely and homogeneously contrasting well-circumscribed lesion aspect of the cauda equina could not completely rule out the other usual diagnoses of this region. Moreover, this lesion remained stable over time and concerned only one or two spinal roots on the preoperative MRI; therefore, it was not believed to be responsible for the clinical signs.

Although rarely symptomatic, intradural extramedullary cauda equina HB is frequent in VHL[9], and acknowledgment of the numerous differential diagnoses is critical for patient selection. More importantly, the onset of MPE in VHL patients has not been described before.

Nervous system manifestations are in the foreground of VHL disease,[11] with retinal and cerebellar HB representing the most common first manifestations of VHL. They harbor significant associated morbidity and include CNS HBs, RH, endolymphatic sac tumors, and, less frequently, CNS metastases.[4] Although MPEs may develop in genetic

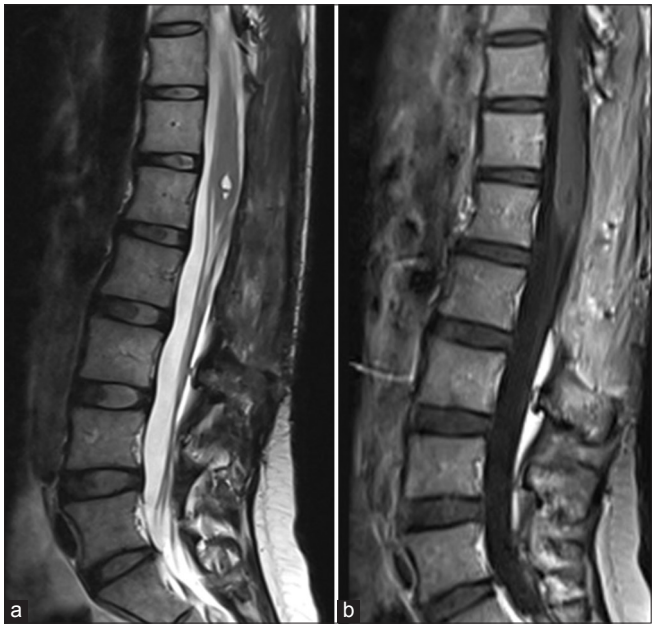

Figure 2: Postoperative magnetic resonance imaging (MRI) performed at 3 months after surgery demonstrating complete resection of both cauda equina and conus medullaris lesions. (a and b) Left image shows a sagittal T2 conus medullaris hypersignal without any signs of residual disease (no contrast enhancement) on T1 gadolinium imaging. MRI showed no signs of recurrent cauda equina myxopapillary ependymoma.

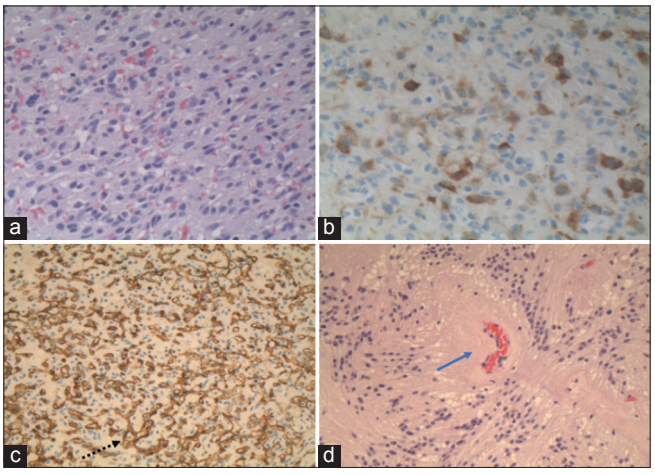

Figure 3: (a) Microscopic examination with hematoxylin and eosin (H&E) (H&E staining) shows multiple small capillary cavities of a hemangioblastoma (HB), (b) demonstrates the positive immunoreactivity of alpha inhibin in HB, (c) shows diffuse positive CD34 markings of numerous vessels (black dotted-arrow) from a World Health Organization grade 1 HB, and (d) under microscopic magnification 400 with H&E, tumor cells are arranged radially around a central vessel (blue arrow), a typical histological feature of myxopapillary ependymomas (i.e., perivascular pseudo rosettes).

disorders such as neurofibromatosis type 2,[5] they are typical sporadic benign tumors not related to VHL. A recent report[3] stated that HBs could mimic ependymoma due to morphological similarities. More interestingly these two entities have been suggested to harbor the same origin.[3] Typical HB shows numerous capillaries and stromal cells and despite molecular advances, the histogenic origin of HB is still unclear.[6,12] Here, we reported a case with two separate tumors, both radiologically and surgically, with a well-differentiated histology. The MPE was more likely to be an incidental tumor, while the HB was obviously related to VHL disease. However, a histological association may exist, although not yet understood, between ependymoma and HB, particularly in the case of VHL disease.[1,3,6]

CONCLUSION

We report the first case of a simultaneous cauda equina MPE and conus medullaris HB. MPE and HBs may occasionally show morphological similarities. Simultaneous spinal MPE and HBs may exceptionally arise in VHL patients, and neurosurgeons should know potential differential diagnosis of primary cauda equina tumors in case of VHL. Finally, VHL disease requires multidisciplinary management to improve the general prognosis and individual care for such a rare entity which is responsible of a significant disease burden and reduce life expectancy, particularly in women.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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