Myelomeningocele

Gregory S. Liptak, MD, MPH,* Nienke P. Dosa, MND, MPH*

Author Disclosure
Drs Liptak and Dosa have disclosed no financial relationships relevant to this article. This commentary does not contain a discussion of an unapproved/investigative use of a commercial product/device.

Objectives

After completing this article, readers should be able to:

1. Recognize the signs and symptoms of ventricular shunt failure (increased intracranial pressure).
2. Identify the signs and symptoms of tethered spinal cord.
3. List signs and symptoms of Chiari II malformation.
4. Apply the three-level classification system to patients who have spinal cord abnormalities.
5. Use system-based recommendations for ongoing surveillance and care of patients who have spinal cord abnormalities.

Introduction

Myelomeningocele (meningomyelocele or spina bifida) is a form of neural tube defect. In this condition, the spinal cord (myelon), its covering (meninges), and vertebral arches develop abnormally early in gestation (Fig. 1). Children born with this condition, which is one of the most complex birth defects compatible with life, have impairments of both the spinal cord and brain. Despite fortification of food with folic acid and the increased use of maternal folic acid, myelomeningocele has not been eliminated. Thus, the primary care pediatrician is likely to provide care for children who have this condition. The pediatrician should work with the child, family, and other clinicians in the community to establish sound chronic disease management procedures for these children, ensuring the provision of continuous and coordinated care of the highest quality.

The prevalence of myelomeningocele varies among countries; in the United States, it is approximately 60 per 100,000 births. The causes remain uncertain, with both environmental and genetic factors playing roles. Prenatal maternal factors such as exposure to alcohol, valproic acid, carbamazepine, or isotretinoin; hyperthermia; malnutrition (especially folate deficiency); diabetes; and obesity all increase the risk of giving birth to a child who has myelomeningocele. Risks also vary by ethnicity and sex, with higher prevalences among Latino and female offspring. The risk is higher among Latino women who have recently moved to the United States than among those who are born in or have lived in the United States for several years. It is unclear what environmental factors, such as nutrition, affect the risk.

Classification

The extent of motor paralysis and sensory loss in patients who have myelomeningocele depends on the location of the defect in the spinal cord; sensory and motor function below that point typically are impaired. The standard classification lists the level(s) of the neurologic impairment such as T12 for involvement at the 12th thoracic level. However, a more functional classification encompasses three categories (Table 1). (1)

The first category includes children who have thoracic or high lumbar level lesions and who lack quadriceps strength. They require extensive bracing for household ambulation and use a wheelchair from an early age for mobility in the community.

The second category includes children who have L3 and L4 functional motor levels and good quadriceps strength and medial hamstring function but lack gluteus medius and gluteus maximus function. This condition results in a Trendelenburg gait that stresses the medial aspect of the knee joint and is energy-inefficient. Such children are advised to use

*Upstate Medical University, Syracuse, NY.

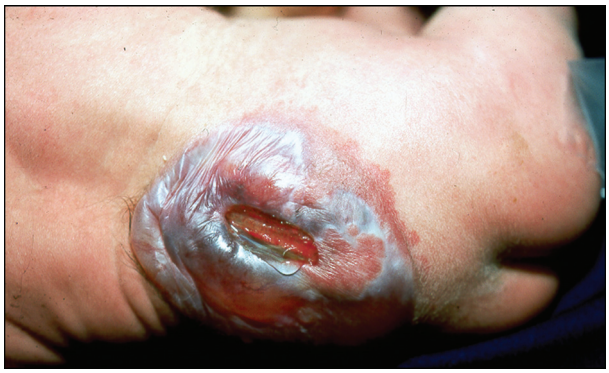

Figure 1. Neonatal open lumbosacral myelomeningocele lesion that is leaking cerebrospinal fluid.

forearm crutches for a swing-through gait and ankle-foot orthoses (braces) to maintain alignment and to improve functional mobility and prevent knee joint arthritis in later years. Although most children in this functional motor group retain community ambulation in adulthood, most eventually prefer to use a wheelchair for longer distances.

