Spina bifida

Important things to know

What is “spina bifida” or a “myelomeningocele”? Spina bifida is a birth defect in which the bones of the spinal column do not form properly. As a result the spinal cord, nerves that arise from it and their covering membrane (the dura) may be exposed to varying degrees. If the area is covered by skin, underlying tissue and muscle this is called a closed defect (spina bifida occulta). A defect where these layers are missing is called spina bifida aperta. In spina bifida aperta the dura can protrude outside the body, this is called a meningocele. When the defect is more pronounced and the dura and spinal cord, or nerves, protrude in the back of the baby this is called a myelomeningocele. Spina bifida occurs in 3-4/10,000 live births in the United States. The defect can be located anywhere on the spine of the fetus but a defect in the lower spine (lumbosacral) is the most common form.

What causes spina bifida? Spina bifida can be caused by a deficiency in folate (a water-soluble vitamin found in green leafy vegetables), a problem with the baby’s chromosomes or by a genetic disorder. If so, the baby may have additional medical problems or organ abnormalities. Spina bifida that occurs by itself and without any identifiable cause is called isolated spina bifida. Because of the range of associated conditions, it is important that spina bifida is thoroughly investigated in order to provide an accurate assessment on the likely effect on the baby.

How can spina bifida harm my baby? Spina bifida can harm the baby’s development in several ways. At the site of the defect, the exposure of the dura and spinal nerves to the amniotic fluid over the course of pregnancy can result in progressive nerve damage. This local nerve damage then results in abnormal function of all nerves below the lesion. Depending on the level of the lesion, this can produce abnormal bladder and bowel emptying, weakness or paralysis of leg movement or decreased sensation. Muscle weakness can result in abnormal position of the extremities, which can become permanent in the developing fetus. Clubfoot is an example of such a deformity. Lesions located higher on the spine generally produce a more severe clinical picture.

The inability of the spinal cord to develop normally, is the second mechanism by which spina bifida affects the babies’ development. During normal development the lower end of the spinal cord moves upward in the spinal canal as the fetal body grows. With spina bifida the position of the cord is fixed and the lower end of the brain is pulled downward as the fetus grows. The opening in the skull through is only large enough for the spinal cord and when the lower part of the cerebellum is pulled down (Chiari II malformation) it blocks the circulation of brain fluid (cerebrospinal fluid=CSF). The accumulation of CSF in the brain results in swelling of all fluid filled spaces of the brain. This produces what is known as hydrocephalus. The pressure on the brain increases and can result in further neurologic damage.

How is spina bifida detected before birth? Prenatally, spina bifida may be suspected by a number of first and second trimester markers. Ultrasound markers include a change in the head shape or the fluid filled spaces in the brain. This can be detected either at the 11-14 week, or 18 week
ultrasounds. In addition, increase in the maternal blood levels of alpha-fetoprotein is a marker for open spina bifida. Whenever such signs are observed, evaluation by a detailed high-resolution level II is necessary to confirm the diagnosis. A fetal echocardiogram and amniocentesis are typically performed to establish if spina bifida is isolated, or if there are additional birth defects or genetic conditions. Magnetic resonance imaging (MRI) relies on different imaging technology and if used in conjunction with ultrasound I can be helpful to define the effects of spina bifida on the developing brain.

How is spina bifida managed?
The principal treatment of spina bifida is the closure of the defect in the spine to restore normal anatomy as much as possible. In addition to the closure of the lesion, treatment of the secondary effects is required. This includes the insertion of a shunt to relieve the hydrocephalus.

Closure of the myelomeningocele can be performed before or after birth. The standard of care is surgical repair 24-48 hours after birth. Under very specific circumstances, and for specific types of lesions, the MOMS trial demonstrated specific benefits and risks of repair before birth. The benefits for the baby include reversal of the Chiari II malformation before birth, reduced need for shunting after birth and a lower incidence of developmental problems at 2 years of age. The risks for the mother include that of surgery, increased rate of rupture of the uterus before birth and the need to deliver all future babies by cesarean section before term.

Prenatal repair requires the highest level of care from multiple medical specialties and therefore can only be performed in specialized fetal myelomeningocele repair centers. The treatment is only offered under very specific circumstances and requires a detailed assessment to determine if all of the requirements are met.

Irrespective whether spina bifida repair was performed before or after birth, long-term care in a specialized spina bifida program is required. The spina bifida program is an interdisciplinary program including pediatricians, neurosurgeons, orthopedic surgeons, urologists, nurses, genetic counselors and social workers. The purpose of this team is to be able to cover all care aspects that may arise in children with spina bifida.

Can spina bifida be prevented?
Folic acid is normally found in a dose of 400 micrograms in prenatal vitamins. It has been shown that a much higher folate dose (4 milligrams daily) can prevent spina bifida. Because spina bifida occurs very early in embryonic development this higher dose of folate has to taken one month before planning to become pregnant. This higher dose of folate is recommended for all women that had a prior child with spina bifida as well as other conditions that are associated with a higher risk for spina bifida such as maternal diabetes, obesity and the use of antiseizure medication.