About Spina Bifida

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Spina bifida is the most common of a group of birth defects called neural tube defects (NTDs). The neural tube is the embryonic structure that develops into the brain and spinal cord. Often called open spine, spina bifida affects the backbone and, sometimes, the spinal cord. It is among the most common severe birth defects in the United States, affecting 1,500 to 2,000 babies (one in every 2,000 live births) each year. Spina bifida and other NTDs occur more frequently among Hispanics and whites of European extraction and, less commonly, among Ashkenazi Jews, most Asian ethnic groups and African-Americans.

How does spina bifida affect a child?
In the embryo, there is a tiny ribbon of tissue that folds inward to form a tube. This structure, called the neural tube, forms by the 28th day after conception. When this process goes awry and the neural tube does not close completely, defects in the spinal cord and in the vertebrae (small bones of the spine) can result. There are three forms of spina bifida:

" Occulta. In this usually symptomless form, there is a small defect or gap in one or more of the vertebrae of the spine. The spinal cord and nerves usually are normal, and most affected individuals have no problems.

" Meningocele. In this rarest form, a cyst or lump consisting of membranes surrounding the spinal cord pokes through the open part of the spine. The cyst, which can vary in size, can be removed by surgery, allowing for normal development.

" Myelomeningocele. In this most severe form, the cyst holds both the membranes surrounding the spinal canal and nerve roots of the spinal cord and, often, the cord itself. Or there may be no cyst, but only a fully exposed section of the spinal cord and nerves. Spinal

fluid may leak out. Affected babies are at high risk of infection until the back is closed surgically, although antibiotic treatment may offer temporary protection. In spite of surgery, some degree of leg paralysis and bladder and bowel control problems remain.
The severity of paralysis is largely determined by the spinal nerves involved. In general, the higher the cyst on the back, the more severe the paralysis. About 80 percent of spina bifida cysts are in the lower back's lumbar and sacral regions.

What causes spina bifida?
Spina bifida usually is an isolated birth defect. Although scientists believe that genetic and environmental factors may act together to cause this and other NTDs, 95 percent of babies with spina bifida and other NTDs are born to parents with no family history of these disorders. While spina bifida appears to run in certain families, it does not follow any particular pattern of inheritance. If one child has spina bifida, the risk of recurrence in any subsequent pregnancy is greatly increased, to about one in 40. If there are two affected children, the risk in any subsequent pregnancy is about one in 20. Spina bifida also can occur as part of a syndrome with other birth defects. Here, inheritance patterns may differ from those of isolated spina bifida.

Women with certain chronic health problems, including diabetes and seizure disorders (treated with certain anticonvulsant medications), have an increased risk (approximately 1/100) of having a baby with spina bifida.

How is spina bifida treated?
Spina bifida occulta usually requires no treatment. Meningocele, which does not involve the spinal cord, can be repaired surgically, usually with no paralysis. Most children with meningocele develop normally. However, affected children should be evaluated for hydrocephalus and bladder problems so they can be treated promptly. A baby with the most severe form of spina bifida, myelomeningocele, usually requires surgery within 24 to 48 hours

after birth. Doctors surgically tuck exposed nerves and spinal cord back inside the spinal canal and cover them with muscle and skin. Prompt surgery helps prevent additional nerve damage from infection or trauma. However, nerve damage that already has occurred cannot be reversed and limb paralysis and bladder and bowel problems usually remain.

As soon after surgery as possible, a physical therapist teaches parents how to exercise their baby's legs and feet to prepare for walking with leg braces and crutches. Studies show that about 70 percent of affected children can walk with or without these devices, although many children will require a wheelchair.

About 90 percent of children with the most severe form of spina bifida develop hydrocephalus, or fluid on the brain. When the cerebrospinal fluid, which cushions and protects the brain and spinal cord, is unable to drain normally, fluid collects in and around the brain, causing the head to be enlarged. Without treatment, mental retardation and other neurologic damage may result.

If the child develops hydrocephalus, fluid can be drained from the brain through surgical placement of a special tube called a shunt. The shunt runs under the skin into the chest or abdomen, and the fluid passes harmlessly into the child's body.

Most children with severe spina bifida have a tethered spinal cord, meaning that the spinal cord does not slide up and down with movement as it should, because it is held in place by surrounding tissue. While most children have no symptoms from this, some suffer progressive loss of function in their legs, and a few develop scoliosis (curvature of the spine). If the spinal cord is surgically untethered soon after these symptoms begin, a child should return to his or her usual level of functioning.

Other chronic complications associated with severe spina bifida include obesity, gut and urinary tract disorders, psychological and sexual issues, and learning disabilities.

According to the Spina Bifida Association of America (SBAA), between 18 and 73 percent of children with spina bifida are allergic to latex (natural rubber), possibly due to intense exposure during surgeries and medical procedures. Symptoms may include watery eyes, wheezing, hives, rash, and even life-threatening anaphylactic reactions. Doctors should use only nonlatex gloves and equipment during any procedures on individuals with spina bifida. Affected individuals and their families should avoid latex items often found in the home and community, such as baby bottle nipples, pacifiers and balloons (a list is available from the SBAA).

With treatment, children with spina bifida usually can become active individuals. At least 70 percent of children with spina bifida have normal intelligence, although some children do have learning problems. Most affected women can have children, but such pregnancies are considered "high risk," as the risk of having a baby with spina bifida is about 1 in 100.

