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Case Report and Brief Review

Adult Spinal Hamartoma Involving Conus Medullaris: Brief Review About Associated Congenital Abnormalities and Surgical Outcome.

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ABSTRACT

Background: Spinal hamartoma is an extremely rare lesion for symptomatic spinal cord compression in adult population. Without any association of spinal dysraphism and neurofibromatosis, only 3 cases have been reported in the literature.

Case description: A 40-year-old man presented with lumbago femorica for 6 months. Gradually the symptoms worsened and he developed features of Cauda equina syndrome. Magnetic Resonance Imaging demonstrated a heterogeneously hyperintense intradural extramedullary mass in both T1 and T2WI, involving conus medullaris and cauda equina, opposite to L1. Patient underwent near total resection of lesion through posterior midline approach. Histopathological features were consistent with hamartoma. Patient neurologically improved and follow up MRI at 6 months showed static small nodular tumor residue.

Conclusion: To the best of our knowledge, current report is the fourth case of spinal hamartoma in adult male, without any association of spinal dysraphism and neurofibromatosis. This may be the first reported case from Bangladesh.

Keywords: hamartoma, neurofibromatosis, spinal dysraphism.

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* Main subject and any subcategories have been classified according to the research topic.
INTRODUCTION

Hamartomas are slow growing benign lesion consist of disorganized mature cells resulting from aberrant differentiation, native to that particular site [1]. During development, premature dysjunction of neural tube from cutaneous ectoderm results in incorporation and differentiation of mesenchyme into the developing neural tube giving rise to spinal hamartoma [2]. They are frequently associated with neurofibromatosis and spinal dysraphism [3-6]. Clinical presentation depends upon the location and extent of spinal cord and / or, nerve root compression [1, 3-10]. MRI is the diagnostic test of choice to evaluate the relationship of the lesion with surrounding neurovascular structures and for operative planning [1,7,11]. Early detection and timely intervention results in favourable outcome [5, 6, 10].

In this paper, we report a case of adult spinal hamartoma of conus medullaris without any associated pathologies. Patient underwent near total resection with significant improvement of neurological functions at 6 months follow up. Beside this, we review the previously reported cases of adult spinal cord hamartoma providing special emphasis on embryogenesis, associated congenital anomalies and surgical outcome.

CASE REPORT

History and clinical examination: A 40-year-old man presented with lumbago with bilateral femorica for previous 6 months. For last 7 days, he developed weakness of both lower limbs with retention of urine for which he underwent catheterization in a local hospital and referred to our facility for further evaluation and management.

A neurological examination revealed decreased muscle power in both lower extremities [left, 4/5; right, 4/5]; diminished knee jerk bilaterally; saddle anaesthesia and straight leg raising test 45 degree bilaterally.

Systemic examination revealed no abnormalities and there was no cutaneous or ocular stigmata for neurocutaneous disorders and occult spinal dysraphism. After clinical evaluation, he diagnosed as a case of cauda equina syndrome.
Surgery: The patient underwent microscope-assisted total laminectomy at the L1, partial laminectomy of D12 after subperiosteal dissection of muscles and soft tissue upto bilateral facet joints. After durotomy, there was yellowish white tumor identified at the level of L1, adhered with the conus medullaris and cauda equina causing significant compression of adjacent neural structures. The resected tumor was capsulated, moderately vascular, and soft to firm in consistency. Near total resection of the lesion with adequate decompression of the cauda equina was achieved.

Histopathology: Microscopic examination showed that the specimen contained mature fatty tissue intermixed with disoriented axons and glial process. No mitotic activity was seen. Features of the lesion was malformative, rather than neoplastic, being consistent with hamartoma [Figure 4].

Post-operative course:
One day after surgery, the patient noticed improvement of lower extremity pain. After one month, all of the neurological deficit improved significantly. Catheter was removed and patient required occasional self intermittent catheterization. We referred him to neurourologist for further management. There was a small nodular tumor residue observed at 6 months follow up MRI.

Figure [3]: After durotomy, there was yellowish white tumor identified, adhered with the nerve roots of Cauda equina [A]; gross appearance of the resected specimen [B].

Figure [4]: Histopathology slide showing intradural lesion composed of adipose tissue, mature cartilage, skeletal muscle, blood vessels and nerve bundles [hematoxylin and eosin, original magnification,100x].

