Case Report

Atypical presentation of extraspinal neurofibroma presenting with acute-onset monoparesis and Horner's syndrome: Case report and review of literature

ABSTRACT

The clinical presentation of spinal or extraspinal neurofibroma is radiculopathy or myelopathy, pain, and motor weakness. Extraspinal neurofibroma presenting with acute-onset monoparesis and Horner's syndrome is very rare. We report the case of a 55-year-old female who presented with acute-onset monoparesis of the left upper limb along with left-side drooping of the eyelid. Imaging revealed C6–D2 extraspinal solitary mass lesion lateral to spinous process with bleed without intraspinal component. The patient underwent an anterior cervical approach and excision of the tumor. Final biopsy report was a neurofibroma. At 3-year follow-up, she recovered from motor weakness, and Horner's syndrome subsided. Extraspinal neurofibroma can present with acute bleed, and surgical outcome is superior in early intervention.

Keywords: Extraspinal, Horner's syndrome, neurofibromatosis, outcome

INTRODUCTION

Neurofibromas are nerve sheath tumors composed of Schwann cells, fibroblasts, and supporting cells termed perineurial cells. They may occur as a sporadic solitary lesion or as a manifestation of neurofibromatosis 1 (NF1).[1] They commonly present as a painless and slow-growing mass. We report the first case of NF1 with extraspinal neurofibroma presenting with acute upper-limb monoparesis and Horner's syndrome.

CASE REPORT

A 55-year-old female presented with sudden-onset neck pain associated with left upper-limb weakness and drooping of the left eyelid. Examination revealed left-sided ptosis and an anisocoria that was greater in the dark. These findings were felt to be consistent with a left Horner's syndrome. Optic discs were healthy bilaterally. Cutaneous examination revealed multiple small neurofibromas and café-au-lait spots. Motor examination showed a flaccid weakness of the left upper limb (Medical Research Council [MRC] Grade: 1/5). The remainder of the systemic examination was unremarkable, and there were no physical signs to suggest a Pancoast tumor. Our patient's mother and brother also had multiple subcutaneous nodules.

Our differential was left apical Pancoast tumor or brachial plexopathy. There was no history of breathlessness, cough, or hemoptysis to suggest apical lung tumor. X-ray chest was normal. There was no history of preceding trauma or fever, which may be responsible for brachial plexopathy.

Magnetic resonance imaging (MRI) (T1-weighted [T1W]) showed a clearly demarcated tumor with bleed [Figure 1a],

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Access this article online	
Website: www.jcvjs.com	Quick Response Code
DOI: 10.4103/jcvjs.JCVJS_86_18	

How to cite this article: Nehete LS, Konar SK, Nandeesh BN, Shukla D, Devi BI. Atypical presentation of extraspinal neurofibroma presenting with acute-onset monoparesis and Horner's syndrome: Case report and review of literature. J Craniovert Jun Spine 2019;10:188-91.

lateral to the spinous process of C6-D2 vertebrae on the left side. MRI (T2W) images showed a lesion abutting the left subclavian artery [Figure 1b and c]. Computed tomography (CT) cervical spine showed scalloping of the transverse processes but no enlargement of neural foramina [Figure 1d]. Contrast study revealed peripheral enhancement and no intraspinal extension [Figure 1e and f]. After appropriate counseling and workup, the patient was offered anterior surgery. Anterior cervical approach and excision of the lesion were done. A transverse skin incision was made 1 cm above the clavicle. After subplatysmal dissection, sternocleidomastoid muscle and inferior omohyoid muscles were seen and retracted. The carotid sheath was identified and retracted laterally. The tumor was seen beneath the prevertebral fascia. Initially, internal decompression was performed, and while dissecting the tumor from the surrounding blood vessels at the inferior pole, the left subclavian artery was injured at the origin of the left vertebral artery. Bleeding was controlled with an angled vascular clip. After achieving hemostasis, the entire capsule was excised. Intraoperatively, the lesion was mildly vascular, firm, and grayish with areas of bleed. Detailed histological examination [Figure 2c] of the excised mass was suggestive of neurofibroma with large areas of fresh hemorrhage and fibrin deposition in a tumor. Postoperative angiogram [Figure 2a and b] showed good filling of the distal subclavian artery and partial filling of the left first (V1) and second (V2) segments of the vertebral artery. The distal vertebral artery was filled by the right vertebral artery. The patient made a good recovery from the operative procedure. At 3-year follow-up, she has complete recovery of motor power except for minimal shoulder abduction weakness (MRC: 4+/5), and Horner's syndrome subsided. After her spine surgery, the patient underwent an orthopedic surgery for long-bone fracture, and metal implants were placed. Henceforth, the MRI scan was not possible. She was evaluated with CT spine (plain + contrast). CT scan showed complete excision of the tumor and vascular clip *in situ* [Figure 3a and b].

