Lumbosacral lipomyelomeningocele with anomalous osseous limb in a 3-month-old female

Sean L. Wilkes, MS, ALM; Jay J. Choi, MD, MSc; and Veronica J. Rooks, MD

A patient with lipomyelomeningocele (known in utero) presented for MRI characterization prior to surgical procedure at three months of age. Cross-sectional imaging revealed a spinal dysraphism of the lower lumbar spine, with a posterior spinal defect spanning L4 to S2 subcutaneous fat intrusion, and distal spinal cord extrusion. An osseous excrescence was also appreciated, articulating with the left iliac bone. This case demonstrates the youngest known lipomyelomeningocele with accessory limb and the abnormal growth of multiple tissue types at the site of spinal dysraphism—a potential consequence of dedifferentiated cell proliferation originating from a secondary neural tube defect or rachipagus parasitic twinning.

Introduction

Spinal dysraphisms are developmental congenital anomalies resulting from dorsal embryo malformations. These malformations are classified as open (as found in spina bifida cystica) or closed (as seen in spina bifida occulta), and are typically discovered in utero, at birth, or during infancy. Lipomyelomeningocele is a closed neural-tube defect (NTD) formed mainly due to a defect in primary neurulation in which mesenchymal tissue enters into neural placode and forms lipomatous tissue (1-3). The spinal cord becomes tethered to the fat and may then be pulled downward in the course of development as well as extruded from the vertebral canal into adjacent tissues. Various clinical impairments may develop.

Case report

A 3-month-old female presented with lipomyelomeningocele for further characterization prior to surgical resection. The patient was born at 39 weeks via spontaneous vaginal delivery with no other known complications. A 3-cm lumbosacral lipomyelomeningocele was discovered via MRI at 28 weeks of gestation. Fetal MR imaging was performed at 28 weeks of gestation using a 12-channel body coil with the same 1.5 T Signa MRI unit. The following sequences were obtained: axial T2 single shot FSE (SSFE, with and without fat suppression); sagittal T2 SSFE; and coronal T2 SSFE. The field of view was 41 x 41 mm, and the matrix size was 384 x 224 with a NEX of 0.57. The slice thickness was 6 mm, with spacing of 1 mm.

Neonatal head ultrasound was performed at birth to evaluate for ventriculomegaly, which was normal. The patient demonstrated normal motor and sensory activity and was subsequently discharged with plans for followup in an outpatient setting and referral to a pediatric neurosurgeon. At three-month followup, the patient continued to demonstrate absence of any sensorimotor or developmental pathology. MR imaging was performed to prepare for neurosurgical intervention. 3D MR imaging of the lumbar spine was performed using a body spine array coil with a 1.5TGE Healthcare. The following sequences were obtained: axial T2 single shot FSE, T1 FSE (with and without fat suppression), and T2 FSE (with and without fat suppression); sagittal T2 SSFE; and coronal T2 SSFE. The field of view was 41 x 41 mm, and the matrix size was 384 x 224 with a NEX of 0.57. The slice thickness was 6 mm, with spacing of 1 mm.

Dr. Wilkes is affiliated with the Uniformed Services University of the Health Sciences, Bethesda MD, and Drs. Choi and Rooks are both in the Department of Radiology, Tripler Army Medical Center, Honolulu HI. Contact Dr. Wilkes at sean.wilkes@usuhs.edu.

Competing Interests: The authors have declared that no competing interests exist.

DOI: 10.2484/rcr.v10i1.1051
and the matrix size was 256 x 128 with a NEX of 2.0. The slice thickness was 5 mm, with spacing of 2 mm. The MR images (Figs 1-3) demonstrated a defect of the posterior elements spanning L4-S2 covered by subcutaneous tissue. Subcutaneous fat was noted within the spinal column at the level of the defect on the left, causing rightward shift of the distal spinal cord and surrounding CSF. The distal spinal cord was low-lying, exiting the defect at the L4-5 level with the CSF-filled meningocele and extending cranially to the L3 level.

A thin curvilinear osseous limb, 4 cm in length with cortex and medullary bone, articulating with the left sacrum, was seen extending posteriorly through the subcutaneous fat, consistent with an osseous limb.

