



## **Myelomeningocele (MMC)**

Patient and family information, brought to you by the Fetal Diagnosis and Treatment Committee of APSA

### **“What is myelomeningocele (pronounced my-elo-men-IN-go-seal)?”**

Myelomeningocele (MMC) is a birth defect when a child’s back (spine) does not form properly and a part of the spinal cord remains exposed and uncovered. MMC is a type of nervous system birth defect called neural tube defect (NTD). It is the most serious form of spina bifida.

The causes of MMC are not completely understood but are thought to include environmental, nutritional, and genetic factors. Spina bifida affects 1.9 to 3.4 out of 10,000 births. This condition usually forms early in the first trimester of pregnancy.

### **“How does myelomeningocele develop?”**

During normal development of a baby in the womb, a layer of tissue forms around the spinal cord and closes along the baby’s back like a zipper, protecting the spinal cord. In babies with MMC, the spinal cord and spine do not form correctly resulting in an incomplete and uncovered spinal cord. There is a sac that sticks out from the back of the baby that includes parts of the spinal cord and the fluid that bathes the brain and spinal cord called cerebrospinal fluid.

### **Signs and Symptoms - “What symptoms will my child have?”**

Depending on the location and size of the exposed spinal cord segment, the severity of injury to the spinal cord can be different and a wide range of symptoms are possible. Some children with MMC have minimal or no symptoms. Some children have bladder or bowel control problems but can walk and do other activities normally or with assistance. Some children remain with weakness or paralysis in their legs and need adaptive devices to help them walk.

NTD is often associated with abnormal flow of cerebrospinal fluid in the brain leading to hydrocephalus. Hydrocephalus is when there is excess fluid within the brain. A procedure to help improve the flow of spinal fluid may be necessary to relieve any buildup of pressure on the brain. Different procedures may be required to relieve excessive pressure on the brain which include a “ventriculo-peritoneal shunt” (a draining tube that diverts the excessive fluid from the brain to

the belly), or “endoscopic ventriculostomy” (using a small camera to create new pathways for the fluid to flow inside the brain).

NTD is often associated with other developmental anomalies of the brain. The most common is called a Chiari II malformation, which is when tissue from the back part of the brain (the cerebellum) pushes on the bottom of the brainstem. While often this does not cause severe symptoms in children, it may lead to difficulty breathing or swallowing and further brain surgery procedures to help alleviate the symptoms.

Other symptoms include:

- decreased feeling and strength in the legs
- weakness in the ankles and feet (referred to as “club feet”)
- inability to walk
- problems with control of urination and stooling with leakage or “incontinence”
- possible learning disabilities
- seizures, usually as a result of brain complications

### **Diagnosis - “What tests are done to find out what my child has?”**

MMC is often diagnosed as a part of routine prenatal care during pregnancy. A blood test showing an elevated alpha fetoprotein (AFP) level in the mother’s blood can indicate a higher risk for MMC or other nervous system birth defects. Ultrasound during pregnancy is used to check for MMC. Your doctor may also request a special scan, magnetic resonance imaging (MRI), to further check your baby’s development. Images of the spine after birth may include MRI, computed tomography (CT or CAT) scan, ultrasound, or plain x-ray.

### **Treatment - “What will be done to make my child better?”**

MMC is a defect that requires surgery to close the defect, usually done within the first 2-3 days of life. We will explain the different surgical approaches below but it will be up to your maternal fetal medicine specialists, fetal surgeon, and pediatric neurosurgeons to determine whether you are an appropriate candidate for each type of repair. Not everyone will qualify for prenatal repair and it can be based on individual centers as well as mother and fetal criteria.

