**Myelomeningocele**

*With emphasis on mid to lower lumbar lesions*

**Introduction**

Myelomeningocele (MMC), a type of spina bifida aperta, is a congenital neural tube defect of the central nervous system1. It is the most severe form of spina bifida1. This diagnosis occurs when the meninges and spinal cord protrude through open or incomplete vertebral arches1. This leads to many impairments and disabilities, including deficits in mobility and strength, bladder and bowel function, cognition, and behavior, among others2,3.

**Epidemiology and Pathophysiology**

An estimated one to five babies per every 1,000 live births in the U.S. are born with MMC4. Further, if a woman has a child with MMC, there is a 3% to 5% chance of having another child with MMC4. The pathology of this condition is not completely understood. It is likely influenced by a combination of environmental and genetic risk factors, notably a lack of folic acid or a family history of neural tube defects. There also appears to be differences based on ethnicity, as the MMC rate in African blacks is 1 in 10,000 live births, compared to Celts who have an incidence rate of 1 in every 80 live births5. Maternal alcohol intake and drug abuse may also contribute to the development of MMC5.

During the embryonic period, or more specifically at the third week of pregnancy, failure of neural tube and mesenchymal closure exposes these tissues to the environment of the uterus, and trauma or exposure to amniotic fluid can lead to damage of the cord throughout gestation1,4. This results in a failure of the vertebral arches to form the spinal canal, allowing the meninges and spinal cord to protrude outwards1. The condition is typically diagnosed by 18 weeks of gestation—and 23% of these pregnancies are voluntarily terminated5. However, despite the challenging complications that children with MMC will present with, medical advancements have led to 90% of those with spina bifida living into adulthood6.

**Disease Progression and Impairments**

Given the complex nature of this condition, it is important to consider the many impairments, or effects on body structure and function, that a child will present with. These impairments will affect activity and participation for that child.

*Musculoskeletal Impairments*

Musculoskeletal deformities are another major impairment seen in children with MMC. Muscle weakness in utero causes abnormal positioning and consequential deformities, including clubfoot, equinovarus, calcaneovalgus, and vertical talus7. Throughout childhood, more deformities may arise from normal growth combined with muscle imbalances and postural effects of gravity7. As the child ages and begins to ambulate, abnormal foot positioning puts the child at risk for skin breakdown7. Specifically looking at mid to low lumbar lesions levels (L3-L5), these patients will often present with increased lumbar lordosis, hip and knee flexion contractures, genu and calcaneal valgus malalignment, and pronated feet when weightbearing7. These children will ambulate with a crouch gait as a result of the muscle weakness and orthopedic deformities7. In turn, this type of gait can lead to further flexion contractures, and can negatively impact the child’s ability to function and participate in their environment7.

Along with hip and knee flexion and foot deformities, children with MMC will often present with postural deficits including a forward head, rounded shoulders, anterior pelvic tilt, rotational deformities of the lower extremity5. Scoliosis, kyphosis and lordosis are common postural deformities, with higher lesions being most affected5. These deformities will progress with age, placing the child at risk for decreased chest wall expansion, decreased lung ventilation and increased respiratory infections5. The deformities may also limit the child’s ability to sit or ambulate7.

These musculoskeletal deformities cause poor joint alignment and increased joint stresses during activity5. They can also lead to decreased range of motion (ROM) 5. Decreased ROM can limit a child’s ability to perform ADL’s, get in and out of bed, and ambulate5. ROM deficits that lead to contractures can cause pain and discomfort, especially when lying down, as well as a negative body image5.

The extent of motor paralysis will depend on the level of the spinal cord lesion8. Lesions at L3 and L4 will typically result in adequate quadriceps strength and good medial hamstring function, but will lack function of the gluteus medius and maximus8. This can result in Trendelenburg gait, valgus stress through the knee, and an overall inefficient gait pattern8. Patients with these mid to low lumbar lesions can typically ambulate with ankle foot orthoses and forearm crutches, which will allow for a more efficient swing through gait and improved function, as well as prevention against future arthritis8. These patients will typically use a wheelchair for longer distance mobility8. Overall, 30% to 50% of those with MMC will be functional ambulators, while 30% to 40% will mostly use a wheelchair for mobility9.

The majority of children with MMC will demonstrate deficits of the upper extremity, including weakness, paresthesia, decreased dexterity and numbness10. The motor deficits are related to the Arnold Chiari II malformation, and can result in limb dysmetria, disdiadochokinesis, and decreased functional independence10.