DISCUSSION

Among the paraspinal neurofibromas, 72% were with intradural extramedullary localization, whereas 14% were with extradural, 13% were with both intraspinal and extraspinal components, and 1% was with intramedullary localizations.[2] A detailed history may reveal familial neurocutaneous syndromes that predispose the patient to tumor development. This patient had multiple subcutaneous neurofibromas, café-au-lait spots, and a family history of NF in first-degree relatives, satisfying the diagnostic criteria for NF.[1] The patient in the present study had presented with sudden-onset neck pain associated with left upper-limb weakness and drooping of the left eyelid. Upper-limb weakness can possibly be explained by the mass effect of a tumor on the ventral roots forming the brachial plexus.

The mechanism of Horner's syndrome in this patient could be due to the sudden increase in the size of the tumor due to bleed, causing mass effect on the ascending second-order neuron from the ciliospinal center of Budge–Waller to superior cervical ganglia. In the past, few cases[3-7] of Horner's syndrome in patients with NF have been reported, which are summarized in Table 1.

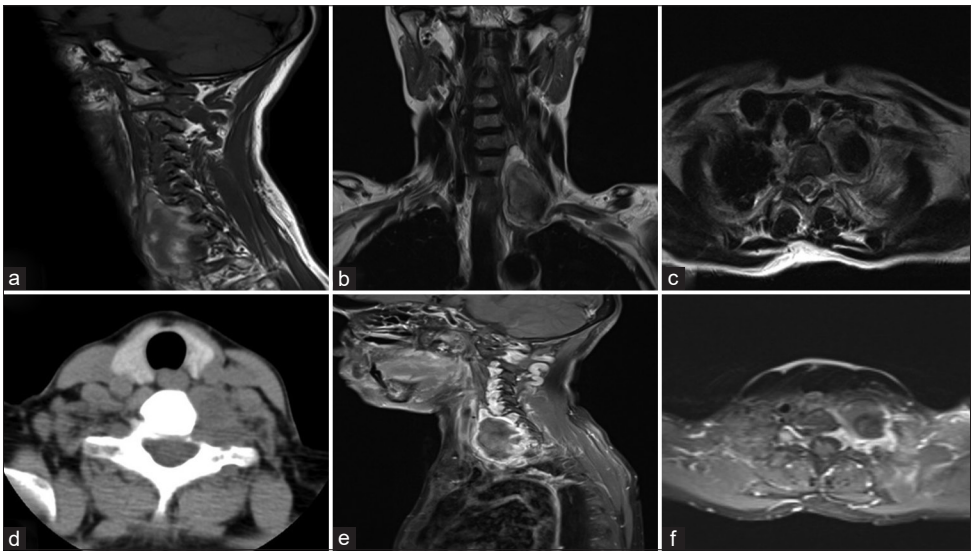

Figure 1: (a) Magnetic resonance imaging (T1 weighted, Sag) showing a solitary lesion adjacent to C6–D2 spinous process, periphery, and lower-part hyperintensity suggestive of bleed. (b) Magnetic resonance imaging (T2 weighted, Cor) showing superoinferior extent (C6–D2) and lower margin abutting the left subclavian artery. (c) Magnetic resonance imaging (T2-weighted, axial) showing extraspinal location with central–posterior hypointensity. (d) Computed tomography, axial, showing erosion of part of transverse process. (e) Magnetic resonance imaging (T1-weighted, contrast) showing peripheral enhancement with central nonenhancing part. (f) Magnetic resonance imaging (T1-weighted, contrast) showing no intraspinal extension

Table 1: Literature review and index case of neurofibromatosis presented with Horner's syndrome

