Double spinal dysraphism

Report of three cases

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The simultaneous presence of multiple spinal neural tube defects is unusual. There have been only a few of these cases reported in the literature. The authors report on three cases of double spina bifida cystica. One patient had two myelomeningoceles (MMCs) at the cervical and lumbosacral regions, one was noted to have both thoracolumbar and sacral defects, and the third presented with double MMCs at lumbar and lumbosacral levels. All three neonates in these cases underwent surgical treatment and ventriculoperitoneal (VP) shunt insertion for associated hydrocephalus. One child died at the age of 2 months despite a well-functioning VP shunt. The other two patients had no complications. Current models of neural tube closure do not thoroughly explain the mechanisms of multiple spinal dysraphism, but the multisite closure model provides a better understanding of caudal neural tube closure than other closure-site models.

KEY WORDS • myelomeningocele • neural tube defect • spinal dysraphism • embryogenesis • pediatric neurosurgery

The presence of MMCs at multiple levels of the spinal cord is a rare condition. To our knowledge, only 11 cases have been published to date.1–4,10–12,15,16 Although several closure-site models for human NTDs have been proposed, the “multisite closure” model of Van Allen and colleagues17 appears to provide the best explanation for the embryogenesis of such conditions. In this report, we present three cases of double spina bifida cystica and review the relevant literature.

Case Reports

Case 1

This 8-day-old girl was found to have two lesions at presentation, one over the neck and the other in the lower back. Physical examination revealed a 5 × 6-cm upper cervical MMC at the midline, with a defined neck and dystrophic skin (Fig. 1 left). There was also a smaller 2 × 2-cm midline defect at the lumbosacral region without any skin coverage on the placode. Her motor and sensory examination results were normal; however, high-pressure bladder and vesicoureteral reflux were discovered by urodynamic tests and voiding cystography, respectively. A triventricular hydrocephalus was detected using brain ultrasonography. In addition, magnetic resonance imaging revealed an associated CM-II (Fig. 1 right). The cervical mass communicated with the dorsal surface of the spinal cord through a band-like tissue that traversed the fascia and the lamina of C-2. The cervical MMC was repaired and the spinal cord was untethered by cutting the stalk. The lumbosacral lesion was also repaired by releasing the exposed atrophic cord from the surrounding tissue, and using standard neural tube reconstruction and dural closure. A VP shunt was inserted later when the patient was 2 months of age. After 2 years, the patient has motor delay, can stand with assistance, and speaks several words. Her sphincter problem is managed using clean intermittent catheterization and anticholinergic therapy.

Case 2

This 3-day-old girl presented with two cystic masses at the midline, located over the lower thoracic and sacral regions (Fig. 2). The lesion overlying the thoracic spine was 3 × 5 cm and covered by a thin purplish squamous epithelium. The 5 × 6-cm sacral lesion contained an infected exposed neural placode with leakage of cerebrospinal fluid. Motor examination revealed weak hip flexion and adduction as well as bilateral knee and foot paralysis. During extensive urological evaluation the patient was found to have neurogenic bladder, and brain ultrasonography confirmed...
the presence of hydrocephalus. Both lesions were explored during the same surgery through circumferential incisions. Microsurgical exploration of the thoracic lesion showed a connection between the spinal cord and the stalk of the cystic mass, which was cut. The sacral MMC was also repaired using a microsurgical technique. The placode was carefully preserved and converted to a neural tube using pial sutures. No early postoperative complications were encountered. A VP shunt was inserted 4 weeks later. Currently, after 1 year of follow up, her neurological deficits are stable and her urological problems are managed conservatively.

Case 3

This 15-day-old girl was admitted with two cystic masses over the lumbar region (Fig. 3). The lumbosacral defect, measuring $7 \times 5$ cm, had a wide neck with an atrophic and membranous skin. The smaller 1.5 $\times$ 1.5–cm lumbar sac, found just 1 cm above the lumbosacral MMC, was separated from it by a small strip of normal-appearing skin. The patient had paraplegia and a neurogenic bladder. The lumbosacral MMC was repaired by untethering the neural placode and reconstructing the neural tube. The proximal lesion was a sac with a thin fibrovascular band attached to the dorsal aspect of the underlying neural tissue. The stalk was removed very close to the cord to complete the untethering procedure. Insertion of a VP shunt was performed at the same surgery to treat the hydrocephalus detected by brain ultrasonography. There were no early postoperative complications, but the patient died at the age of 2 months after developing cyanosis and apnea despite a well-functioning VP shunt, most probably due to a CM-II.