**Discussion**

Lipomyelomeningoceles are a category of spinal dysraphisms due to a defect in primary neurulation from non-disjunction in which mesenchymal tissue enters into the neural placode and forms lipomatous tissue (Fig. 4). The spinal cord then becomes tethered to the lipomatous mass. Our patient, with documented intrauterine meningocele, presented at birth with a lipomyelomeningocele at the lumbar-sacral level associated with an osseous appendage. Similar anomalous findings have been reported previously by Lee et al. and Wasnik et al. (4, 5). Wasnik et al.
scribed the appendage as a "rudimentary accessory limb," while Lee et al. argued that because of the absence of limb structures such as muscle, long bones, and digits, a more accurate moniker would be "anomalous bone associated with spinal dysraphism." A number of similar cases have been previously described since 1975 (6-14).

Here we delineate the youngest account of a lipomyelomeningocele, in a 3-month-old female with an anomalous bone articulating with the left iliac bone. Our review of the literature did not reveal any prior documentation of such an anomaly at this early an age. The underlying cause of this abnormality is not entirely understood; however, multiple theories have been proposed.

Most NTDs are suspected to result from an arrest in the process of neural-tube closure (15). Various theories describing the precise process of neural-tube closure persist, but most have in common a "zipper" mechanism whereby the dorsal surfaces of the neural folds fuse in a cranial-to-caudal fashion, with a number of alternate proposed sites of initiation (16-19). This stepwise process is carefully modulated by a complex network of genes and is influenced by a wide range of environmental factors. Consequently, a number of chromosomal abnormalities and genetic disorders have been associated with failure in the process and resultant NTDs, including autosomal trisomy, cerebrocostomandibular syndrome, Waardenburg syndrome, and a mutation in BMP4 (15). Among nutritional components, a deficiency in folate (which is particularly critical to DNA and RNA synthesis) has also been shown to result in NTDs. Finally, environmental factors that have been shown to be associated with increased risk of NTD include radiation exposure, prenatal maternal alcohol abuse, maternal infection, and exposure to benzene.

Gardner and Egar proffer two classifications of NTDs: primary NTD, as described above, wherein the neural tube fails to close, resulting in simple spinal dysraphism; and a secondary NTD, in which a closed neural tube ruptures as a consequence of the overproduction of neural-tube fluid, which can then leak into subcutaneous space (20, 21). The fluid contains Schwann cells, which are thought to revert to a multipotent state capable of differentiating into various tissue types, including bone, fat, cartilage, muscle, and neural tissue. This secondary NTD, with subsequent invasion and proliferation of dedifferentiated cells, may explain the formation of non-neural tissues, such as the lipoma and anomalous bone in our patient, as well as the occasional development of fully formed accessory limbs (13).

An alternate explanation for the development of this anomalous bone structure is the parasitic twin hypothesis, which presupposes a multiple gestation pregnancy. In this case, the failure or maldevelopment of one embryo is thought to result in its attachment to or subsumption by the healthier twin (22). This can lead to the growth of a rudimentary accessory limb that articulates with the lumbar or sacral spine (23) and may, in some instances, be associated with a spinal dysraphism such as lipomyelomeningocele (13). It is therefore not unreasonable to posit that the anomalous osseous structure seen in our patient may represent a poorly developed limb resulting from rachipagus parasitic twinning.

However, the anomalous limb in our patient was poorly developed and lacked cartilage or surrounding muscle, which seems more suggestive of differentiation from multipotent cells as an origin, rather than parasitic twinning.

**Conclusion**

Closed spinal dysraphisms resulting from NTDs can be associated with a variety of anomalous tissue formations, including muscle, fat, and bone. The underlying process by which the formation of this non-neural tissue occurs remains unclear. When a closed spinal dysraphism is noted, assessing for the possibility of an accessory limb or additional teratomatous element is important for proper surgical planning.

**References**

1. Sarris CE, Tomei KL, Carmel PW, Gandhi CD. Lipomyelomeningocele: pathology, treatment, and outcomes. *Neurosurg Focus.* 2012 Oct;33(4):E3. [PubMed]
2. Finn MA, Walker ML. Spinal lipomas: clinical spectrum, embryology, and treatment. *Neurosurg Focus.* 2007;23(2):E10. [PubMed]
3. Kanev PM, Lemire RJ, Loeser JD, Berger MS. Management and long-term follow-up review of children with lipomyelomeningocele, 1952–1987. *J Neurosurg.* 1990 Jul;73(1):48-52. [PubMed]
4. Lee SH, Je BK, Kim SB, Kim, BH. Adult with sacral lipomyelomeningocele covered by an anomalous bone articulated with iliac bone: Computed tomography and magnetic resonance images. *Congenit Anom (Kyoto).* 2012 Jun;52(2):115-8. [PubMed]
5. Wasnik APAS, Lalchandani UR, Gujrathi R, Pai BU. Rudimentary third lower limb in association with spi-
Lumbosacral lipomyelomeningocele with anomalous osseous limb in a 3-month-old female