The third category includes children born with sacral level lesions. These children can be subdivided into those who have higher sacral levels and weakness in the ankle plantar flexors and who have lower sacral levels and some weakness of the intrinsic muscles of the foot but good gastrocnemius strength and no discernible gait abnormality. All children in this category have an excellent prognosis for lifelong ambulation. Those who have weak plantar flexors benefit from ankle-foot orthoses but generally do not need to use crutches.

Table 1. Functional Levels in Children Born With Myelomeningocele (1)

Level	Implications for mobility
High lumbar–thoracic	Can walk for short distances using long leg (high) braces. By early adolescence, most use wheelchair for mobility.
Low lumbar	Can walk with short leg braces and forearm crutches.
High sacral	Can walk with a gluteal lurch using braces to stabilize the ankle and foot. Walking ability usually is retained through adolescence.

Consequences

Brain Abnormalities

Abnormalities of the brain (Fig. 2) include hydrocephalus, Chiari type II malformation, agenesis of the corpus callosum, and other anomalies such as hypoplasia of the cranial nerve nuclei and diffuse microstructural anomalies. Such defects are associated with learning disabilities, including nonverbal learning disorder, attention-deficit/hyperactivity disorder (ADHD), and problems with executive function. Unless ventriculitis has occurred, most children affected by myelomeningocele have overall intelligence in the normal range. Hydrocephalus occurs in 60% to 95% of children who have myelomeningocele and is more common with higher-level lesions. Hydrocephalus usually is treated with a ventricular shunt, although third ventriculostomy (surgical placement of an opening between the third and fourth ventricles) is being used more frequently in some settings (eg, developing countries). However, shunts fail frequently or become infected and can lead to life-threatening increases in intracranial pressure. Table 2 shows the signs and symptoms associated with shunt failure. *Myelomeningocele is a nonprogressive condition.* Thus, children who exhibit any neurologic deterioration should be evaluated for a treatable cause, especially ventricular shunt failure.

Almost all children who have myelomeningocele above the sacral level also have a Chiari type II malformation of the brain. In this abnormality, the brainstem and part of the cerebellum are displaced downward through the foramen magnum into the upper cervical spinal canal rather than remaining within the skull.

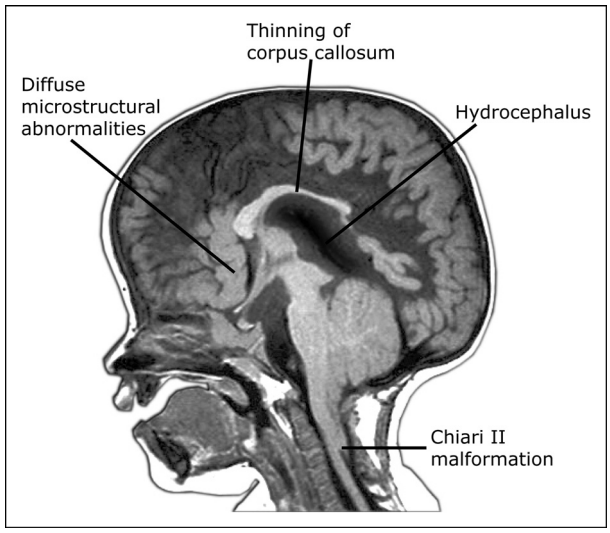

Figure 2. Cranial magnetic resonance imaging showing abnormalities commonly found in myelomeningocele.

Table 2. Signs and Symptoms of Increased Intracranial Pressure (Ventricular Shunt Failure)

Typical

- Headache, irritability, lethargy
- Vomiting, anorexia
- Increasing head circumference, bulging anterior fontanelle
- Esotropia, diplopia, paralysis of upward gaze

Subtle or Confusing

- Cognitive change, such as decline in school performance
- Moodiness, change in personality
- Signs of Chiari malformation (Table 3)
- Signs of tethered spinal cord (Table 4)

Symptoms and signs of Chiari II malformation are shown in Table 3. New-onset Chiari symptoms often are related to increased intracranial pressure, as from ventricular shunt failure. Subtle symptoms such as obstructive sleep apnea also can occur and should be evaluated promptly with otolaryngologic consultation and sleep study.