Figure [5]: Follow up MRI-T2WI mid sagittal section, after six months showing small residue of tumor with encysted fluid collection at the operative area.
DISCUSSION

Hamartomas are the matured specialized cells, resulting from anomalous differentiation of stem cells or tissues native to that particular site. They are benign lesion in which tissues are arranged in disorganized fashion [1]. Though relatively uncommon in the brain, spinal hamartoma is an extremely rare lesion contributes to symptomatic spinal cord compression in adult population and to the best of our knowledge, only ten cases have been reported in the literature, including the present one [Table 1] and this is the first reported case from Bangladesh.

Associated anomalies

Spinal hamartomas are frequently associated with Neurofibromatosis and Spinal dysraphism [3-6,12], specially in paediatric population. Their cutaneous and ocular stigmata helps in early evaluation of this rare entity [7,11,12]. However, association with Klippel feil syndrome, spinal giant cell angioblastomas, Proteus syndrome, pseudo-pancreatic cyst have also been reported [5,8,13].

During review of the previously reported literature, only three documented cases were found having no features of such association [1,9,10]. Beside this, there is no genetic predisposition found for this hamartomatous lesion [14].

Considering their association, Castillo et al. [7] classified spinal hamartomas into two categories: one is “vascular and neuroglial hamartoma,” associated with neurofibromatosis and the other as “midline spinal hamartoma” which is sometimes associated with spinal dysraphism.

Developmental background

The exact pathogenesis of spinal hamartoma is still a matter of debate. However, it is proposed that, in the embryonic stage, premature dysjunction of developing neural tube from cutaneous ectoderm induces surrounding mesenchyme enters between the neural tube and overlying cutaneous ectoderm [Figure 1A, B].

Later on, this aberrant mesenchyme differentiates to form other soft tissues, such as adipose and muscle tissues [1,2], nerve tissue [3], cartilage, bone, abnormal vessels, glands, synovial membranes, lymphoid tissues, and urinary tract tissue [8].

Considering the tissue architecture, their distinction from teratoma is difficult. However, mature teratoma is believed to have the elements from all three germ layers whereas hamartomas don’t have. Beside this, teratoma carries the risk of malignant transformation [1].

<table>
<thead>
<tr>
<th>S/N</th>
<th>Author</th>
<th>Year</th>
<th>Age</th>
<th>Sex</th>
<th>Site</th>
<th>Presentation</th>
<th>Associated anomalies</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>01</td>
<td>Brownlee et al.</td>
<td>1998</td>
<td>52</td>
<td>F</td>
<td></td>
<td>D12, IM</td>
<td>Conus medullaris syndrome</td>
<td>Neurofibromatosis type I</td>
</tr>
<tr>
<td>02</td>
<td>Riley et al.</td>
<td>1999</td>
<td>26</td>
<td>M</td>
<td>C2,</td>
<td>Neck pain, weakness of right upper extremity</td>
<td>None</td>
<td>Biopsy and detethering of the spinal cord. Develop chemical meningitis and hydrocephalus after 2.5 weeks post-op and treated accordingly.</td>
</tr>
<tr>
<td>04</td>
<td>Perrini et al.</td>
<td>2005</td>
<td>20</td>
<td>M</td>
<td>C4,</td>
<td>Spastic quadriplegia</td>
<td>Klippel-Feil Syndrome, spinal dysraphism</td>
<td>Maximum safe surgical resection, detethering of spinal cord. Tetraparesis improved, bladder function remain unchanged. Static residue at 6 months F/U MRI.</td>
</tr>
<tr>
<td>05</td>
<td>Lin et al.</td>
<td>2011</td>
<td>73</td>
<td>F</td>
<td>L3, EM</td>
<td>Cauda equina syndrome</td>
<td>None</td>
<td>GTR with vertebroplasty. Resolution of symptoms after 1 month.</td>
</tr>
<tr>
<td>06</td>
<td>Rao et al.</td>
<td>2013</td>
<td>55</td>
<td>F</td>
<td>D12-L2</td>
<td>Weakness of left leg</td>
<td>Hyperpigmentation, non healing ulcer</td>
<td>Excision of the cyst. F/U not mentioned.</td>
</tr>
<tr>
<td>07</td>
<td>Shindo et al.</td>
<td>2015</td>
<td>75</td>
<td>M</td>
<td>D3,IM</td>
<td>Chest pain</td>
<td>None</td>
<td>Maximum safe surgical resection. Static at F/U MRI after 2 years.</td>
</tr>
<tr>
<td>09</td>
<td>Present case</td>
<td>2020</td>
<td>40</td>
<td>M</td>
<td>L1, EM</td>
<td>Cauda equina syndrome</td>
<td>None</td>
<td>Maximum safe surgical resection. Static at F/U MRI after 6 months.</td>
</tr>
</tbody>
</table>

Histopathology

Histologically, spinal hamartoma composed of adipose and muscle tissues [1, 2], nerve tissue [3], cartilage, bone [4], abnormal vessels, glands, synovial membranes, lymphoid, and urinary tract tissue [5].

