Terminal myelocystocele

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Object. The authors describe the cases of eight patients with terminal myelocystoceles and report on their initial diagnoses, neuroimaging findings, surgical procedures, and clinical follow-up examinations.

Methods. There were seven girls and one boy. The initial age at diagnosis ranged from prenatal age (determined by imaging) to 14 years. Operative correction was performed from 6 weeks to 14 years of age. One patient presented with a large prevertebral (intraabdominal) meningocele. Four patients have associated abdominal wall defects and are significantly handicapped. Two are wheelchair bound and the other two ambulate with prosthetic devices; all are incontinent. The four patients without abdominal wall defects are fully ambulatory; one wears an ankle–foot orthosis. One patient has normal bowel and bladder functions, and the other three require intermittent clean catheterization. No patient to date has had clinical evidence of retethering, although a low-set conus medullaris and residual lipoma were visible on postoperative neuroimaging. No patient in this series had hydrocephalus or Chiari malformation.

Conclusions. It is concluded that patients with myelocystoceles without abdominal wall defects have a favorable neurological outcome when compared with those having ventral wall defects.

KEY WORDS • myelocystocele • spinal dysraphism • cloacal extrophy • pediatric neurosurgery

A terminal myelocystocele constitutes a distinct clinical entity that presents as a skin-covered mass overlying the distal dorsal spine. The low-lying distal spinal cord migrates into a dorsal meningocele and expands into an ependyma-lined cystic structure continuous with the central canal overlying the distal spine.10–12

Abdominal wall defects such as extrophy of the cloaca (vesicointestinal fissure) are commonly associated with spinal dysraphism.7 The most common form of skin-covered dysraphism is a terminal myelocystocele,6,12 although terminal myelocystoceles can occur independently of abdominal defects.6,14 Authors of previous reports have primarily described patients who had terminal myelocystoceles associated with abdominal wall defects.6,14 In the present article we report on a series of patients with and without this association to delineate the difference in the outcome of these two subsets.

Clinical Material and Methods

Patient Population

Between 1985 and 2002, in eight patients (seven girls and one boy) a diagnosis of a terminal myelocystocele was established at the Children’s Hospital of San Diego. Four of the patients had no abdominal wall defects and the others did (such as cloacal extrophy and associated anomalies).

Abbreviation used in this paper: MR = magnetic resonance.

Age at Time of Diagnosis

Patients were placed into one of two groups according to the age at which the terminal myelocystocele was discovered. The infants (five) presented at birth with a skin-covered mass, and only one had an associated abdominal wall defect identified at 4 months of age. The diagnosis in the latter patient was delayed because she initially presented with bladder extrophy. The spinal lesion was later revealed on the pelvic MR image. One of the infants had been suspected to have a terminal myelocystocele on prenatal imaging, and this has been previously reported.13 In three patients a diagnosis was established in childhood (two) and adolescence (one). All three had bladder extrophy and associated anomalies.

Neuroimaging Procedure

All patients underwent preoperative spinal as well as cranial MR imaging either prior to or after spinal surgery. All of the preoperative spinal MR imaging studies revealed the previously described findings of the terminal myelocystocele:12,15 a low-set conus medullaris with a distal cystic dilatation (syringocele), tethering of the distal cord to the area of the meningocele–lipoma interface with a variable mass effect from the meningocele, and a spinal/subcutaneous lipoma. In addition, one patient who was 8 years old at the time of presentation had a very large intraabdominal ventral meningocele and a paraspinal mass suggestive of a dermoid tumor.
Physical Examination

Infants. The four infants presented with a skin-covered mass of variable size in the lumbosacral region that was soft to palpation. A cutaneous hemangioma was present in two of them. All had good muscle bulk and strength in the lower extremities, except for one who had unilateral weakness in dorsi- and plantar flexion, and a slight cavus deformity of the right foot. All four had good range of motion of the lower extremities. Two of the patients had good pelvic tone on perineal palpation. In the fifth infant a diagnosis was made at 4 months of age, and she was the only one of this group who had an abdominal wall defect. She had significant muscle wasting of the legs, with practically no dorsi- or plantar flexion of both feet and clubfoot deformity.

Children and Adolescent. All three patients had in common a skin-covered, soft-tissue mass in the lumbosacral region as well as abdominal wall defects. One child’s myelocystocele was found at 26 months of age. She had a congenitally amputated right leg and a clubfoot deformity of the left foot with no dorsi- or plantar flexion and significant wasting of her left leg muscles and a partially repaired abdominal wall defect.