Osteoporosis and osteopenia are more common in patients with MMC9. Decreased bone density can be affected by many different functional and medical factors, such as ambulatory vs. nonambulatory status, as well as neurologic level, bladder control and risk of metabolic acidosis, and vitamin D deficiency9. A fracture in this population can be especially debilitating, as it can lead to a spiraling effect of immobilization, further decreased bone density, and an ensuing fracture9.

*Neurologic Impairments*

Neurologic impairments can further affect the level of activity and participation for patients with MMC Hydrocephalus and the Arnold Chiari II malformation are common impairments seen in children with MMC1. Hydrocephalus, which is caused by enlarged cerebral ventricles and increased cerebrospinal fluid (CSF), affects more than 85% of patients with MMC1. About 80% of these patients will need a ventricular shunt in order to manage the CSF volume and help prevent further intellectual and neurologic impairments1. Arnold Chiari II malformation occurs when the hindbrain descends through the foramen magnum, which subjects the child to brainstem compression1. Thirty percent of children will only have mild symptoms, while 5% will demonstrate “Chiari crisis”, characterized by stridor, failure to thrive, a weak cry, apnea and cyanosis7. Brainstem compression will cause cerebellar dysfunction, complications from the medullary respiratory center, involvement of cranial nerves 9 and 10, and hydrocephalus1. It is important to address this via surgery and a ventricular shunt, as this is a life threatening condition1.

 An estimated 15% to 20% of patients with MMC will experience seizures during childhood, likely due to shunt malfunction and infection7. Oculomotor disorders, such as strabismus and visual tracking deficits, can decrease a patient’s ability to interact and focus on their environment7.

Neurogenic bowel and bladder pose serious health and social implications to MMC patients. Interruption of the S2 through S4 spinal segments and decreased anal sensation can cause the anal sphincter to become spastic, flaccid, or hypotonic, leading to an inability to control the bowels5. Fifty percent of those with MMC will present with a detrusor dyssynergy, where the bladder contracts but the external sphincter is unable to relax and allow excretion of the urine11. If unmanaged, this detrusor dyssynergy and an overactive pelvic floor can lead to serious renal damage, which can start in the first 6 months of life11. These patients can experience high bladder pressures, putting them at an increased risk for urinary tract infections11. Clean intermittent catheterization is instituted immediately after closure of the open back lesion. Children with lower lumbar lesions should be self sufficient by 6 to 8 years of age, and a timed voiding schedule will help to decrease any unwanted leakages5,11. Urinary management is a life long necessity, with aims of preserving renal function, improving quality of life, promoting urinary dryness and independence with bowel and bladder management, and improving self image and sexuality11.

Sensory loss is common, though it is often asymmetrical in presentation7. Patients with lower lumbar lesions will often lack sensation around the perineum, anus and feet7. Sensation deficits do not necessarily correlate to the dermatome of the lesion level, so it is important to perform an in depth evaluation of sensation. More than 50% of patients with MMC will develop a latex allergy during childhood8. This can be life threatening, and it is recommended to avoid latex products from the time of birth8.

Patients with midlumbar lesions may present with spasticity in their lower extremities. Increased tone and spasticity of the hip and knee flexors can further limit a child’s ability for proper positioning and ability to sit or ambulate12. Spasticity can sometimes contribute to hip dislocation or subluxation due to an increased pull from the hip adductors and decreased influence from the hip extensors and abductors in these patients12. Likely, spasticity may be present in the upper extremities, leading to decreased ability to perform ADL’s13.

Individuals with MMC typically have cognition deficits and intellectual disabilities7. Executive function, attention, organization, abstract reasoning, and visual perception are often affected7. Complications with the ventricular shunt can further affect intellectual disability7. Verbal reasoning skills are typically better than nonverbal reasoning skills, and lower level lesions (lumbar and sacral) tend to show less severe deficits compared to higher level lesions7.

*Integumentary and Cardiopulmonary*

Due to the sensory deficits in this population, these patients are at increased risk for decubitus ulcers, burns and abrasions8. The ischial tuberosities, the coccyx, and bony protrusions on the feet and ankles are the most susceptible pressure points8. Pressure relief is an important part of prevention, as ulcers will lead to prolonged hospital stays and possible osteomyelitis8. Poor bowel and bladder management will place these patients at further risk for maceration and skin breakdown8.

