



Spina Bifida (Myelomeningocele)

A birth defect in which an area of the spinal column doesn't form properly, leaving a section of the spinal cord and spinal nerves exposed through an opening in the back.

The most common and serious form of spina bifida is called myelomeningocele (MMC), in which part of the spinal cord and surrounding nerves push through the open bones (vertebrae) in the spine and protrude from the fetus' back. Usually, the exposed spinal cord and nerves are contained in a sac that is exposed to amniotic fluid. Continuous bathing of the fragile developing spinal cord in amniotic fluid over the course of gestation is thought to result in progressive neurologic injury.

Spina bifida occurs in 3.4 per 10,000 live births in the United States and is the most common central nervous system birth defect. Between 1,400 and 1,500 babies are born with spina bifida in the U.S. each year. Liveborn infants with myelomeningocele have a mortality rate of approximately 10 percent.

The MMC lesion can occur at any level on the developing spine, but most are found in the lumbo-sacral region. Depending on the lesion's location, MMC may cause bladder and bowel problems; sexual dysfunction; weakness and loss of sensation below the defect; orthopaedic malformations such as club feet or problems of the knees or hips; and inability to move the lower legs (paralysis). Generally, the higher the defect is located on the spine, the more severe the complications.

In many cases, the brain develops a Chiari II malformation, in which the hindbrain herniates or descends into the upper portion of the spinal canal in the neck. This herniation of the hindbrain blocks the circulation of cerebrospinal fluid, causing hydrocephalus (accumulation of fluid in the brain), which can injure the developing brain.

Prenatal Repair — A Significant Advance

Traditional surgical repair of spina bifida takes place 24 to 48 hours after birth. We have long theorized that if the defect could be repaired much earlier (*in utero*), neurologic damage might be minimized.

The Center for Fetal Diagnosis and Treatment conducted extensive laboratory research, the findings from which supported this theory. Between 1998 and 2003, the Center team performed prenatal spina bifida repair in 58 mothers and observed significant benefit in the babies.

To answer the question conclusively, a randomized, controlled clinical trial was initiated and funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development, comparing outcomes after prenatal and postnatal repair in 183 patients.

GROUNDBREAKING ANSWERS The trial demonstrated that outcomes after prenatal repair are improved to the degree that the benefits of the surgery outweigh the maternal risks. This represents the most significant advance in the history of treatment for spina bifida.

Specifically, the study found that prenatal repair resulted in:

- Reversal of the hindbrain herniation component of the Chiari II malformation
- Reduced need for ventricular shunting (a procedure in which a thin tube is introduced into the brain's ventricles to drain fluid and relieve hydrocephalus)
- Reduced incidence or severity of potentially devastating neurologic effects caused by the spine's exposure to amniotic fluid, such as impaired motor function

Prenatal repair is a complex and challenging procedure, requiring the most expert, comprehensive care for both mother and fetus. The surgical team's level of experience in all aspects of care surrounding the operation are of paramount importance.



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Evaluation at CHOP

As with other birth defects, most MMCs are discovered by routine ultrasound evaluation between 16 and 18 weeks into the pregnancy. Sometimes, a blood test known as maternal serum alpha fetoprotein (AFP) is also positive, triggering the ultrasound. Sonographic features suggestive of MMC include a cystic mass anywhere on the spine. If no skin covers the defect, AFP levels in the blood and amniotic fluid will be elevated and support the diagnosis.

Mothers referred to CHOP's Center for Fetal Diagnosis and Treatment undergo a comprehensive, one-day evaluation that includes:

- High-resolution level II ultrasound – to confirm location of the defect and to assess for any other birth defects
- Ultrafast fetal MRI – to confirm presence of the Chiari II malformation and to screen for evidence of any other neurologic abnormalities
- Fetal echocardiogram – to determine any problems with the heart

Treatment

After evaluation, you will meet with our multidisciplinary team, including a fetal surgeon, a neurosurgeon, an anesthesiologist, a maternal-fetal medicine specialist and a coordinator. The purpose of this meeting is to review your test results, confirm the diagnosis, explain treatment options and potential outcomes, and answer all of your questions.

The decision of whether prenatal or postnatal repair is appropriate is influenced by several considerations, including gestational age, the level of the MMC lesion on the spine, presence of the Chiari II malformation and a number of important maternal health factors. Prenatal repair also requires a significant commitment on the part of the mother and her support person.

The Center team helps each family fully understand the benefits and risks of treatment so they can make the best decision for their unique situation. Our team also counsels and supports families who choose to terminate the pregnancy.

