Terminal Myelocystocele: 
Important Differential Diagnosis in the 
Prenatal Assessment of Spina Bifida

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The most common lumbosacral mass of a fetus is a meningomyelocele that is not covered by skin; however, skin-covered lesions do occur, and they often have a quite different clinical cause and outcome. It is often difficult to make this differentiation on ultrasonography; when a skin-covered mass in the lumbosacral region is encountered, the following differential diagnoses should be considered. The most common skin-covered lesion is the lumbosacral lipoma (i.e., lipomeningocele or lipoma), followed by the teratoma, hamartoma, and the other rarer tumors and mass lesions.1 The TM appears as a skin-covered mass in the lumbosacral region, sometimes associated with urologic and gastrointestinal tract abnormalities.2,3 Although TMs may appear similar to myelomeningoceles on prenatal ultrasonography, it is of the utmost importance to distinguish them, as the outcome and prognosis and thus the prenatal counseling will be quite different.4 In addition to the presumed difference in pathophysiology, there is no familial occurrence and therefore no genetic predisposition for future offspring.2,4,5 We present a patient with TM and describe the prenatal findings, postnatal studies, and clinical course.

CASE REPORT

A 30 year old primigravida woman was seen for prenatal care 8 weeks after her last menstrual period, without complaints. No family history of neural tube defects existed, and AFP level was normal (0.55 multiple of the median). An ultrasonographic examination performed for dating and anatomy demonstrated a 19 1/2 week fetus by measurements and a lumbosacral mass of intermediate echogenicity, with splaying of the posterior elements from the third lumbar vertebra to the lower sacrum. The fetus demonstrated bilateral lower extremity motion, a normal-shaped cranium, no evidence of ventriculomegaly, and a normal posterior fossa, and the presumptive diagnosis was that of myelomeningocele. Amniocentesis revealed the fetus to have a normal karyotype and normal amniotic fluid AFP level (1.5 multiples of the median). A repeat

ABBREVIATIONS

AFP, Alpha-fetoprotein; MR, Magnetic resonance; TM, Terminal myelocystocele; OEIS, Omphalocele, exstrophy, imperforate anus, spinal defects

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sonogram at 26 weeks showed appropriate fetal growth, the lumbosacral defect, continued limb movement, normal posterior fossa, and normal-sized cerebral ventricles. Termination of the pregnancy was discussed. The couple elected to continue the pregnancy. Pediatric neurosurgery and Fetal Diagnosis Center consultations were obtained. Three-dimensional ultrasonographic examination of the fetus was performed after informed consent was obtained. Splaying of the posterior elements of the vertebral bodies was demonstrated from the second lumbar vertebral body to the sacrum. A mass was seen protruding from the back, with an echogenic extension of tissue into the dominant mass, suggesting lipomatous infiltration (Figs. 1, 2). The pediatric neurosurgery consultant suspected a skin-covered lesion compatible with a spinal hamartoma, a myelocystocele, or, less likely, a spinal teratoma. The baby was delivered at 38 weeks’ gestation by elective cesarean section, weighing 3600 g, with Apgar scores of 6 and 9 at 1 and 5 min, respectively. A large skin-covered dorsal midline lumbar mass was noted, and neurologic examination of the female infant revealed mild bilateral S2 denervation by plantar grasp weakness. Cranial MR imaging showed a normal brain, including the posterior fossa. The spinal MR image revealed the characteristic findings of a TM: a lumbosacral spinal defect extending from L2 to S2, with splaying of the posterior elements from the superior lipomatous structure within the distal cord (lipoma). The cord was low (tethered) as a result of the previously mentioned abnormalities (Figs. 3, 4). Results of renal and bladder ultrasonographic examinations were normal. The child was discharged home, where she did well, and was electively admitted for surgery at 6 weeks of age. At operation the mass was resected, beginning at the third lumbar vertebra; with microsurgery the dorsal portion of the myelocystocele was amputated and the conus released. Postoperatively, the infant was unchanged neurologically and did well. She was discharged on the third postoperative day, and on a 4 month follow-up visit she was found to be developing normally except for weakness of plantar grasp on the left side. Yearly neurosurgical and urologic assessments will be performed until body growth is completed.

DISCUSSION

TMs constitute approximately 3.5 to 7% of the skin-covered dysraphic states overlying the lower spine. TM is a rare form of occult spinal dysraphism occurring in the lumbosacral region. It is characterized by a large, ependyma-lined, cystic dilation of the caudal end of the central canal of the spinal cord; it projects dorsally through a lamina defect, with overlying varying amounts of lipomatous subcutaneous tissue. Myelocystoceles are associated with a tethered cord and meningocele, which communicates with the spinal subarachnoid space, but not with the central canal cyst.
TMs have a distinctly different pathophysiology and anatomy from those of cervical myelocystoceles and therefore are probably a different category of a spinal dysraphic state. The embryology of the TM also appears to be different from that of a myelomeningocele. Most of the central nervous system forms by the process of neurulation, whereby the neural plate formed of thickened embryonic ectoderm folds upon itself to form the neural tube. Both ends of this neural tube close by day 27 or 28 of gestation. An abnormality of closure of the caudal neuropore is responsible for defects such as myelomeningoceles. The caudal neural tube forms from the caudal cell mass of undifferentiated cells. These cells transform to a neural appearance and canalize, completing this process by day 48 of gestation. The rostral portion of the caudal neural tube, which gives rise to the caudal spinal cord (i.e., the future conus medullaris), joins that part of the cord formed by neurulation. Within this portion of the caudal neural tube is an area of dilated central canal known as the ventriculus terminalis. From around day 48 of gestation until some time postnatally, atrophy of the caudal neural tube rostrally to the level of the ventriculus terminalis proceeds, resulting in formation of the filum terminale. This process, known as retrogressive differentiation, in addition to disproportionate growth of the spine and spinal cord, is responsible for the relative ascent of the caudal end of the spinal cord to its final adult position at the L1-2 interspace.

