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Case Reprot 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 Resonence Imagimg 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.

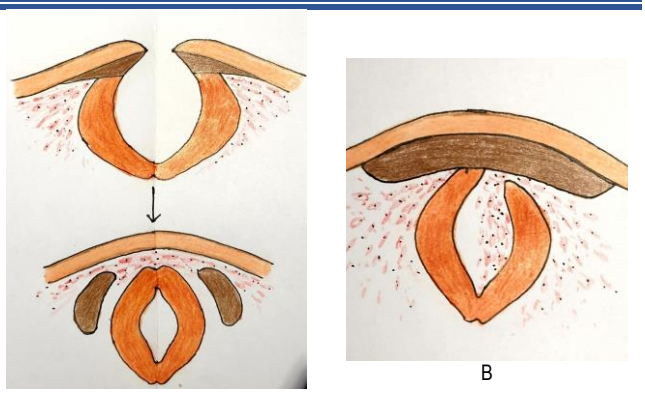

Figure [1]: Schematic picture demonstrates normal neurulation process [A]; premature disjunction of neuroectoderm results in incorporation of surrounding mesenchyme to the inner surface of neural tube which differentiates into fatty issue, along with other connective tissue giving rise to spinal hamartoma [B] [Here, light brown-cutaneous ectoderm, dark brown-neural crest, orange-neural tube, red-mesenchyme].

Neuroimaging: Spinal magnetic resonance imaging revealed a heterogeneously hyperintense intradural extramedullary lesion in both T1WI & T2WI, approximately 3x 2 cm in size at the L1 level [Figure 2A, B]. Based on the findings, the differential diagnosis should include a focal subacute hematoma and fat containing neoplasm, such as hamartoma or teratoma. Plain x-ray of the dorsolumbar spine revealed no abnormalities of vertebral body and arch.

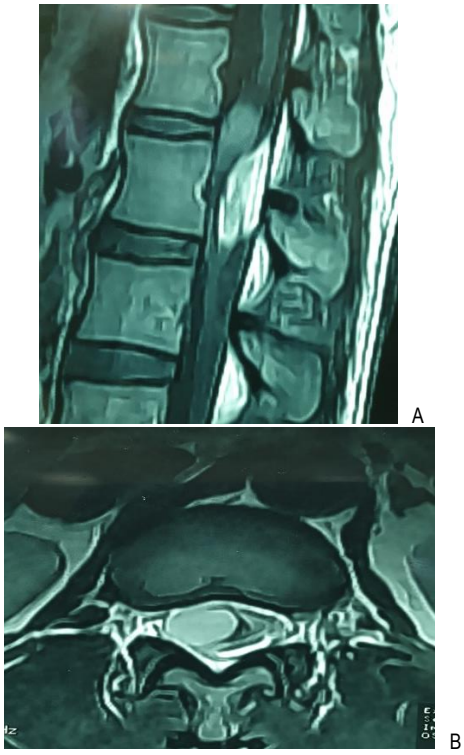

Figure [2]: Midsagittal T1WI [A] and axial T2WI demonstrates a heterogenously hyperintense lesion opposite to the L1, compressing the conus medullaris and cauda equina significantly. There is hypertrophy of the facet joints and ligamentum flavum.

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.

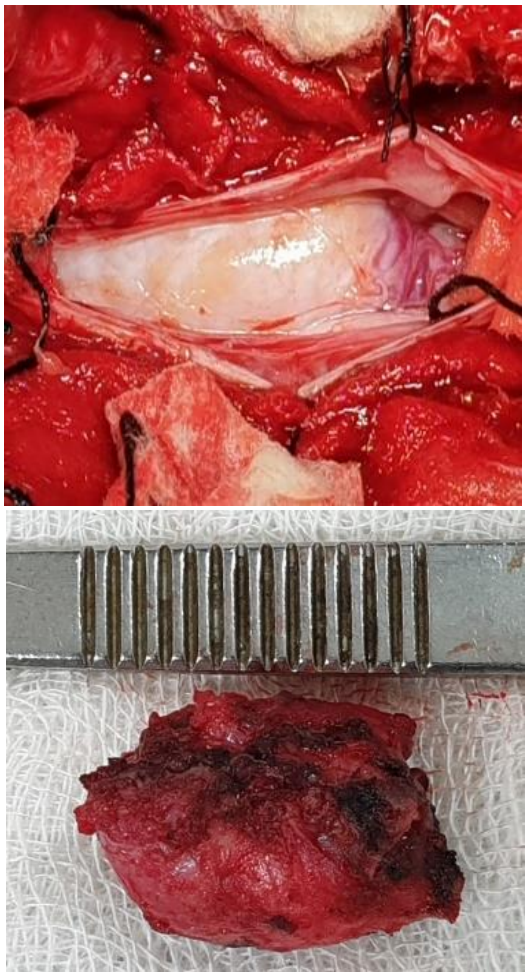

Figure [3]: After durotomy, there was yellowish white tumor identified, adhered with the nerve roots of Cauda equina [A]; gross appearance of the resected specimen [B].