Strabismus occurs in about 20% of children who have hydrocephalus and myelomeningocele. Many children who have myelomeningocele and hydrocephalus receive a diagnosis of a learning disability. They often have impairments in executive function that have an impact on education, social, and self-help skills. Executive function skills include the abilities to plan, initiate, sequence, sustain, inhibit competing responses, and pace work. Deficits in attention frequently are diagnosed in these children but do not respond as well to stimulant medications as children who have ADHD without myelomeningocele. Precocious puberty is a common occurrence in females who have myelomeningocele with hydrocephalus and is believed to be related to a disorder of the hypothalamus.

Approximately 15% of individuals born with myelomeningocele develop a seizure disorder (epilepsy). The seizures usually are generalized tonic-clonic and respond well to antiepileptic medications. Sometimes, new-onset seizures may occur as a manifestation of shunt infection or, rarely, shunt failure.

Table 3. Signs and Symptoms of Chiari II Malformation

- Dysphagia, including difficulty swallowing, poor or prolonged feeding, pooling of oral secretions and feedings, cyanosis, choking, coughing, and nasal regurgitation
- Hoarseness or stridor
- Aspiration with or without pneumonitis
- Breath-holding spells
- Apnea, including disordered breathing during sleep
- Stiffness, weakness, and decreased function in the arms
- Opisthotonos

Spinal Cord Abnormalities

Spinal cord malformation results in loss of both sensation and motor function. Loss of sensation leads to decubitus ulcers and damage from burns and abrasions. If not diagnosed early, decubitus ulcers may require prolonged hospitalizations for debridement, pressure relief, and plastic surgery. Common pressure points include the ischial tuberosities (suggesting inadequate pressure relief from wheelchair cushions), the coccyx (seen with poor mattresses or with prolonged bedrest), and the bony prominences of the feet and ankles (due to poorly fitting shoes or orthoses). Ulcers that are continuously contaminated by stool or urine do not heal well. A deep ulcer may lead to osteomyelitis. The most important method of promoting healing is to remove the pressure from the ulcer.

Loss of efferent nerve stimuli results in neurogenic bowel and bladder. Loss of motor function in the lower extremities leads to loss of mobility and to musculoskeletal deformities such as flexion contractures or torsion of a bone. Vertebral bony anomalies (as well as paralysis) lead to scoliosis and kyphosis. Regular evaluations by an orthopedist to determine joint mobility, positioning of the extremities, and spinal curves are indicated. Loss of efferent fibers in sexually mature males leads to impotence as well as to retrograde ejaculation, in which the semen goes into the bladder rather than being ejected out of the urethra.

The spinal cord in most children who have myelomeningocele is positioned lower in the spinal column than in other individuals. It may be attached to surrounding tissue or split (diastematomyelia or diplomyelia). These defects can lead to subsequent neurologic damage as the child grows. Table 4 lists the signs and symptoms of tethered spinal cord. As with the Chiari malformation, the new onset of tethered cord symptoms often is related to increased intracranial pressure, as from ventricular shunt failure. Because most children who have myelomeningocele have a low-lying conus, magnetic resonance imaging (MRI) of the spinal cord may not be helpful, and

Table 4. Signs and Symptoms of Tethered Spinal Cord

- Weakness in lower extremities, deterioration of gait
- Pain in back or legs
- Atrophy of muscles in lower extremities
- Sensory loss or change in lower extremities
- Change in deep tendon reflexes
- Change in bowel or bladder function
- Local swelling in the back
- New orthopedic contracture, such as pes cavus or foot- or leg-length discrepancy
- Rapidly progressive scoliosis
- New decubitus ulcer

the presence of a low-lying conus does not indicate symptomatic tethering. MRI, however, may show an abnormality such as a lipoma, syrinx, or split cord. Serial neurologic evaluations, muscle strength testing, and urodynamics can be helpful in diagnosing tethered spinal cord.