There is no detectable mitotic activity and Ki 67 should provide negative results [3]. Our patient presented with matured adipose tissue with disorganized axonal profile and glial process. We therefore believe that this case shows the characteristics of pure hamartoma.

Clinical presentation

Spinal hamartoma may be more likely to pass unnoticed until adulthood if the clinical condition of the patient is intact and no associated lesions. The age of patients with adult spinal hamartoma ranges from 20 years to 75 years [Table 1].

They are located in the cervical, thoracic, and lumbar regions or at junctional zone with slight male dominance [male: female = 5:4]. Clinical presentation depends upon the location and extent of compression of spinal cord and/or, nerve roots. Back pain [4,9], chest pain [10], muscle wasting [4,5,8,9], conus medullaris syndrome [3], cauda equine syndrome [1,6] have been reported in the literature. Some-times, tethering of the spinal cord and/or, filum terminale present as painful scoliosis [6,15].

Neuroimaging

On MRI, midline spinal hamartoma is characterized by a well-defined, exophytic mass, arising from the dorsal surface of the spinal cord with signal intensity similar to that of normal cord tissue on both T1- and T2-weighted imaging, with no contrast enhancement [7,9]. However, presence of adipose tissue, muscles and other structures give rise to heterogenous signal intensity, as in our reported case. Sometimes, teratoma mimics the neuro-imaging features which shows gadolinium enhancement whereas hamartoma is non enhancing lesion [10]. But their definitive diagnosis is only possible after histopathology.

Surgery and outcome

The indication for surgery in a patient with a spinal hamartoma is the presence of spinal cord and/or, root compression, giving rise to neurological deficit. Due to their developmental background, their infiltrating nature often precludes gross total resection. So, maximum safe surgical resection with preservation of the existing neurological functions should be the aim of surgery.

Based upon the previous literature, total tumor resection [6,7], subtotal tumor resection [3,7,11,16,17], or only biopsy [9] in questionable cases have been reported. In addition, detethering of the spinal cord, excision of the derma sinus and fistula should be done [5,6,9].

In our case, due to adherence of the lesion with multiple nerve roots and conus medullaris, we went for near total resection to preserve the neurological function. Although it’s a slow growing benign lesion, near total or subtotal resection always carry the risk of recurrence. Follow up MRI after variable periods demonstrated static tumor residue [3,5,10]. However, Mittler et al. [16] reported a paediatric spinal hamartoma in 4-year-old child, which recurred several times and 17 years after the initial diagnosis as hamartoma, transformed into glioblastoma multiforme. Most of the patients were reported to improve their neurological function significantly after the operation. However, Riley et al. [9] observed chemical meningitis, 2 weeks after the initial surgery. Patient treated conservatively and later on, underwent shunt surgery due to development of hydrocephalus.

Conclusion:

Adult spinal cord hamartoma is extremely rare etiology for symptomatic cord compression. Their genetic analysis and proposed pathogenesis can't explain their existence without any association of spinal dysraphism or neurofibromatosis. Without any cutaneous or ocular stigmata, their definitive diagnosis often delayed. However, early diagnosis and timely intervention is mandatory for good neurological outcome.
Declarations:

Authors' contributions:

- Conception, diagnosis and design: Dr. Nazmin Ahmed, Dr. Bipin Chaurasia, Vishal Chavda, Giuseppe Umana, Gianluca Scalia, Ottavo Tomasi
- Manuscript preparation: Dr. Nazmin Ahmed
- Radiology diagnosis: Dr. Shahidul Islam Khan
- Literature search: Dr. Avijit Dey
- Technical revision: Dr. Bipin Chaurasia
- Manuscript editing: Dr. KM Tarikul Islam
- Final approval of manuscript: Dr. Shahidul Islam Khan

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Ethics approval: There is no ethics issue in this paper.

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