Can spina bifida be prevented?
Studies show that, if all women in the United States took enough of the B vitamin folic acid every day before and during early pregnancy, up to 70 percent of neural tube defects (including spina bifida) could be prevented. The key is having enough folic acid in the system before pregnancy and during the earliest weeks of pregnancy, before the neural structures close. The March of Dimes recommends that women take a multivitamin containing 400 micrograms of folic acid daily, and eat a healthy diet including foods rich in folic acid. This is the most reliable way of ensuring that a woman gets all the folic acid she needs.

Foods that contain folate (natural folic acid) include: orange juice, other citrus fruits and juices, leafy green vegetables, beans and whole-grain products. Multivitamins, fortified breakfast cereals, and enriched grain products contain a synthetic form of folic acid that is more easily absorbed by the body than the natural form. It is estimated that about half of food folate is absorbed by the body, while most (about 85 percent) of folic acid in fortified foods and virtually all (100 percent) of the folic acid in a vitamin supplement are absorbed. This is why the March of Dimes, Centers for Disease Control and Prevention (CDC), and Institute of Medicine recommend that women who could become pregnant consume 400 micrograms a day of the synthetic form. However, women should not take more than 1,000 micrograms (or 1 milligram) without their doctor's advice.

Women who already have had a baby with spina bifida or another NTD, as well as women who have spina bifida, diabetes or seizure disorders, should consult their health care providers before another pregnancy about the amount of folic acid to take. Studies have shown that taking a larger dose of folic acid daily (4 milligrams), beginning at least one month before pregnancy and in the first trimester of pregnancy, reduces the risk of having another affected pregnancy by about 70 percent.

Can spina bifida be detected prenatally?
Spina bifida often can be detected before birth using two or more tests. Most health care providers now routinely offer pregnant women a blood test called the maternal serum alpha-fetoprotein (MSAFP) screening test (part of the "triple screen"). This test detects pregnancies at higher-than-average risk of spina bifida and other NTDs, as well as certain other birth defects, including Down syndrome.

If a woman has a high MSAFP test result not caused by factors such as a miscalculation of fetal age, her health care provider probably will recommend two additional tests that are accurate in detecting severe spina bifida. These are a detailed ultrasound examination of the fetal spine, and amniocentesis to measure levels of alpha fetoprotein (AFP) in amniotic fluid.

What are the benefits of detecting spina bifida before birth?
When spina bifida is diagnosed before birth, health care professionals can provide parents with information and support. They can plan for delivery in a specially equipped medical center so that the baby can have any necessary surgery or treatment soon after birth.

In 1991, March of Dimes grantee David B. Shurtleff, MD, and others at the University of Washington in Seattle, found that cesarean delivery prior to the onset of labor can reduce the severity of paralysis in babies with spina bifida. If a baby is prenatally diagnosed with spina bifida, parents can discuss the possibility of a planned cesarean delivery with their doctors.

More than 100 babies have undergone experimental prenatal surgery to repair severe spina bifida before birth. A 1995 study by March of Dimes grantee N. Scott Adzick, MD, Children's Hospital of Philadelphia, suggests that chemical and physical trauma to the exposed spinal nerve tissue in the womb contributes to paralysis after birth, and that early repair may help prevent paralysis and other complications. To date, this procedure is being performed at only two medical centers, Children's Hospital of Philadelphia and Vanderbilt University Medical Center in Nashville, Tenn. Preliminary results appear promising: fewer babies who have had surgery require shunts to drain fluid from their brains. However, it is too soon to know how well most of these babies will walk, and there is a high risk that the procedure can induce preterm labor.

Is the March of Dimes conducting research on spina bifida?
March of Dimes grantees are searching for genes that may cause a predisposition to spina bifida and other NTDs and for improved treatments. One of our grantees has recently identified a mutation in a gene that helps to regulate how the body processes folic acid. Since maternal folic acid deficiency can lead to a baby with spina bifida, this mutant gene may increase the risk of having a child with spina bifida.

March of Dimes grantees also are seeking to improve the treatment of children with spina bifida through improved surgical approaches. One grantee is developing programs to prevent behavior and adjustment problems during adolescence in children with spina bifida. Another is studying how specific memory deficits may play a role in academic difficulties in some children with spina bifida. The March of Dimes also heads the National Council on Folic Acid, an alliance of organizations working to prevent NTDs.

What resources are available for families affected by spina bifida?
Genetic counselors can help families with questions about the chances of having affected children, and can explain prenatal testing for NTDs like spina bifida. The SBAA, with chapters nationwide, also can provide information and a newsletter.

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Addendum: New Fetal Surgery Trial
The most severe form of spina bifida (open spine) is myelomeningocele (see above). It occurs in one out of every 1,000 pregnancies, and nearly always requires surgery as soon as possible after birth, usually within the first day or two. Based on the hope that

the earlier the surgical repair could be done, the better would be the chance of an improved outcome for the baby, pioneer surgeons in recent years developed an experimental technique for performing the surgery prenatally, somewhere between the 19th and 25th weeks of pregnancy. These efforts at fetal surgery sometimes seem to result in a beneficial outcome for some of the serious complications of myelomeningocele. However, as yet, there are insufficient data to know if the results of fetal surgery were worth the added risk. Potential complications include greatly increased chances of premature delivery for the baby, with its own risks to the child. There are also risks for the mother, who has to undergo otherwise unnecessary surgery, with the risk of suffering long-term adverse effects, in order to allow access to the fetus.