Case Report

Infantile Presentation of Lehman Syndrome with Multiple Lateral Meningoceles, Dural Ectasias, and Herniation of Conus: A Rare Case Report

Mohan Amuthabarathi, Kramadhari Harshith, Krishnan Nagarajan

Department of Radiodiagnosis, Jawaharlal Institute of Postgraduate Medical Education & Research (JIPMER), Pondicherry, India

Lehman syndrome, or lateral meningocele syndrome, is a rare disorder of skeletal malformation, characterized by facial dysmorphism and multiple lateral meningoceles. We present a case of a 5-month-old girl who presented with macrocephaly, developmental delay, and failure to thrive. A whole spine magnetic resonance imaging was carried out, which showed multiple bilateral well-defined cystic masses within the neural foramina involving the entire spine, predominantly the thoracolumbar regions, with neural foraminal widening and dural ectasia suggestive of multiple lateral meningoceles.

KEYWORDS: Dural ectasia, lateral meningocele, Lehman syndrome, magnetic resonance imaging

INTRODUCTION

Lehman syndrome, or lateral meningocele syndrome (LMS), first described in 1977 by Lehman et al., is a rare disorder of which only a few cases have been described in the literature. The syndrome is characterized by multiple lateral meningoceles, abnormal facies, and skeletal abnormalities. The meningoceles are commonly located and largest in the thoracolumbar region. This can damage the spinal nerve roots leading to neurogenic bladder, paresthesia, and weakness of the lower limbs. The motor milestones are delayed but intelligence is usually normal. Typical facial features are also characteristic. In this report, we discuss a case of a 5-month-old girl who had bilateral multiple-level lateral meningocele without any associated neurofibromatosis, or Marfan's syndrome.

CASE REPORT

A 5-month-old girl presented with complaints of failure to thrive, developmental delay, and macrocephaly. The patient was a first-born child and born to a second-degree consanguineous marriage with uneventful antenatal history. Physical examination findings showed increased head circumference, epicanthic folds, and micrognathia. No features of either Marfan's syndrome or neurofibromatosis type 1 (NF1) were noted. Neurosonography of child's cranium and computed tomography (CT) brain showed ventriculomegaly with prominent cerebrospinal fluid (CSF) spaces. The patient was referred for Ultrasonography (USG) of abdomen for ruling out other anomalies. Screening USG spine showed bilateral symmetric elongated cystic outpouchings in the entire paraspinal regions, which were communicating with the spinal canal [Figure 1]. A solid hyperechoic structure was seen herniating into the one of the sac at the right lumbar region, which was found to be spinal cord. The patient underwent magnetic resonance imaging (MRI) for further evaluation. Whole spine MRI showed multiple lateral CSF-intensity protrusions lined by meninges through the neural foraminal expansions involving the entire spine, which were communicating with the spinal canal, suggesting meningoceles [Figure 2]. Dural

Address for correspondence: Dr. K Nagarajan, Department of Radio-Diagnosis, Jawaharlal Institute of Postgraduate Medical Education & Research (JIPMER), Pondicherry 605006, India. E-mail: lknagarajan1@gmail.com

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: WKLHRPMedknow_reprints@wolterskluwer.com

How to cite this article: Amuthabarathi M, Harshith K, Nagarajan K. Infantile presentation of Lehman syndrome with multiple lateral meningoceles, dural ectasias, and herniation of conus: A rare case report. J Pediatr Neurosci 2020;15:111-5.
ectasia was noted predominantly posterior to the cord in thoracolumbar region. Herniation of the conus was noted into meningeal sac through the dilated right neural foramen at L2–L3 levels [Figure 3]. Scalloping of the posterior body of thoracic and lumbar vertebrae were also noted. MRI of the brain showed cerebellar tonsillar herniation with crowding at the level of the foramen magnum causing hydrocephalus. The corpus callosum was thinned because of the ballooned third ventricle [Figures 3 and 4].

Figure 1: (A and B) USG images in sagittal plane showing lateral meningoceles and dural ectasias. (C and D) USG image in parasagittal plane showing herniated conus in lumbar neural foramen

Figure 2: (A and B) Sagittal T2-weighted MR images of spine showing dural ectasia in thoracolumbar region and scalloping of the vertebral bodies. (C) STIR coronal image showing multiple lateral meningoceles in thoracolumbar region. (D) T2-weighted axial sections showing lateral meningoceles with enlarged neural foramen
**DISCUSSION**

Reduced meningeal elasticity within the thecal sac presents with isolated or multiple dilatations of the CSF space. This manifests with formation of cysts and ectasias at variable levels of the central nervous system with a series of neuroanatomical features including dural ectasias, Tarlov cysts, arachnoid cysts, and lateral meningoceles. Dural ectasia refers to congenital ballooning or widening of the dural sac associated with herniation of nerve root sleeves.\(^\text{[2]}\) Lateral or anterior spinal meningocele are relatively rare congenital anomalies where protrusion of dura mater and arachnoid extending laterally through an enlarged intervertebral foramen into the paraspinal, intrathoracic, or retroperitoneal region.\(^\text{[3]}\) Lateral meningocele may be unilateral or bilateral and may be solitary or multiple.