#### **Postnatal repair: Surgery shortly after birth**

Goals:

- To close the MMC defect
- Protect the spinal cord from further damage and infection
- Halt cerebrospinal fluid leakage

### Procedure:

This surgery is done within the first 2-3 days of your baby's life, usually as soon as your doctors believe it is safe. During this procedure, your baby will be completely asleep under general anesthesia. This will allow the surgeons to do their delicate work on your baby's spine without worrying about the baby moving and will prevent your baby from feeling pain during the procedure. The surgeon will then close the layers of the spinal cord covering (the dura), the skin and muscles that did not close over the spinal cord. If the defect is too large to close with the baby's own tissues, a "graft" of tissue may be used to reduce tension and close the defect effectively. Occasionally, plastic surgeons may help with this closure. After surgery there will be a detailed plan to care for the needs of the baby. Your baby will stay in the NICU (neonatal intensive care unit) until well recovered.

### Benefits:

- Your child will likely reach full term before delivery, allowing their lungs and other vital organs to grow completely before birth
- Potential leakage of cerebrospinal fluid will be managed and contained shortly after birth
- The spinal cord will be covered to protect it from further damage

### Risks:

- Damage to the spinal cord which may have occurred during pregnancy that cannot be reversed after birth
- Further damage to the spinal cord during surgery may occur
- Infection of the surgical site may develop
- The spinal cord can be damaged further as the baby grows due to cord tethering

## **Prenatal (*in utero*) repair: Surgery Before the Baby is Born**

### Goals:

- To close the MMC defect early in pregnancy
- To stop the leakage of fluid from the back.
- Protect the spinal cord and brain from further damage that may occur during pregnancy
- Improve the Chiari malformation by reversing the "hindbrain herniation".

### Procedure:

A scientific landmark study called “MOMS” (Management of Myelomeningocele Study) proved the benefits of fetal surgery for some fetuses with MMC. This trial compared outcomes after fetal surgery (done before birth) to those obtained by the traditional surgery done after birth. Those fetuses who had prenatal repair required a shunt only about ½ as often as those who had the repair done after birth. Also, those fetuses who had prenatal surgery showed better mental development and walking compared to those who had surgery after birth.

We are still learning the best way to perform fetal surgery for MMC. There are different surgical approaches. Each center may do things differently and it is reasonable to ask the specialists about the options and results for each approach. In nearly all cases, the operation on the mother is similar with a c-section like incision on the abdomen. In the “open approach”, the uterus is opened, and the defect is repaired directly. In the “fetoscopic approach”, 2-3 tiny incisions are made on the uterus and the defect is repaired with long instruments and video guidance. There are advantages and disadvantages with each approach. A particular approach may be best for the particular circumstances of you and your baby. In general, it is harder to do the operation fetoscopically but the smaller incisions used in this case may allow a vaginal delivery and may pose less long-term risks to the uterus during this and future pregnancies.

Regardless of the approach, after prenatal surgery is performed, the fetus and mother will be monitored closely with ultrasound and MRI scans. The mother continues on with the pregnancy for as long as possible. The goal is to keep the baby in the womb until at least 37 weeks’ gestation before delivery. The mode of delivery can be vaginal or Cesarean which depends on the exact surgical approach and the recommendations of your fetal surgical team.

#### Benefits:

- Fetal repair of MMC decreases the risk of hydrocephalus (increased pressure in the brain) and need for a decompressive shunt.
- The MOMS trial proved that in-utero repair with the open technique allows for increased motor function and improved ability to walk independently.
- Fetal repair of MMC may also improve urinary control but there are still challenges for many babies and most will need care from a urologist into adulthood.

#### Risks:

- Incomplete closure of the defect, requiring additional surgeries after birth
- Damage to the spinal cord during surgery
- There are risks to the mother, including rupture of membranes (water breaks) causing early delivery.
- With early/premature delivery, the baby may be born before their lungs and other vital organs are mature and fully functional. If this happens, your baby may require a lengthy admission in the Neonatal Intensive Care Unit (NICU) under close supervision of a team of doctors, nurses, and other health care professionals.

- The spinal cord can still become damaged further as the baby matures and grows usually related to cord tethering.

## Home Care - “What do I need to do once my child goes home?”

Once your baby is home with you, their care plan will be similar no matter what surgery they had. They will be followed closely by a team of doctors and other medical professionals to make sure they are developing well and reaching their full potential. Often, care centers will help you organize your appointments so that you can see multiple doctors at one visit to care for your child with MMC. It is beneficial to have your baby cared for by a multi-disciplinary spina bifida team or clinic.