Bowel and Bladder Dysfunction

Bladder and bowel dysfunction occur in virtually all children who have myelomeningocele, even in those who have sacral level lesions. Bladder dysfunction can be categorized as failure to store urine or failure to empty urine and may be related to the bladder, external sphincter, or both. Failure to empty urine increases the risk of urinary tract infections. The combination of a tight bladder outlet and increased tone in the bladder leads to urinary reflux and hydronephrosis. Urinary reflux plus infection leads to renal damage and failure over time. Renal failure remains a common cause of death in adults born with myelomeningocele. Bladder pressure is reduced by using daily clean intermittent catheterization (CIC). The urinary tract is imaged ultrasonographically at regular intervals beginning in infancy. Bladder function also may be evaluated by using cystometrography (urodynamics). In some infants who have increased bladder pressure, however, CIC is not successful and a vesicostomy (a surgical opening through the abdominal wall into the bladder) is performed, allowing urine to drain directly into the diaper. In older children and teens, the bladder may be augmented, as by opening the bladder and sewing on a piece of small intestine. Children born with myelomeningocele typically receive regular care from a urologist.

Neurogenic bowel initially is managed with timed toileting, a diet high in fiber, and the use of stool softeners or laxatives; regular enemas delivered through a balloon or wedge catheter may be attempted. Constipation is common, and rectal prolapse may occur. If conventional measures do not work in managing incontinence and constipation, a surgical procedure, the antegrade colonic enema, may be indicated. In this procedure, a hole is made in the abdominal wall that can be connected directly to a hole in the colon. A gastrostomy feeding button is placed through the hole on the abdominal wall into the colon. Alternatively, a piece of appendix may be interposed between the bowel wall and the intestine. In either case, daily boluses of liquids are pushed into the colon, evacuating it and preventing fecal incontinence.

Latex Allergy

For reasons that are not completely clear, more than 50% of children who have myelomeningocele develop an allergy to latex in childhood. The risk of allergy increases as the child gets older and may be life-threatening. Avoiding latex-containing products from the time of birth is recommended.

Prenatal Screening and Newborn Care

Prenatal screening can identify many affected fetuses. Between the 16th and 18th weeks of pregnancy, maternal serum is analyzed routinely for alpha-fetoprotein (AFP), which is elevated when the fetus has an open myelomeningocele. If the AFP value is abnormal, high-resolution ultrasonography is used to detect specific abnormalities of the fetal head and back. If myelomeningocele still is suspected, amniocentesis is performed to analyze amniotic fluid concentrations of AFP and acetylcholinesterase, an enzyme found in cerebrospinal fluid. At the same time, chromosomes are evaluated to rule out conditions associated with myelomeningocele, such as trisomy 13. A woman carrying a fetus that has a myelomeningocele should be referred for delivery to a tertiary medical center that specializes in the care of these children.

The newborn who has myelomeningocele should receive routine care in the delivery room, but the lesion on the back should be protected from contamination. To prevent infection of the central nervous system, the open lesion is covered and the infant is given parenteral antibiotics until surgical closure. Typically, the lesion on the back is closed surgically within 72 hours. The infant is evaluated neurologically to determine the level of lesion.

Orthopedic evaluation is performed to identify curvatures of the spine, such as kyphosis, and joint contractures, such as talipes equinovarus (clubfoot). Urologic

evaluations, including measurement of creatinine and blood urea nitrogen and renal ultrasonography, are used to evaluate renal function and to rule out hydronephrosis or other renal anomalies, such as horseshoe kidney. Voiding cystourethrography is obtained to evaluate for reflux and to help determine whether antibiotic prophylaxis is indicated. Cranial imaging using ultrasonography or MRI is performed to evaluate for hydrocephalus and structural evidence of the Chiari II anomaly. Daily measuring of head circumference, monitoring of neurologic signs such as irritability, and repeated cranial imaging are used to guide the management of hydrocephalus. If the cerebral ventricles are enlarging significantly, surgery to shunt the ventricles is performed. Family support, financial counseling, and genetic counseling are provided.