LMS or Lehman syndrome is characterized by multiple lateral meningoceles in the absence of neurofibromatosis or Marfan's syndrome.\(^\text{[4]}\) Presentation is commonly during the fourth and the fifth decades of life\(^\text{[3]}\) and overall, female patients are more commonly affected. Lateral meningoceles commonly occur in thoracic and lumbar regions followed by the cervical area,\(^\text{[6-8]}\) and rarely in the sacral region.\(^\text{[5,9,10]}\)

LMS has been previously reported in 11 patients, five females, and six males, belonging to 9 families.\(^\text{[11]}\) LMS has been observed in two instances of vertical transmission, and seven sporadic cases with age at diagnosis ranging from 25 months to 33 years.\(^\text{[2]}\) The genetic basis of LMS is still unknown. Familial recurrence was seen in two instances with transmission from an affected mother to her daughter.\(^\text{[3]}\)

Heterozygous mutations in the last exon of the \textit{NOTCH3} gene were recently shown to be associated with Lehman syndrome in six individuals. \textit{NOTCH3} is a part of an evolutionarily conserved family of cell surface receptors that regulate gene expression and subsequently cell growth and proliferation.\(^\text{[12]}\) The available data support the hypothesis that the majority of LMS arise from \textit{de novo} mutations.\(^\text{[11]}\) The molecular cause of LMS may affect the various connective tissue components, such as collagen XII or other constituents of the extracellular matrix, which are selectively or markedly expressed in the meninges.\(^\text{[2]}\)

The frequently associated distinctive craniofacial features are down-slanting palpebral fissures, ptosis, mandibular hypoplasia, a high palate, and skeletal abnormalities such as hypoplasia of the posterior arch of the atlas, short stature, scoliosis, and kyphosis. Other

---

**Figure 3:** (A) T2-weighted coronal image showing multiple lateral meningoceles with herniation of conus into right lumbar neural foramen. (B and C) T2-weighted right parasagittal MR images showing lateral meningoceles and herniated conus through the neural foramina in the lumbar spine. (D) T2-weighted axial section at L2–L3 levels showing herniation of conus into the meningocele through the right neural foramen
craniofacial features have been described that include hypertelorism, malar hypoplasia, cleft palate, dental crowding, broad, flat or high nasal bridge, and low-set posteriorly angulated ears. Systemic findings include scoliosis/kyphosis, joint hypermobility, wormian bones, loose skin, hyperextensibility, hypotonia, developmental delay, conductive hearing loss, umbilical or an inguinal hernia, cryptorchidism, congenital heart defects, and vertebral anomalies.

Patients with LMS can present with back pain with or without lower limb weakness, atypical pelvic mass/pain, failure to thrive, retrocardiac mass, thoracic kyphosis/kyphoscoliosis, and kidney and ureter dislocation. Therefore, in LMS, chronic symptoms usually develop from compression/deformation by meningeal protrusions on adjacent structures (nerves, viscera, vertebral bodies). Age at onset is unpredictable, although it could be partly related to the volume of the paraspinal mass. Although lateral meningoceles may be congenital in origin, symptom onset is likely influenced by the location of the cyst(s) and altered CSF flow dynamics. Two pediatric patients had presented with failure to thrive, indicating that LMS may rarely present with homeostatic symptoms of unknown origin possibly shared with other hereditary connective tissue disorders. In contrast, when the meningoceles are small, the patient may be asymptomatic, and the lesion may be incidentally diagnosed on a routine chest radiograph.

On conventional radiographs, meningoceles may appear as sharply defined round, smooth, or lobulated paraspinal masses. On CT scan, these lesions appear as well-defined, homogeneous, and low (CSF) attenuation paravertebral masses with or without enlargement of the neural foramina. MRI findings are diagnostic and better delineate the details of multiple lesions including paravertebral expansion and dural ectasias with scalloping of the pedicles, laminae, vertebral bodies, and an enlarged spinal canal.

Hajdu–Cheney and LMS share some physical findings, including thickened skull vault, Wormian bones, midface hypoplasia, hypertelorism, small mandible with an obtuse mandibular angle, biconcave vertebrae, and joint laxity. Acro-osteolysis, a hallmark finding in Hajdu–Cheney syndrome, is absent in LMS.