Prenatal Repair If a mother whose fetus has an MMC is a candidate for prenatal repair at The Children's Hospital of Philadelphia, the surgery is performed between 19 and 25 weeks' gestation. Mothers must be willing to stay in Philadelphia with a support person for the surgery and for the duration of the pregnancy to allow close monitoring.

In prenatal repair, the mother undergoes general anesthesia, which also relaxes the uterus and anesthetizes the fetus. Fetal surgeons perform a laparotomy (an incision across the mother's abdomen), the uterus is opened and the baby's back is rotated into view. A pediatric neurosurgeon removes the MMC sac, if one is present, and closes the surrounding tissue and skin over the defect to protect the spinal cord from exposure to the amniotic fluid. The uterus and the abdominal incision are then closed. A maternal-fetal medicine specialist performs sterile intraoperative ultrasound to map the position of the placenta and the fetus, and a fetal cardiologist uses echocardiography (ultrasound to assess the fetal heart) to examine fetal heart function during the surgery. These measures are used to ensure the utmost maternal and fetal safety.

The mother usually remains in the hospital for three to five days and is on modified bed rest for two weeks after surgery to reduce the risk of preterm labor. For the remainder of the pregnancy, follow-up includes weekly visits to the Center for ultrasound monitoring and routine prenatal care. The baby is delivered by planned cesarean section at 37 weeks, if labor does not commence sooner.

Mothers who elect fetal repair will also be asked to sign a consent for postnatal follow-up of the baby by our Center. This follow-up will require the family to return to Philadelphia for evaluation when the child reaches 12 months, 30 months and five years of age. Detailed follow-up not only provides excellent care for the child, it adds to a growing base of knowledge that will benefit future generations of children with spina bifida.

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Treatment *continued*

Postnatal Repair If postnatal repair is recommended or elected, the pregnancy is monitored — often at the mother's home hospital — and a cesarean delivery is planned at CHOP at 37 weeks. Within the first 24 to 48 hours of life, the newborn undergoes repair of the defect using the same approach in prenatal repair. After surgery, the baby will receive care in our Newborn/Infant Intensive Care Unit.

Delivery for all MMC babies takes place in CHOP's Garbose Family Special Delivery Unit (SDU), allowing for the highest level of immediate care for the newborn, as well as expert obstetric services for the mother — all within a leading pediatric hospital. The world's first birth facility designed exclusively for pregnancies complicated by birth defects, the SDU is a vital part of our team's ability to provide comprehensive care for spina bifida.

Long Term: The Spina Bifida Program Children's Hospital provides comprehensive long-term follow-up for children with spina bifida, from the time of repair through adolescence. Our dedicated Spina Bifida Program's interdisciplinary team includes a pediatrician, nurse, social worker, physical therapist, neurosurgeon, orthopaedist, urologist and genetic counselor — all focused on the care of these children. As needed, other subspecialists within CHOP are consulted.

The spina bifida team works closely with each child's primary care pediatrician and school, and helps families access appropriate community services.

FETUS WITH NORMAL SPINE

▲ FETUS WITH MYELOMENINGOCELE

1. Part of the spinal cord and spinal nerves, usually encased in a sac, protrude through an opening in the back and are exposed to the amniotic fluid.
2. The brainstem (hindbrain) descends, or herniates, into the spinal canal in the neck and blocks the circulation of cerebrospinal fluid. This can cause a damaging buildup of fluid in the brain called hydrocephalus.

▲ FETUS AFTER SURGICAL REPAIR

1. Fetal surgery repairs the defect, returning the spinal tissue to its proper place and covering the opening in the fetus' back.
2. The hindbrain herniation gradually reverses after repair, and the brain stem returns to its normal position.

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Insurance and Financial Matters

Our Center's team is experienced in working with many insurance plans to facilitate care for families who seek our services. We also have resources available to assist families with travel-related and other expenses associated with coming to The Children's Hospital of Philadelphia. We are happy to discuss your individual situation and needs.

To learn more about MMC treatment at the Center for Fetal Diagnosis and Treatment, please visit fetalsurgery.chop.edu/spinabifida.

To learn more about the Special Delivery Unit and take a virtual tour, visit fetalsurgery.chop.edu.



About Our Center Expectant parents from all 50 states and more than 50 countries have found hope at the Center for Fetal Diagnosis and Treatment at The Children's Hospital of Philadelphia, recognized as a pioneer in the highly specialized field of fetal medicine. We devote every resource to providing comprehensive care focused on the best outcome for babies yet to be born.

Contact us with questions or concerns at 1-800-IN-UTERO (468-8376) or visit fetalsurgery.chop.edu.