Ongoing Surveillance and Care

Myelomeningocele has been described as the most complex birth defect compatible with life. Figure 3 shows the most common findings and their relationship to brain and spinal malformations. Affected children need complex, coordinated care to optimize their health and well-being. After the newborn period, the child should be followed on a regular basis and monitored over time in the medical home. Periodic evaluations include:

- Detailed neurologic evaluations of upper and lower extremities. Deterioration of upper extremity function may indicate worsening hydrocephalus due to a problem with the ventricular shunt, a problem with the spinal cord or brainstem (eg, syrinx), or posterior fossa compression from the Chiari malformation. Deterioration of lower extremity function or bowel and bladder function may be a sign of worsening hydrocephalus or tethered spinal cord.
- Monitoring for signs and symptoms of increased intracranial pressure from shunt failure (Table 2). Early recognition of shunt failure or infection is critical.
- Assessment of growth, nutrition, and physical fitness. In older children, arm span may be substituted for height. Head circumference should be measured regularly to evaluate for increasing ventricular size. Children born with myelomeningocele are at increased risk for obesity. Emphasis should be placed on increasing physical activities such as aerobic conditioning (eg, wheelchair sports) and strength training (eg, lifting free weights). The child's adaptive physical education program should be evaluated to ensure that he or she is truly participating in sufficient activities. Exercise should be combined with a healthy balanced diet; dietary restrictions of calories may be indicated, especially because individuals who have paraplegia need fewer calories to grow and maintain normal weight. Affected children also are likely to have short stature due to a combination of failure of leg growth, spinal curves, and occasionally, a deficiency of growth hormone.
- Developmental surveillance and screening, including evaluation of gross motor, fine motor, social, language, and cognitive abilities. Infants born with myelomeningocele should be referred to their local Early Intervention Program by 6 months of age. Children exhibiting development outside the expected norms for this population should have formal psychometric evaluations. Before school entry, all children who have myelomeningocele should undergo a formal psychoeducational evaluation to determine whether they have a learning disability and assess their strengths and weaknesses.
- Assessment of bladder and bowel function. Urinary tract infections (UTIs) occur commonly in children

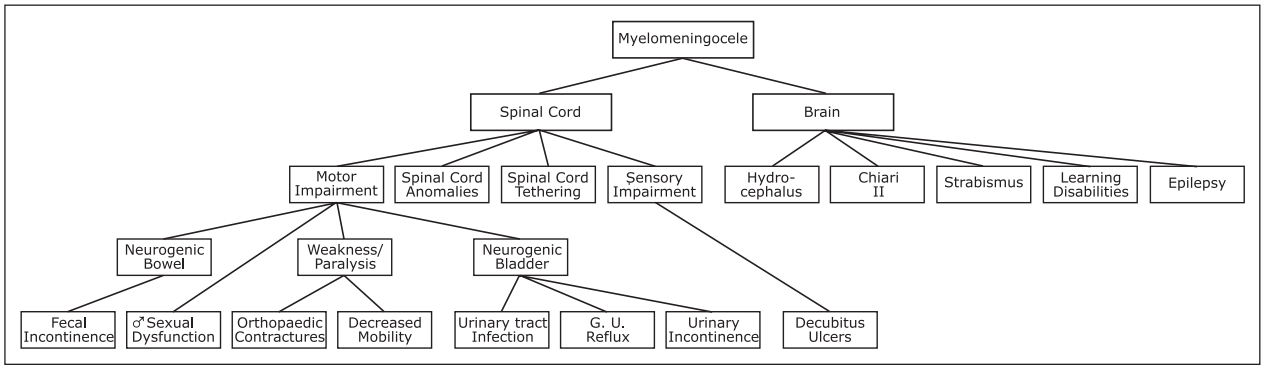

Figure 3. The most common findings in children born with myelomeningocele and their relationship to brain and spine malformations. GU=genitourinary

and teens who have myelomeningocele. However, urine cultures may reveal greater than 100,000 organisms/mL in asymptomatic individuals. Increasingly, these asymptomatic infections are not being treated with antibiotics. However, children who have positive urine cultures and symptoms such as fever, chills, and change in urine function (eg, increased leaking) should be treated with appropriate antibiotics; therapy may require parenteral antibiotics. The use of prophylactic antibiotics to prevent subsequent UTIs is controversial. Regular follow-up with a urologist is indicated. Bowel function, including constipation and continence, are evaluated at every visit.