Discussion

Coexistence of MMCs at multiple spinal cord levels is unusual. We encountered a few cases of double MMCs in the literature, one of which was a case of a triple NTD and a parietooccipital encephalocele, together with cervical and thoracolumbar MMCs. Two of the double MMC cases presented with associated neurofibromatosis Type 1. Among the remaining cases, six patients had a thoracic and/or lumbosacral MMC, whereas the other two were reported to have coexisting cervical and thoracolumbar/lumbar defects. Hydrocephalus was detected in five of these cases. A VP shunt was inserted in three of these patients, one of whom died of bronchopneumonia due to shunt complications.

Neural tube fusion in human embryos begins at postgestational Day 22. The rostral neural tube fuses first at approximately Day 24, followed by caudal neuropore closure at approximately Day 26. By Day 28, the neural tube is completely closed in normal embryos. Myelomeningoceles are believed to result from defective closure of the caudal neural tube between Days 26 and 28.

Previously, human neural tube closure was believed to begin at the region between the third and sixth somites, and continue bidirectionally and continuously (the “zipper” or “single-site” model). In the single-site closure model, cranial and spinal NTDs are assumed to develop from the failure of rostral and caudal neuropore closure, respectively; however, this model cannot explain the mechanism of all types of NTDs and their site-specific variations.

In 1993, Van Allen and coworkers extrapolated mouse model data of neural tube closure to the human and proposed the multisite closure model. According to this model, neural tube closure in humans, as in mice, occurs at sever-
al sites, and NTDs represent defects at one or more of five closure sites. Closure 1 begins in the midcervical region and proceeds bidirectionally. Closure 2 initiates at the prosencephalic–mesencephalic junction and progresses bidirectionally, forming two cranial neuropores. Caudally, closure 2 terminates at the superior aspect of the rhombencephalon where closure 2 meets closure 3 rostrally. Closure 3 appears at the most rostral end of the neural fold, continuing caudally to meet closure 2 and close the rostral neuropore. Closure 4 begins at the caudal end of the rhombencephalon and proceeds rostrally to meet closure 2. Closure 5 is assumed to commence at the most caudal end of the neural tube, progressing rostrally to meet caudal closure 1. As secondary neurulation by canalization is estimated to extend to the S-2 level, the region between L-2 and S-2 is a likely site of closure 5. The closure of the neural tube initiates at closure site 4, and is followed by closure sites 2, 3, 1, and 5.17

All clinically observed NTDs can be described by this model. The cervical MMC in Case 1 is assumed to have resulted from a defect at rostral closure 1, or at the junction of closure 4 and closure 1. The lumbosacral lesions in Cases 1 and 3 and the sacral myeloschisis in Case 2 may have developed from a defect at closure 5, whereas the lumbar lesion in Case 3 could have arisen from the failure of closure 1 to meet closure 5. Defects at caudal closure 1 could have resulted in the thoracic cystic mass in Case 2.

By providing a good explanation for NTDs in humans, the multisite closure model has been the foundation of further studies of neural tube closure in human embryos. Among others, Golden and Chernoff,3 Sanchis Calvo and Martínez-Frías,13 and Seller14 achieved similar results, but Nakatsu et al.7 and O’Rahilly and Müller9 challenged the applicability of the mouse closure model to humans and proposed new models that suggested only two and three closure sites, respectively. Nevertheless, none of these recent models can thoroughly explain the mechanisms of caudal NTDs and the presence of multiple spinal NTDs in our patients.

Although the multisite closure model of Van Allen and colleagues17 appears to provide the best current definition of the anatomical origin of caudal NTDs, there are still many uncertainties about the mechanism and sequence of the simultaneous appearance of multiple spinal dysraphisms at distant levels. Future studies may provide a better understanding of these rare cases.

References


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