**Pediatrician:** Your child will be followed closely by their general doctor, called a pediatrician, like any other child and will have regular appointments to track their progress as they mature. This doctor will be your first contact when you have concerns about your child and will be able to answer many questions about your child’s condition and how their development compares to their peers.

**Pediatric Neurosurgeon:** These doctors are responsible for surgical treatments of your child. They are very knowledgeable about the options for treating your child and will carry out surgeries on their back before they’re born or while they are very young. A pediatric neurosurgeon is an expert in brain and spinal cord surgery on children and will be helpful in making sure your child is not in danger of any other neurological problems. A few common reasons a pediatric neurosurgeon may advise surgery in babies with a corrected MMC include finding a tethered spinal cord, or a blockage or leaking of the fluid that keeps the brain safe.

**Pediatric Gastroenterologist:** These specialists focus on the gastrointestinal system including the small bowel and large bowel. They are experts in managing neurogenic bowel and this is important for children who may have difficulty stooling or controlling bowel movements. They will help you come up with a plan for a good bowel regimen.

**Pediatric General Surgeon:** Pediatric general surgeons will be responsible for evaluating how well your child’s gut is working and will help to determine if they need surgeries to help them digest food better. There are some pediatric general surgeons who are expert in fetal surgery and help to organize the medical teams that perform these procedures.

**Pediatric Urologist:** Urologists focus on the urinary system, which includes the kidneys and bladder. This is important for children with MMC because they often have reduced ability to control or contract their bladder. Holding urine too long or being unable to release urine can be very dangerous for your baby’s kidneys, which are very important for their health long-term. To keep your baby’s kidneys safe, they will need to meet regularly with a urologist while they are young, and many will need close urological follow up for their entire life.

**Pediatric Orthopedic surgeon:**

Pediatric orthopedic surgeons specialize in bones, muscles and tendons that can affect joint or limbs. One important goal is to assist with walking so they can help with orthotics, bracing, and casting. They are also experts in soft tissue surgery such as Achilles tendon release and lengthening if needed.

**Physical Therapist/Occupational Therapist:** These health care providers work on strengthening and coordination with your baby and help them to engage in typical childhood milestones and experiences. When they are young, this can include helping your baby work on their neck muscles and how they move. Later in their life, they help decide the best mobility options for your child and their individual needs and focus their work on helping them participate in school and other activities.

### Long Term Outcomes - “Are there future conditions to worry about?”

Though it may seem daunting, there are many treatment options available for your baby, and your medical care team will do their best to care for you and your child. Many people with MMC or other forms of spina bifida can live long and meaningful lives, and there are many support groups available for parents and patients with MMC. More often times than not, it takes a multidisciplinary clinic and approach to care for your child. We recommend referring parents and families to the Spina Bifida Association (<https://www.spinabifidaassociation.org>) for additional information and guidance.

### By the Numbers- Important Statistics to know about MMC and prenatal repair:

- Approximately 1.9 to 3.4 children out of 10,000 are born with spina bifida in the United States.
- In adulthood, approximately 33% of those with spina bifida will require assistance with activities of daily living.
- Prenatal surgical intervention reduces likelihood of shunt placement by approximately half (40% of babies with fetal surgery require shunt compared to 82% of post-natal repairs).
- Prenatal surgery is associated with twice the likelihood that the patient will be able to walk without assistive devices (42% of people with prenatal repair have been shown to walk independently compared to 21% of those with neonatal repair).
- In-utero surgery is associated with a younger gestational age at birth, increasing the risk of other complications associated with prematurity.