Although the exact mechanism is unknown, the insult responsible for the formation of the TM probably occurs during the process of retrogressive differentiation. An obstruction of the outflow of cerebrospinal fluid from the neural tube is theorized to cause progressive dilation of the terminal ventricle. This dilation disrupts the dorsal mesenchyme (i.e., future posterior elements of the spinal canal) but not the superficial ectoderm, resulting in spina bifida with an intact skin cover. The ballooning terminal ventricle carries with it the surrounding arachnoid, creating a circumferential flaring caused by the enlarging terminal cyst that also tethers the spinal cord.

**Figure 2** Surface-rendered image of three-dimensional ultrasoundography of myelocystocele. A mass (arrowheads) is seen protruding from the back, which corresponds to the myelocystocele (sagittal or profile image of the fetus). Note that the head would be at the superior aspect of the image and the feet at the inferior aspect. I, Iliac bone; arrows, sacrum.

**Figure 3** Spinal sagittal MR image (spin echo, TR 500, TE 15) reveals a tethered cord (small light arrow), the terminal syrinx (large light arrow), the meningocele (small dark arrow), and the dorsal lipoma (large black arrow).
cord by preventing its normal ascent because of its size.\textsuperscript{5} The ependymally lined terminal cyst communicates freely with the central canal of the spinal cord, and the meningocele communicates with the subarachnoid space surrounding the spinal cord. However, the cyst and meningocele do not communicate.\textsuperscript{4,5,13}

Although myelocystoceles may be isolated, as in this case report, they are often found in association with the midline abdominal and pelvic defects termed the OEIS complex,\textsuperscript{2} which consists of omphalocele, extrophy of the bladder, imperforate anus, and spinal defects and is often considered to be in the spectrum of limb–body wall complex. Other defects are also common, including abnormalities of the external genitalia and upper urinary tract, malrotation and bowel shortening, and extremity deformities such as clubfeet, polydactyly, and syndactyly.\textsuperscript{2,3} Lipomeningoceles, lipomyelomeningoceles, tethered cord, meningoceles, lipomas, syringomyelias, fatty fila, and hemivertebrae have also been described with this complex.\textsuperscript{2,3}

Identification of a mass in the lumbosacral region of the spine on the prenatal ultrasonographic examination of the fetus should suggest the possibility of meningomyelocele, as well as skin-covered lesions. Misinterpretation of a myelocystocele as a myelomeningocele could have tragic consequences, such as termination of pregnancy. The ultrasonographer should be familiar with the appearance of a TM on ultrasonography: splaying of posterior elements of vertebral bodies with protruding mass, extension of echogenic material from the spinal canal into the mass, normal fetal head with normal-sized ventricles, normal posterior fossa, and no evidence of a deformed skull (i.e., lemon sign). Three-dimensional ultrasonographic imaging was useful in this case in that the level of the defect was accurately identified. It was possible to view the transverse plane through the spine at the same time as the rendered image of the spine, which allowed referencing of the specific vertebral level. The obstetrician should be highly suspicious of the diagnosis of a myelocystocele when maternal serum AFP and serum acetylcholinesterase levels are normal.

Clinically, neonates with TM have milder symptoms than those with meningocele. Although neonates have been neurologically intact,\textsuperscript{13} most authors report some degree of lower extremity dysfunction.\textsuperscript{2–5} Patients may be seen with normal neurologic function and then deteriorate over time secondary to the tethering of the distal cord.\textsuperscript{4} In patients without the chromosomal anomalies, intellectual development is usually normal.\textsuperscript{4,5} In addition, although few reports describe hindbrain abnormalities (i.e., Chiari malformations) associated with terminal myelocystocele,\textsuperscript{8,13} most reports show no association with Chiari malformations or hydrocephalus.

Although the numbers of reported cases are small and the issue has not been studied in depth, these disorders seem to be sporadic in that no evidence exists that TM or the OEIS syndrome is an inherited condition.\textsuperscript{2,4} This problem needs to be studied further to more appropriately counsel parents regarding the risks of recurrence of these disorders in future offspring. Teratogens such as hydantoin,\textsuperscript{2} Eoperamide,\textsuperscript{13} and retinoic acid\textsuperscript{13} have also been implicated as a possible cause of these disorders.

Patients with TM often have little or no neurologic deficit, and, therefore, the question of whether to treat these lesions does arise. We believe that TMs should be surgically treated and the spinal cord untethered on an elective basis soon after birth because of the reversible nature of the deficits when treated and the progressive symptoms when left untreated.\textsuperscript{3,4} If treated early, the prognosis for these children both intellectually and from a functional motor standpoint is excellent.\textsuperscript{3,5}

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Figure 4 Spinal axial MR image (spin echo, TR 600, TE 50) shows the previously described findings on the sagittal image. The terminal syrinx (small light arrow) is surrounded by the meningocele (small black arrow) and the subcutaneous lipoma interface (large black arrow).
REFERENCES