Lateral meningoceles and dural ectasia have been described either as an isolated defect or as a feature of genetic disorders, such as NF1, Nevo syndrome, and Marfan syndrome. In addition, in patients with NF1, lateral meningoceles are usually isolated and most frequently found at the thoracic level and diagnosed after the fourth decade of life. Nevo syndrome, an allelic condition of the kyphoscoliotic type of Ehlers–Danlos syndrome, caused by pathogenic mutations of PLOD1, might present with dural ectasia, joint and skin laxity, kyphoscoliosis, and hypotonia at birth. However, patients with Nevo syndrome typically show...
Amuthabharathi, et al.: Rare case of Lehmans’s syndrome in an infant

Surgical excision is recommended for symptomatic lesions, particularly when there are neurological deficits or bowel and bladder involvement,[5,8] respiratory distress, or rapid progress in the size of the meningocele.[15,16]

Bilateral multiple-level meningoceles without associated features of neurofibromatosis or Marfan’s syndrome is a rare entity. LMS syndrome is one such condition, which needs to be considered in a patient with multiple lateral meningoceles when associated with abnormal face, neurological, and skeletal findings. MRI is diagnostic in depicting the lesions. The clinicians and radiologists should be aware of this rare clinical entity.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

REFERENCES
1. Correia-Sá I, Horta R, Neto T, Amarante J, Marques M. Lehman syndrome: a new syndrome for Pierre Robin sequence. Cleft Palate Craniofac J 2015;52:369-72.
2. Castori M, Morlino S, Ritielli M, Brancati F, De Bernardo C, Colombi M, et al. Late diagnosis of lateral meningocele syndrome in a 55-year-old woman with symptoms of joint instability and chronic musculoskeletal pain. Am J Med Genet A 2014;164A:528-34.
3. Kumar BE, Hegde KV, Kumari GL, Agrawal A. Bilateral multiple level lateral meningocele. J Clin Imaging Sci 2013;3:1.
4. Gripp KW, Scott Cl Jr, Hughes HE, Wallerstein R, Nicholson L, States L, et al. Lateral meningocele syndrome: three new patients and review of the literature. Am J Med Genet 1997;70:229-39.
5. Seddighi A, Seddighi AS. Lateral sacral meningocele presenting as a gluteal mass: a case report. J Med Case Rep 2010;4:81.
6. Sharma V, Newton G. Lateral cervical meningocele. J Korean Med Sci 1992;7:179-83.
7. Shore RM, Chun RW, Strother CM. Lateral cervical meningocele. Clin Pediatr (Phila) 1982;21:430-3.
8. Göçer AI, Tuna M, Gezercan Y, Boyar B, Bağdatoğlu H. Multiple anterolateral cervical meningoceles associated with neurofibromatosis. Neurosurg Rev 1999;22:124-6.
9. Erkulvrawat S, ElGamal T, Hawkins J, Green JB, Srinivasan G. Intrathoracic meningoceles and neurofibromatosis. Arch Neurol 1979;36:557-9.
10. Kaur N, Mishra SC, Vijayaragvan P, Minocha VR. Lateral sacral meningocele as a gluteal swelling: an unusual presentation. J Indian Med Assoc 2005;103:554-6.
11. Alves D, Sampaio M, Figueiredo R, Leão M. Lateral meningocele syndrome: additional report and further evidence supporting a connective tissue basis. Am J Med Genet A 2013;161A:1768-72.
12. Ejaez R, Qin W, Huang L, Blaser S, Tetreault M, Hartley T, et al.; Care4Rare Canada Consortium. Lateral meningocele (Lehman) syndrome: a child with a novel NOTCH3 mutation. Am J Med Genet A 2016;170A:1070-5.
13. Reis C, Carneiro E, Fonseca J, Pereira P, Vaz R, Pinto R, et al. Epithelioid hemangioendothelioma and multiple thoraco-lumbar lateral meningoceles: two rare pathological entities in a patient with NF-1. Neuroradiology 2005;47:165-9.
14. Gripp KW. Lateral meningocele syndrome and Hajdu-Cheney syndrome: different disorders with overlapping phenotypes. Am J Med Genet A 2011;155A:1773-4; author reply 1775.
15. Mizuno J, Nakagawa H, Yamada T, Watabe T. Intrathoracic giant meningocele developing hydrothorax: a case report. J Spinal Disord Tech 2002;15:529-32.
16. Ebara S, Yuzawa Y, Kinoshita T, Takahashi J, Nakamura I, Hirabayashi H, et al. A neurofibromatosis type 1 patient with severe kyphoscoliosis and intrathoracic meningocele. J Clin Neurosci 2003;10:268-72.