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

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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 proton density FSE, T1 FSE (with and without fat suppression), and T2 FSE (with and without fat suppression); sagittal T1 FSE non-fat-suppressed and T2 FSE; and gadolinium-enhanced axial and sagittal T1 FSE (with fat suppression). The field of view was 28 x 28 mm,

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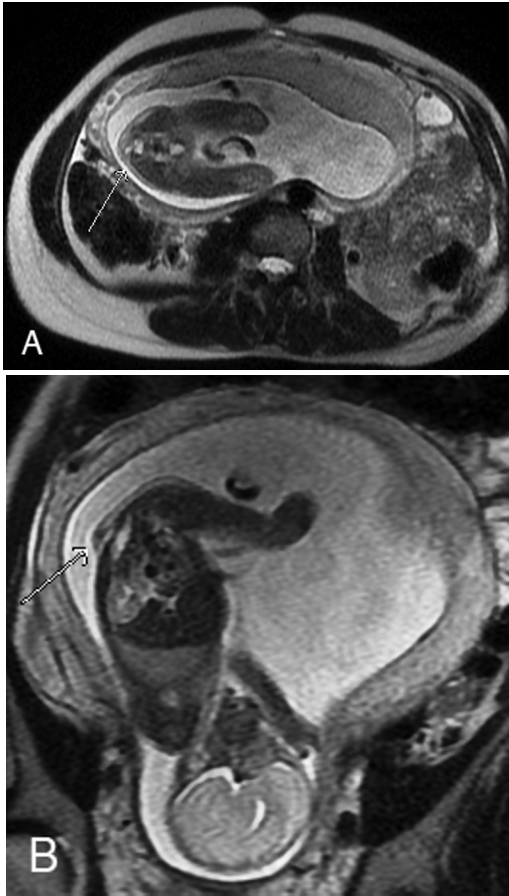
Fig. 1. Axial T2 SSFSE non-fat-suppressed image. The arrow demonstrates protrusion of CSF and neural elements through a covered posterior fusion defect (A). Sagittal T2 SSFSE non-fat-suppressed image demonstrates protrusion of CSF and neural elements through a covered posterior fusion defect (B).
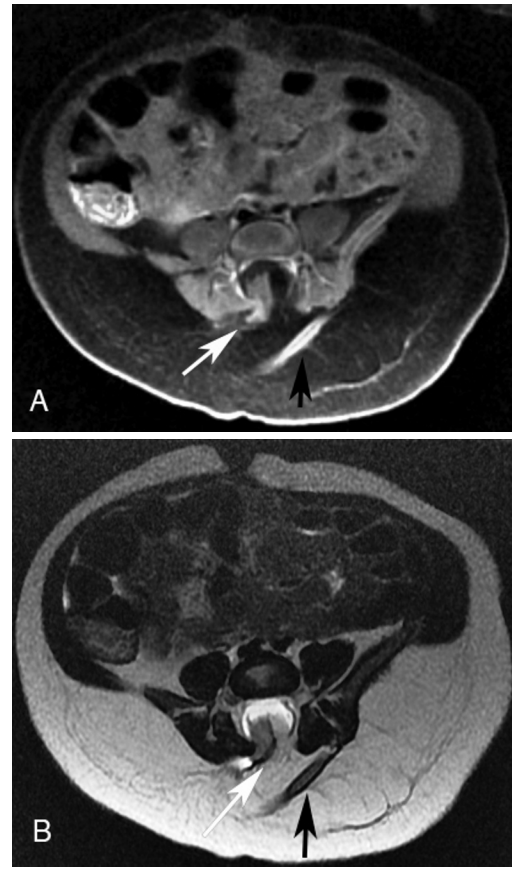

Fig. 2. Axial T1 postcontrast, fat sat demonstrates sacral defect (white arrow) and the sacral accessory limb (black arrow) (A). Axial T2 FRFSE non-fat-suppressed image demonstrates the same (B).

Fig. 3. Sagittal T2 FRFSE image demonstrates a lipomyelomeningocele (arrow), not in communication with an adjacent simple cyst.

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. de-
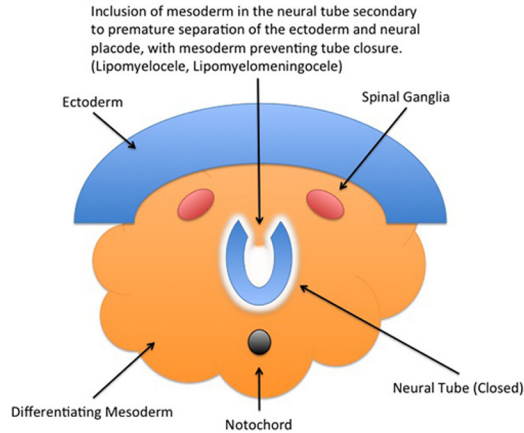
Fig. 4. Premature disjunction abnormality of primary neurulation. Lipomyelomeningocele results from mesenchymal tissue entering the neural placode, leading to the formation of lipomatous tissue and tethering of the spinal cord to the lipomatous mass.

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 lipomyelomenignocele (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 dyraphism is noted, assessing for the possibility of an accessory limb or additional teratomatous element is important for proper surgical planning.

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