As mentioned, the high prevalence of spinal deformities can compromise ventilation and put the child at risk for respiratory infection5. Central respiratory function is the single most frequent cause of death in patients with MMC14. Inactivity, decreased aerobic fitness, and obesity may increase the risk for cardiovascular disease in this population, however research is somewhat inconclusive in this area15. Researchers did find that nonambulators were more at risk for CVD if they had a cluster of risk factors, and higher aerobic fitness tended to result in no CVD risk15.

**Activity and Participation**

Due to the complex nature of body structures and function in patients with MMC—including motor and sensory function, orthopedic deformities, respiratory involvement, and cognition deficits—activity and participation are greatly affected. At an early age, physical impairments may result in decreased mobility (*activity*), and decreased curiosity and initiative to explore and participate in the child’s environment5. This can lead to passively dependent behavior that can stay with the child as he ages5.

In patients with lower lumbar lesions (L4/L5), scoliosis and lordosis(*body structure/impairment*) may lead to activity limitations such as decreased sitting balance21, and a delay in independent transfers and ambulation. This can potentially affect a child’s participation in school and hobbies; it may limit a child’s participation in sitting at the lunch table and eating with their peers, or may further limit the ability to walk from class to class with their school mates. Decreased weakness and dexterity in the upper extremity may limit independence in fine motor movements or ADL’s such as feeding and dressing (*activity*), leading to a decreased ability to fully participate at work, school, or to live alone17.

One study found that wheelchair dependence and incontinence were activity limitations that significantly affected social participation in young adults with spina bifida16. Further, physical impairments (e.g. paralysis, contractures, hip dislocations) led to wheelchair use and an inability to use public transportation or to access certain buildings (*activity*)16, while speech and comprehension impairments led to decreased communication skills(*activity*)17. In turn, this kept the subjects from taking part in social activities with their friends and family (*participation*)16. Cognitive impairment and executive dysfunction, particularly related to language, attention, memory and perception, can lead to activity limitations such as decreased problem solving, conversation skills, and ability to read22. In turn, this can limit the child’s ability to succeed at school, be comfortable in social settings, perform self care, or hold a job to financially provide for oneself22.

Physical impairments also negatively affected the patient’s ability to work a job, participate in leisure activities16, or obtain a driver’s license17. Participants with spina bifida who had MMC, a higher lesion level, or hydrocephalus had greater participation restrictions in terms of needing special secondary education, being unable to work a regular job, not having a partner for support, and requiring special care for living arrangments16. It was interesting to note that the ability to participate in volunteer activities was a protective factor against negative long term outcomes, as it led the participant to feel more empowered and confident in themselves17.

Researchers also found that environmental factors such as a lower family income and lower education level of the parents were associated with decreased social participation for the child with spina bifida17. Parental attitudes may also play a role2. Personal factors such as self-worth and self-esteem will effect the quality of life for patients with spina bifida2. Some research has demonstrated an inverse relationship between severity of disability and the emotional aspect of quality of life2; those with less severe physical impairments (e.g. are able to ambulate) demonstrated greater emotional problems compared to those with more severe physical impairments2. It was hypothesized that the higher functioning individuals spent more time with peers, but were more distressed about incontinence and urologic issues2.

**Practice Pattern23**: 5C, Impaired motor function and sensory integrity associated with nonprogressive disorders of the central nervous system—congenital origin or acquired in infancy or childhood

**Examples of Outcome Measures for Body structure/function, Activity, Participation, and QOL23**

1. International Myelodysplasia Study Group Criteria for Assigning Motor Levels
2. PEDI
3. FAM
4. Bayley Scales of Infant Development
5. Beck Depression Inventory
6. SF-36
7. Childhood Health Assessment Questionnaire
8. Strength and ROM tests
9. Head circumference
10. Clinical Test of Sensory Integration for Balance, Pediatric
11. Hydrocephalus Outcome Questionnaire
12. Oxygen Consumption, heart rate
13. Adolescent Self Management and Independence Scale
14. Spina Bifida Health Related Quality of Life Scale
15. WeeFIM
16. Harter Self-Perception Profile for Adolescents
17. Health Utilities Index Mark 3
18. Barthel Index
19. Acute Care Index of Function
20. Craig Handicapped Assessment Reporting Technique
21. Fatigue Severity Scale
22. Toddler and Infant Motor Evaluation
23. 6MWT
24. Gait speed

**Focused Intervention**

Given the varying impairments and activity limitations in this population, it will be necessary to use many different interventions. Interventions for ambulation in children with lower lumbar lesions was something of particular interest to me. There is so much research that is being done on treadmill training in stroke patients, and it was very interesting to learn that treadmill training is also being used in the spina bifida population. In children with lower lumbar lesions, ambulation often becomes too strenuous and inefficient during the adolescent years, making it difficult to keep up with peers18. Muscle weakness in the lower extremities results in a strenuous gait pattern, and higher energy expenditure causes fatigue and decreased aerobic fitness18. Because of this increase in energy expenditure, researchers found that ambulation eventually decreases with age18.