6. Theander G. Malformation of the iliac bone associated with intraspinal abnormalities. *Pediatr Radiol*. 1975 Sep 15;3(4):235-8. [PubMed]

7. McAlister WH, Siegel MJ, Shackelford GD. A congenital iliac anomaly often associated with sacral lipoma and ipsilateral lower extremity weakness. *Skeletal Radiol*. 1978:3(3):161-6. DOI 10.1007/BF00347363

8. Cohen JY, Lebatard-Sart RER, Lajat Y, Mitard D, David A. Sacral intraspinal lipoma associated with genital iliac anomaly. *Childs Brain*. 1981;8(3):181-8. [PubMed]

9. Krishna A, Chandha S, Mishra NK, Gupta AK, Upadhyaya P. Accessory limb associated with spinal bifida. *J Pediatr Surg*. 1989 Jun;24(6):604-6. [PubMed]

10. Scalif JH, Kendall BE, Kingsley DPE, Britton J, Grant DN, Hayward RD. Closed spinal dysraphisms: Analysis of clinical, radiological, and surgical findings in 104 consecutive patients. *Am J Roentgenol*. 1989 May;152(5):1049-57. [PubMed]

11. Parkinson D. Accessory limbs and spinal dysraphism. *J Neurosurg*. 1991 Sep;75(3):498-9. [PubMed]

12. Krishna A, Lal P. Accessory limbs associated with spina bifida - a second look. *Pediatr Surg Int*. 1999;15(3-4):248-50. [PubMed]

13. Murphy RF, Cohen BH, Muhlauer MS, Eubanks JW, Sawyer JR, Moisan A, et al. An accessory limb with lipomyelomeningocele in a male. *Pediatr Surg Int*. 2013 Jul;29(7):749-52. [PubMed]

14. Bayri Y, Tannkulu B, Eksi MS, Dagcinar A. Accessory lower limb associated with spina bifida: case report. *Childs Nerv Syst*. 2014 Aug 5. [Epub ahead of print]. [PubMed]

15. Padmanabhan R. Etiology, pathogenesis, and prevention of neural tube defects. *Conagen Anum (Kyoto)*. 2006 Jun;46(2):55-67. [PubMed]

16. Copp AJ, Bernfield M. Etiology and pathogenesis of human neural tube defects: Insights from mouse models. *Curr Opin Pediatr*. 1994 Dec;6(6):624-31. [PubMed]

17. Van Allen MI. Multisite neural tube closure in humans. *Birth Defects Orig Artic Ser*. 1996;30(1):203-25. [PubMed]

18. Nakatsu T, Uwabe C, Shiota K. Neural tube closure in humans initiates at multiple sites: Evidence from human embryos and implications for the pathogenesis of neural tube defects. *Anat Embryol (Berl)*. 2000 Jun;201(6):455-66. [PubMed]

19. O’Rahilly R, Muller F. The two sites of fusion of the neural folds and the two neuromeres in the human embryo. *Teratology*. 2002 Apr;65(4):162-70. [PubMed]

20. Gardner WJ. Hypothesis; over distention of the neural tube may cause anomalies of non-neural organs. *Teratology*. 1980 Oct;22(2):229-38. [PubMed]

21. Egar MW. Accessory limb production by nerve-induced cell proliferation. *Anat Rec*. 1988 May;221(1):550-64. [PubMed]

22. Hoefel CC, Nguyen KQ, Phan HT, Truong NH, Nguyen TS, Tran TT, et al. (2000). Fetus in fetu: a case report and literature review. *Pediatrics*. 2000 Jun;105(6):1335-44. [PubMed]

23. Chadha R, Lal P, Singh D, Sharma A, Choudhury SR. Lumbosacral parasitic rachipagus twin. *J Pediatr Surg*. 2006 Jan;41(1):e45-8. [PubMed]