- Assessment of vision. All children born with myelomeningocele should see an ophthalmologist early in life.
- Assessment of independent skills (activities of daily living). Discussions of expectations for adult independence should be undertaken, even in preschool-age children.
- Dental screening. All children should be evaluated regularly by a dentist. Because of the complexity of myelomeningocele, dental care may be neglected.

Some children who have myelomeningocele have unrecognized pain that can affect their quality of life and be associated with depression.

- Assessment of skin integrity. Potential pressure areas such as the buttocks, perineum, back, and lower extremities are evaluated, especially in the older child and adolescent.
- Pain monitoring. Some children who have myelomeningocele have unrecognized pain that can affect their quality of life and be associated with depression. Better recognition and treatment of pain may significantly improve quality of life and help identify treatable conditions such as tethered spinal cord or overuse syndromes (eg, shoulder pain in a person using a wheelchair).
- Assessment of bone density. Children and teens who have myelomeningocele are at increased risk for osteopenia and pathologic fractures, especially after prolonged immobility due to circumstances such as recovering from surgery. Ensuring adequate intake of calcium and vitamin D should be part of routine care. Because no current evidence-based treatment to prevent or improve osteopenia in this population exists, obtaining routine bone density studies is controversial.

Transition to Adult Care

The primary care pediatrician has an important role in helping the adolescent who has myelomeningocele make the transition to adult care. The transition to adult-oriented care is influenced by the severity of the condition, the support system, and the availability of community resources. To be effective, formal transition planning should begin when the adolescent is no older than 14 years of age. Information should be provided to the adolescent in a manner that is appropriate for his or her developmental abilities, especially because most have some form of learning disability. Before the transition occurs, the adolescents should incorporate self-care skills into their daily lives, as in taking responsibility for their medications and procedures (such as catheterization). Individuals who have myelomeningocele may need social skills training to help them with assertiveness, including speaking on their own. The pediatrician helps to ensure that medical information is transferred to the adult-oriented clinicians. For adolescents and young adults who have significant cognitive delay, issues of guardianship should be addressed before the person legally becomes an adult.

Sexuality and reproductive health should be discussed routinely and referrals made to a urologist, if indicated, for discussion of medical and surgical treatment of neurogenic impotence and retrograde ejaculation. Sexually active young women should be counseled to check the skin of the perineal area and to use water-soluble lubricant during intercourse. The primary care pediatrician also should be familiar with the Centers for Disease Control and Prevention (CDC) recommendations for prenatal folate consumption to prevent recurrence of neural tube defects in babies of young women who have spina bifida as well as couples who have borne a child who has a neural tube defect. The recurrence risk is 3% to 5%, which can be reduced to less than 1% with administration of 4 mg folate daily in the periconceptual period (for 1 to 3 months before conception and during the first trimester). Pediatricians also play an important role in educating the general population about CDC recommendations for daily folate

consumption to prevent birth defects. Brochures in both English and Spanish can be downloaded from www.SBAA.org.