Age/sex	Title	Author, journal	Clinical presentation	Imaging	Histology	Treatment
31/female	NF Type 1 presenting with Horner's syndrome	Cackett *et al.*,[2] Eye 2005	Two-month history of a drooping left eyelid	Nonenhancing dumbbell-shaped mass extending from the root of the neck anterior to the first rib to approximately 2 cm above the level of the aortic arch	MPNST	Excision
31/male	Horner's syndrome in NF Type 1	Lee *et al.*,[4] J Craniofac Surg 2015	Drooping left eyelid	Well-defined left posterior mediastinal mass close to vertebral bodies of upper dorsal spine at the level of T1-T5	Neurofibroma	Excision
23/male	MPNST presenting with Horner's syndrome	Basuthakur *et al.*,[5] J Assoc Phys India 2013	Dull-aching pain in the right shoulder and anterior chest wall for 1 year which radiated to the inner aspect of the arm and forearm. He also complained weakness of the right hand, loss of sweating of right side of the face, and shrunken right eye for 1 year	Well-defined, irregular, enhancing mass in the right upper lobe	MPNST	CT-guided biopsy
24/female	Neurofibroma of the cervical sympathetic chain presenting with Horner's syndrome	Ruckley and Blair,[6] J Laryngol Otol 1986	Right neck swelling, right Horner's syndrome	6-4 cm hard mass fixed to larynx and right lobe of thyroid	Neurofibroma	Excision
6/female	A case of segmental NF presented with Horner's syndrome	Oguz *et al.*,[7] Turk Pediatri Ars 1993	Horner's syndrome, neurofibroma involving brachial plexus	NA	Neurofibroma	NA
55/female	NF1 with extraspinal neurofibroma presented with Horner's syndrome	Present case	Sudden-onset neck pain associated with left upper limb weakness and drooping of the left eyelid	Solitary oval-shaped mass lesion lateral to spinous process of C6-D2 vertebrae on the left side	Neurofibroma	Excision

NA - Not available; MPNSTs - Malignant peripheral nerve sheath tumors; NF - Neurofibromatosis; CT - Computed tomography

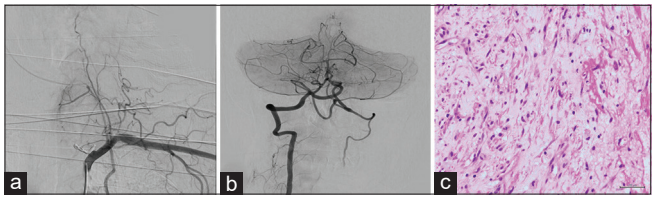

Figure 2: (a) Digital subtraction angiography subclavian injection showing distal filling. (b) Digital subtraction angiography right vertebral injection showing proximal filling of the left vertebral artery. (c) Histopathological examination: (H and E, ×200): microphotograph showing a loose hypocellular spindle cell neoplasm with wavy nuclei and myxoid change. The neoplasm is highly vascular with fibrin deposits. The features are suggestive of neurofibroma

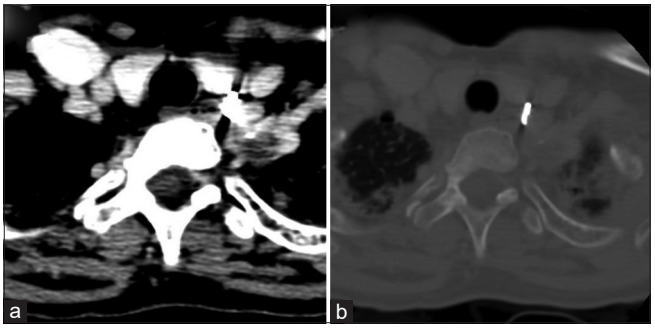

Figure 3: (a) Computed tomography spine (axial, contrast) showing complete excision of the tumor. (b) Computed tomography spine (axial, bone window) showing vascular clip *in situ*

A hemorrhagic complication in neurofibroma or schwannoma is uncommon, which may be seen either at operation or following trauma.[8] Acute hemorrhage following minor trauma may occur due to the increased vascular fragility of the dysplastic vessels associated with the neurofibroma[9] or vascular invasion by the tumor.[10] Furthermore, Schwann cells have angiogenic potential which may contribute to the hypervascularity of tumor. Various factors such as angiogenin, transforming growth factors α and β, epidermal growth factor, fibroblast growth factors, and acidic and basic fibroblast growth factors have been described for angiogenesis.[11]

The literature review showed extraspinal neurofibroma presenting as acute-onset monoparesis, and Horner's syndrome is not reported yet. In the past, acute-onset paraplegia or quadriplegia due to bleed in the schwannoma has been reported.[12,13]

CONCLUSION

An extraspinal neurofibroma presenting with sudden-onset monoparesis and Horner's syndrome is very rare. Acute hemorrhage within the tumor may probably be the reason for acute presentation. Prompt evaluation and surgery is the goal in this rare type of cases.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published, and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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