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]**.

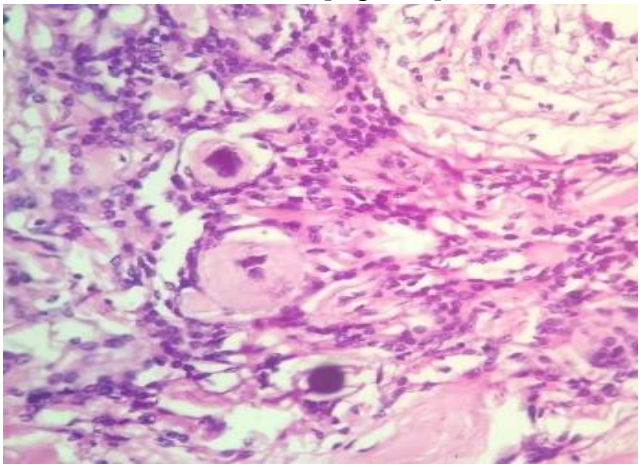

Figure [4]: Histopathology slide showing intradural lesion composed of adipose tissue, mature cartilage, skeletal muscle, blood vessels and nerve bundles [hematoxylin and eosin, original magnification,100A].

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 ocassional 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.

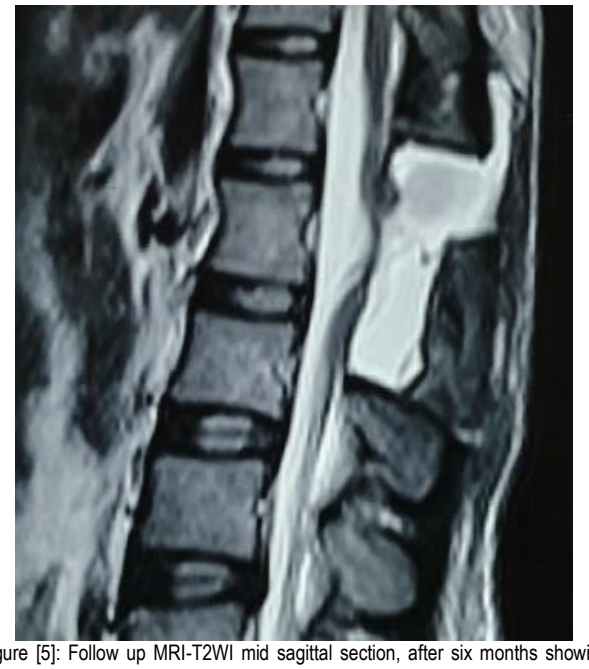

Figure [5]: Follow up MRI-T2WI mid sagittal section, after six months showing small residue of tumor with encysted fluid collection at the operative area.

Table [1]: Reported cases of spinal hamartoma in adult population till date [1, 3- 6, 8-10]

S/N	Author	Year	Age	Sex	Site	Presentation	Associated anomalies	Management
01.	Brownlee *et al*.	1998	52	F	D12, IM	Conus medullaris syndrome	Neurofibromatosis type I	Maximum safe surgical resection. Static residual mass at 2 years F/U MRI.
02.	Riley *et al*.	1999	26	M	C2, EM	Neck pain, weakness of right upper extremity	None	Biopsy and detethering of the spinal cord. Develop chemical meningitis and hydrocephalus after 2.5 weeks post-op and treated accordingly.
03.	Sen *et al*.	2005	62	F	L4-L5, EM	Cauda equina syndrome	Spinal dysraphism, epidermoid cyst, dermal sinus	Maximum safe surgical resection with fistulectomy. Complete resolution of symptoms at F/U examination.
04.	Perrini *et al*.	2005	20	M	C4-5,	Spastic quadriparesis	Klippel Feil Syndrome, spinal dysraphism	Maximum safe surgical resection, detethering of spinal cord. Tetraparesis improved, bladder function remain unchanged. Static residue at 6 months F/U MRI.
05.	Lin *et al*.	2011	73	F	L3, EM	Cauda equina syndrome	None	GTR with vertebroplasty, Resolution of symptoms after 1 month.
06.	Rao *et al*	2013	55	F	D12-L2	Weakness of left leg	Hyperpigmentation, non healing ulcer	Excision of the cyst. F/U not mentioned.
07.	Shindo *et al*	2015	75	M	D3,IM	Chest pain	None	Maximum safe surgical resection. Static at F/U MRI after 2 years.
08	Krishnan & Saha	2016	23	M	L3-4, EM	Back pain, muscle wasting of right leg	Spinal dysraphism	Maximum safe surgical resection. Static at F/U MRI after 1 year.
09.	Present case	2020	40	M	L1, EM	Cauda equina syndrome	None	Maximum safe surgical resection. Static at F/U MRI after 6 months.

M: male, F: female, C: cervical, D: dorsal, L: lumbar, EM: extramedullary, IM: intramedullary, GTR: gross total resection, F/U: follow up

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], specialy 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 dintinction 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].

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 [8].