Family Support

A child born with myelomeningocele places psychological, financial, and social stress on the family. In the newborn period, parents are faced with the loss of the expected normal child; the presence of a child who has an unanticipated problem and uncertain future; and the need to make immediate medical decisions, including giving consent for surgery. Throughout childhood and adolescence, the family must face the daily burden of care, which requires a great commitment of time and energy. The care of a child who has myelomeningocele also is expensive both directly in terms of medical and related expenditures and indirectly in terms of costs for transportation, time lost from work, illnesses, hospitalizations, structural changes to the home, special clothing, and special child care arrangements. Family support, including financial counseling, and family-centered care should be provided. This care includes evaluation of the parental relationship with the child and the presence of stress and social supports. Determining the functioning of siblings also is important. Families may benefit from respite care. Children who have myelomeningocele and their families also may benefit from referrals to community resources, including parent support and advocacy groups and community programs of recreational and adaptive sports.

Delivery of Care

Children born with myelomeningocele typically receive care from multiple clinicians, including orthopedists, orthotists, urologists, and neurosurgeons, as well as physical, occupational, and sometimes speech therapists. They usually receive services through Early Intervention or school-based programs. Parents, teachers, and therapists often approach pediatricians for advice on medical and habilitative care. To optimize the care of children who have myelomeningocele, medical homes should collaborate and coordinate with medical and nonmedical providers in the community. Primary care physicians should facilitate linkages to appropriate services. Communication and accurate transfer of information, such as informing the school nurse of changes in the child's seizures, improves care and outcomes. (2) Clinical settings should be physically accessible, and billing should reflect time spent in direct and indirect care, such as care coordination. Attention should be given to social and financial issues as well as to clinical care.

Women carrying fetuses found to have myelomeningocele should be referred for delivery to a tertiary medical center that specializes in the care of these children. All children who have myelomeningocele should be followed during childhood by a multidisciplinary team that includes experts in child development, neurosurgery, orthopedics, urology, orthotics, social work, nursing, physical and occupational therapies, and plastic surgery. Finally, the adolescent's care should be transferred to skilled adult medical practitioners.

Summary

- Based on strong research evidence, myelomeningocele is a condition that affects both the spinal cord and the brain in most children.
- Based on some clinical research, myelomeningocele is a nonprogressive condition. Thus, any neurologic deterioration should be evaluated for a treatable cause. Priority should be given to the evaluation for increased intracranial pressure by evaluating the ventricular shunt.
- Based on expert opinion, the care of children born with myelomeningocele should be provided within the medical home as well as with multidisciplinary teams of specialists. Collaboration among all providers can improve patient outcomes.
- Based on strong research evidence, the birth prevalence of myelomeningocele can be decreased by the use of folate supplementation in the periconceptual period.

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PIR Quiz

Quiz also available online at http://www.pedsinreview.aappublications.org.

1. You are seeing a 12-year-old girl who has myelomeningocele in your office for complaints of radiating back pain over the past 3 months. She has not changed her bowel or bladder management program but is now having new urinary leakage between catheterizations. The additional symptom that *most* supports your presumptive diagnosis is:

   A. Declining school performance.
   B. Deteriorating gait.
   C. Difficulty swallowing.
   D. Early morning headache.
   E. Worsening handwriting.

2. An emergency department physician calls you about her evaluation of a 14-month-old child who has myelomeningocele and a ventriculoperitoneal shunt. Results of computed tomography scan and shunt series are unchanged from previous studies, but the child is not "normal," according to the parents. Which of the following is *most* concerning for a shunt malfunction?

   A. Ankle clonus.
   B. Limited upward gaze.
   C. Patulous anus.
   D. Pes cavus.
   E. Swelling on back.

3. During a neonatal consultation for an infant born with a neural tube defect (neurologic level L3), the family asks if their child will walk. The *most* accurate statement regarding this child's future walking ability is that she will walk:

   A. Independently.
   B. With ankle bracing.
   C. With high leg bracing.
   D. With hip bracing.
   E. With low leg bracing and forearm crutches.

4. You are seeing an 8-month-old boy who was born with myelomeningocele for a routine follow-up evaluation in the clinic. His parents have been reading on the Internet about the Chiari II malformation and ask if their son has this problem. The clinical finding *most* commonly associated with Chiari II malformation is:

   A. Bulging fontanelle.
   B. Leg pain.
   C. Scoliosis.
   D. Stridor.
   E. Urinary incontinence.

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