In the second child a diagnosis was made at 8 years of age, and she also had a partially repaired abdominal wall defect. She presented with severe abdominal distention, and the diagnosis of a large prevertebral meningocele was made based on the MR image. She had significant muscle wasting of both lower extremities and some dorsi- and plantar flexion of the right foot, but none on the left. She was wheelchair bound.

A diagnosis was made in the third patient at 14 years of age. She had severe wasting of both lower extremities with global weakness involving the distal musculature more so than the proximal. She walked with a limp and required a bilateral ankle–foot orthosis. Her knee reflexes were brisk, and ankle reflexes were absent. Low-back pain was present, and it was exaggerated during spinal flexion. All three patients had absent and/or denervated sphincters.

Operative Interventions

The children who had a diagnosis in infancy all underwent surgery in early life (at 1, 6, 9, 10, and 100 days of age). Those in whom the diagnosis was made in childhood and adolescence underwent operative intervention at that age (2, 8, and 14 years). In the infants with no abdominal wall defects the procedure was performed with continuous rectal electromyographic monitoring via the urodynamic monitoring system. The lipomatous mass and the meningocele were dissected with a freehand CO2 laser to minimize blood loss. The CO2 laser was subsequently attached to the operating microscope, the syringocele was drained by myelotomy, and the cord was untethered. All patients were discharged to their home between postoperative Days 3 and 7.

Results

Immediate Postoperative Course

Two patients acquired postoperative dysesthesias in a lower extremity that spontaneously resolved. One patient experienced immediate weakness in one leg, which dissipated over several weeks. Persistent subcutaneous fistula of the cerebrospinal fluid developed in two patients. To avoid wound dehiscence, they were treated with a temporary subcutaneous lumboperitoneal shunt. Several months later, as the fistulas resolved, the shunts were removed. A wound infection developed in one of these patients after the placement of the shunt, and the patient was treated with antibiotic agents and shunt removal.

Long-Term Follow Up

The duration of follow up in this series ranged from 0.5 to 9 years (0.5, 1, 2.5, 3.5, 5, 6.5, 7, and 9 years). All patients who were walking prior to surgery retained ambulation. All infants who underwent surgery in early life acquired age-appropriate levels of ambulation. Only one patient acquired sphincter continence, at 3 years of age, and she had no abdominal wall defects.

All patients underwent spinal MR imaging approximately 6 months after their operation. All of them had a persistent low-set conus medullaris and residual lipoma. One patient had a small, recurrent syringocele that was asymptomatic. The patient with the intraabdominal meningocele had a complete resolution as revealed on follow-up MR imaging. During surgery, it was drained by aspiration through the opening via a posterior approach and the dural connection was repaired. The dermoid tumor discovered using MR imaging in a location ventrolateral to the spinal lumbar spinal column has not yet been surgically addressed.

No patient in this series has suffered from hydrocephalus. All patients demonstrate age-appropriate behavior and none has evidence of developmental delay. Those who are of school age are in an age-appropriate classroom setting.

Operative Intervention

The patients with ventral wall defects underwent multiple surgical procedures to repair them. In addition, orthopedic procedures were required either to stabilize the pelvis and/or to address the lower-extremity congenital anomalies or problems arising from muscle imbalance. These patients required multiple (range five [three patients]–12 [two patients]) interventions. As a consequence, they were hospitalized multiple times.

The four patients without ventral wall defects required no abdominal and/or pelvic surgical procedures. The patient who underwent tendon transposition surgery in one foot was the only one who requires an orthotic device for ambulation (an ankle–foot orthosis). The remainder of the patients—other than the one who underwent myelocystocele repair with tethered cord release—have not required operative interventions. As a consequence, they have had fewer hospitalizations. Table 1 summarizes the clinical aspects of the patients in this series.

Multidisciplinary Support

All patients are being followed up in the multidisciplinary setting of the Spinal Defects Clinic. This is a team approach, with the physicians and allied health resources combining to maximize the overall long-term monitoring and care that children with congenital spinal conditions require.

Discussion

The exact cause of terminal myelocystoceles is unknown,
Clinical outcome in patients with myelocystoceles

We must note the unique patient reported by Cartmill, et al.,7 with terminal myelocystocele who presented with progressive neurological deficits without a visible lumbosacral mass. The authors of this report emphasize the importance of early neuroimaging in children with progressive neurological dysfunction of the lower extremities as a way of detecting this variation of the disease process.

Conclusions

The current patient series demonstrates significant clinical course differences in patients with terminal myelocystoceles with or without abdominal defects. The terminal myelocystocele was discovered earlier in life in patients without abdominal defects. The patients with abdominal wall defects consequently underwent surgical exploration and untethering later in life. The patients with abdominal wall defects, as expected, underwent far more procedures due to their clinical condition. The patients without abdominal defects seemed to experience better neurological outcomes.

References


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