Considering the effects that decreased ambulation can have in terms of participating in leisure, work, social, or even ADL activities, it is an important area to address, particularly for children with lower lumbar lesions who are capable of ambulating. Researchers recruited 41 ambulatory children with spina bifida ages 6 to 18 years old to assess the effect of a 12 week treadmill training program on aerobic fitness and ambulation18. The experimental group completed a treadmill program that used varying increases in speed intervals based on peak heart rate and the child’s baseline speed during the 6 Minute Walk Test18. The subjects began with three 6 minute repetitions on the treadmill, and worked up to three 10 minute repetitions at varying speeds18. A target heart rate of 60% to 70% of max heart rate was used based on recommendations for children with chronic disabilities18. Level of fatigue and rating of perceived exertion influenced the change in speed intervals throughout the 12 week program18. Those in the control group continued with their regular care and normal physical activity routines18.

After 12 weeks of training, the experimental group demonstrated an increase of 38.7 meters during the 6MWT, while the control group showed a decrease of 2 meters18. Walking speed and VO2 consumption also significantly increased in the experimental group compared to the controls18. There were also significant differences in self perceived change—72% and 50% of the experimental group reported positive changes in both endurance and ambulation, compared to only 5% and 0% in the controls18. The intervention group maintained these changes at a 3 month follow up18. Additionally, 30% of the experimental group continued with the treadmill program while 27% chose to start up a new activity such as biking, horseback riding, or participating in physical education in school18. Not only did the intervention result in increased endurance and ambulation, but allowed for increased confidence and social interaction with peers through increased participation18. This appears to be a promising intervention for ambulatory children with MMC that can lead to increased activity and participation.

There is some evidence to suggest that treadmill training can even be used in perambulatory toddlers with spina bifida. A case report regarding an 18 month old with an L4/5 lesion used body weight supported treadmill training combined with walker stepping in order to promote stepping skills and functional gait19. At the beginning of the trial, the child was able to pull to stand, but seemed ‘very fearful’ of taking a step19. She was unable to ambulate with a walker, even with maximum assistance19. Her treadmill starting speed was 0.2 m/s based on an algorithm used for children with Down Syndrome19. The 18 week program gradually progressed the treadmill speed, and practiced under varying conditions (e.g. barefoot, with shoes, with orthotics, with weights) 19. The toddler showed improvement in tolerance for upright activity and ambulation throughout the program—after 3 weeks of the training program, the child was able to ambulate 12 steps with the walker, and at 18 weeks she was able to ambulate 153 meters with her walker and verbal encouragement19. On average, the child spent 25 minutes per week practicing stepping throughout the trial19. This case report demonstrates how combined treadmill and overground training can lead to increased functional mobility, as well as decreased caregiver assistance. It may result in monumental functional gains that will allow the child more independence and interaction with her environment.

Based on research in infants with Down Syndrome, treadmill stepping may even benefit infants with MMC20. In infants with Down Syndrome, treadmill stepping 3 to 5 times per week led to an earlier onset of walking and an increase in step frequency20. Based on these results, a similar study using infants with MMC used a treadmill to facilitate the stepping response20. The researchers used various types of sensory input during the treadmill training, and found that visual flow input and increased friction (by covering the treadmill with Dycem) resulted in the greatest increases in stepping rate20. This increased sensory input during treadmill training could potentially be used in perambulatory infants as a way to build strength and control, and to promote an earlier onset of walking20. Further, increased sensory input could positively affect young children with MMC who are learning to ambulate.

Clinically, I can use this evidence as part of my interventions with children with MMC. If I am treating a young child who is showing developmental delay in terms of ambulation, I can use the model from the case report to create a combined treadmill and overground training program to help facilitate ambulation. The model provides clear indicators for when to progress the speed of the treadmill, when to initiate ambulating with a walker, etc. Additionally, for older children, I can use treadmill training to help increase endurance, which will have secondary benefits of improved participation and self confidence. The randomized control trial provides an outline of target heart rates and appropriate perceived exertion to ensure the child is practicing at a safe level, while also providing appropriate parameters to gradually increase the intensity and duration over time. This is a good example of a functional activity that is specific to every day activity for a child with MMC.

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