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*. [18] 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, Ottavio 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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Patient consent: An informed written consent was obtained from the patient.

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

REFERENCES

1. Lin HC, Yu CP, Liu MY, Lee HS. A spinal hamartoma in an aged woman. Kaohsiung J Med Sci. 2011 Jul;27[7]:295-8. doi: 10.1016/j.kjms.2010.09.007.

2. Barkovich AJ. Pediatric neuroradiology, fourth edition. Philadelphia, PA: Lippincott Williams & Wilkins, 2005: 801 – 868

3. Brownlee RD, Clark AW, Sevick RJ, Myles ST. Symptomatic hamartoma of the spinal cord associated with neurofibromatosis type 1. Case report. J Neurosurg. 1998; 88 [6]: 1099-103. doi: 10.3171/jns.1998.88. 6.1099.

4. Krishnan P, Saha M. Osseous dysraphic hamartoma - A completely formed, aberrantly located, supralaminar rib in a lumbar lipomyelocele. Neurol India. 2016 Jan-Feb; 64[1]: 182-4. doi: 10.4103/0028-3886.173626.

5. Perrini P, Scollato A, Guidi E, Benedetto N, Buccoliero AM, Di Lorenzo N. Tethered cervical spinal cord due to a hamartomatous stalk in a young adult. Case report. J Neurosurg. 2005 Mar;102[2 Suppl]:244-7. doi: 10.3171/jns. 2005.102.2.0244.

6. Sen O, Kayaselcuk F, Yalcin O, Erdogan B, Aydin MV, Zorludemir S, Altinors N. Lumbar meningeal hamartoma and epidermoid cyst associated with spinal dysraphism in an elderly patient. Neurosurg Rev. 2005 Apr;28 [2]: 159-62. doi: 10.1007/s10143-004-0351-8.

7. Castillo M, Smith MM, Armao D. Midline spinal cord hamartomas: MR imaging features of two patients. AJNR Am J Neuroradiol. 1999 Jun-Jul; 20 [6]:1169-71. PMID: 10445466.

8. Rao DN, Chakravarthy VK, Aruna E, Ratnam GV, Rao DR. Midline Spinal Cord Hamartomas—A Report of Six Cases. Int J Clin Med. 2013 Jun 4;4[06]:291. DOI: 10.4236/ijcm.2013.46051

9. Riley K, Palmer CA, Oser AB, Paramore CG. Spinal cord hamartoma: case report. Neurosurgery. 1999 May; 44 [5]: 1125-7; discussion 1127-8. doi: 10.1097/00006123-199905000-00109.

10. Shindo D, Shimono T, Takami T, Tanaka S, Tsukamoto T, Miki Y. Spinal hamartoma in an elderly man. Jpn J Radiol. 2015 Nov;33[11]:706-9. doi: 10.1007/s11604-015-0475-5.

11. Bining HJ, Saigal G, Goswami RS, Gill D, McNeely PD. Spinal hamartoma in a neonate: Unusual presentation and MR imaging findings. AJNR Am J Neuroradiol. 2006 Apr;27[4]:810-2. PMID: 16611769.

12. Malelak EB, Lauren C, Argie D, Nugraheni T. Congenital Midline Spinal Hamartoma in a 5-Month-Old Infant. World Neurosurg. 2021 Jan;145:142-147. doi: 10.1016/ j.wneu.2020.09.065.

13. Gupta S, Kumar A, Gangopadhyay AN, Kumar M. Spinal cord hamartoma with pseudopancreatic cyst. Indian J Pediatr. 2008 Mar;75[3]:281-3. doi: 10.1007/s12098-008-0060-3.

14. Bartels RHMA, Grotenhuis JA, Stegmann APA, Brunner H. Genetic analysis of spinal dysraphism with a hamartomatous growth [appendix] of the spinal cord: a case series. BMC Neurol. 2020 Apr 6;20[1]:121. doi: 10.1186/s12883-020-01710-7.

15. Davidson L, Gonzalez-Gomez I, McComb JG. Thoracic spinal cord tethering resulting from an exophytic hamartoma. Pediatr Neurosurg. 2009; 45[3]:244-6. doi: 10.1159/ 000224623.

16. Abel TJ, Chowdhary A, Jallo G, Wang PP, Burger P, Avellino AM. Thoracic spinal cord compression by intramedullary hamartomatous tissue in a young boy: case report. Neurosurgery. 2008 Jun;62[6]:E1380-1; discussion E1381. doi: 10.1227/01.neu.0000333314.40920.5e.

17. Yano S, Hida K, Nagashima K, Iwasaki Y. Spinal fibrous hamartoma of infancy: case report. Neurosurgery. 2004 Sep; 55 [3]: 712. doi: 10.1227/01.neu.0000134614. 73236.a4.

18. Mittler MA, Walters BC, Fried AH, Sotomayor EA, Stopa EG. Malignant glial tumor arising from the site of a previous hamartoma/ganglioglioma: coincidence or malignant transformation? Pediatr Neurosurg. 1999 Mar;30[3]:132-4. doi: 10.1159/000028780.

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