### Other resources:

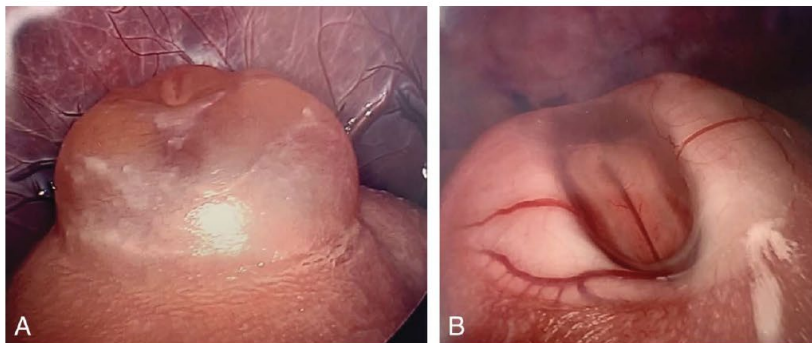
Spina Bifida Association was created in 1973 with the goal of helping patients, families, caregivers of spina bifida. Their mission includes supporting pillars of research, education & support, clinical care, network building and advocacy. Below are the website link and also the spina bifida clinical care partners.

<https://www.spinabifidaassociation.org/>

<b>Spina Bifida Clinic Care Partner Center</b>	<b>Location</b>
Children's of Alabama	Birmingham, Alabama
The University of Alabama at Birmingham	Birmingham, Alabama
District Medical Group	Phoenix, Arizona
Phoenix Children's Hospital	Phoenix Arizona
Arkansas Children's Hospital	Little Rock, Arkansas
Kaiser Northern California Regional Spina Bifida	Oakland, California
Loma Linda University Children's Hospital	Loma Linda, California
Children's Hospital Los Angeles	Los Angeles, California
Shriners Hospitals for Children, Northern California	Sacramento, California
UCSF Benioff Children's Hospital	San Francisco, California
Stanford University	Palo Alto, California
Children's Hospital Colorado	Aurora, Colorado
Connecticut Children's Hospital	Hartford, Connecticut
Orlando Health Arnold Palmer Hospital for Children	Orlando, Florida
Ann & Robert H. Lurie Children's Hospital of Chicago	Chicago, Illinois
Riley Children's Hospital	Indianapolis, Indiana
Children's Hospital New Orleans	New Orleans, Louisiana
Kennedy Krieger Institute	Baltimore, Maryland
Boston Children's Hospital	Boston, Massachusetts
Gillette Children's Specialty Healthcare	Saint Paul, Minnesota
Children's Mercy Kansas City	Kansas City, Missouri
Golisano Children's Hospital	Syracuse, New York
Duke University Hospital	Durham, North Carolina
Cincinnati Children's Hospital Medical Center	Cincinnati, Ohio
Nationwide Children's Hospital	Columbus, Ohio
Oklahoma University Children's Pediatric Urology	Oklahoma City, Oklahoma
Oregon Health and Science University	Portland, Oregon
Penn State Health	Hershey, Pennsylvania
Children's Hospital of Philadelphia	Philadelphia, Pennsylvania
University of Pittsburgh Medical Center	Pittsburgh, Pennsylvania
UPMC Children's Hospital of Pittsburgh	Pittsburgh, Pennsylvania
Monroe Carell Jr. Children's Hospital at Vanderbilt	Nashville, Tennessee
Texas Scottish Rite Hospital for Children	Dallas Texas
Cook Children's Medical Center	Fort Worth, Texas
Texas Children's Hospital	Houston, Texas
University of Texas Spina Bifida Clinic	Houston, Texas
Primary Children's Hospital/Intermountain Healthcare	Salt Lake City, Utah
UW Health/American Family Children's Hospital	Madison, Wisconsin
Children's Wisconsin	Milwaukee, Wisconsin

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**Fig. 1.** Fetoscopic images of open neural tube defect captured at the time of fetoscopic spina bifida repair at 26–27 weeks of gestation. **A.** Myelomeningocele-type open spina bifida lesion with neural placode visible at the center of the myelomeningocele sac. **B.** Myeloschisis-type open spina bifida lesion with neural placode visible at the base of the defect.

*Chmait. Prenatal Management of Spina Bifida. Obstet Gynecol 2023.*