Introduction to Spina Bifida

Written and Reviewed by: My Child Without Limits Advisory Committee

Spina bifida, which literally means "cleft spine," is a term used to describe the incomplete development of the brain, spinal cord, and/or meninges (the protective covering around the brain and spinal cord).

Who Does Spina Bifida Affect?

In the United States, spina bifida is the most common defect of the neural tube (a structure in an embryo that eventually becomes the baby's spinal cord and brain). It affects 1,500 to 2,000 of the more than 4 million babies born in the country each year.

What Causes Spina Bifida?

The exact cause of spina bifida is not known. Scientists think there are genetic, nutritional, and environmental reasons. However, research studies have shown that not enough folic acid -- a common B vitamin -- in the mother's diet is a key cause of spina bifida and other neural tube defects. This is why vitamins that are given to pregnant mothers typically contain folic acid as well as other vitamins. (See "Can spina bifida be prevented?" for more information on folic acid.)

How is Spina Bifida Diagnosed?

In most cases, spina bifida is diagnosed before birth. However, some mild cases may go unnoticed until after birth. Very mild cases, in which there are no symptoms, may never be detected.

Prenatal (Before Birth) Diagnosis
Certain screening tests, including blood tests, ultrasound, and amniocentesis, can help doctors to tell if the fetus has a neural tube defect.

Postnatal (After Birth) Diagnosis
Mild cases of spina bifida that are not diagnosed during prenatal testing may be detected after the child is born. This usually happens when an X-ray is taken during a routine examination. If spina bifida is suspected, doctors may use magnetic resonance imaging (MRI) or a computed tomography (CT) scan to get a clearer view of the spine and vertebrae. If hydrocephalus (excess fluid in the brain) is suspected, the doctor may request a CT scan and/or X-ray of the skull.

What Are the Different Types of Spina Bifida?

There are four types of spina bifida: occulta, closed neural tube defects, meningocele, and myelomeningocele.

Occulta is the mildest and most common form. With occulta one or more vertebrae (bones that make up the spine) are not properly formed. The name "occulta," meaning "hidden," means that the malformation, or opening in the spine, is covered by a layer of skin. This form of spina bifida does not usually causes disability or symptoms.

Closed neural tube defects are the second type of spina bifida. This form consists of a group of spinal defects in which the spinal cord is marked by a malformation of fat, bone, or membranes. In some patients there are few or no symptoms; in others the malformation causes partial paralysis with urinary and bowel problems.

In the third type, **meningocele**, the meninges (protective coverings around the spine) push out from the spinal opening, and may or may not be covered by a layer of skin. Some patients with meningocele may have few or no symptoms. Others may experience symptoms that are similar to closed neural tube defects.

Myelomeningocele, the fourth form, is the most severe. This occurs when the spinal cord shows through the opening in the spine. resulting in partial or complete paralysis of the parts of the body below the spinal opening. The paralysis may be so severe that the patient with this type of spina bifida is unable to walk and may have urinary and bowel dysfunction.

What Are the Signs and Symptoms of Spina Bifida?

There are different types of spina bifida. Therefore the symptoms vary from person to person, depending on which type they have.

Often, individuals with the occulta and closed neural tube defects types of spina bifida have no outward signs of the disorder. This is because the defect is hidden under the skin. These types of defects, may be noticed early in life if there is an abnormal tuft or clump of hair, or a small dimple or birthmark on the skin at the site where the spine is not properly formed.

With other types of spina bifida, meningocele and myelomeningocele, there will usually be a fluid-filled sac -- visible on the back -- sticking out from the spinal cord. In meningocele, a thin layer of skin may cover the sac. In most cases of myelomeningocele, there is no layer of skin covering the sac and a portion of spinal cord tissue is usually exposed.

Can Spina Bifida Be Prevented?

Folic acid, also called folate, is an important vitamin in the development of a healthy fetus. Although taking this vitamin cannot guarantee that a mother will have a healthy baby, it is believed that it can help. Recent studies have shown that by adding folic acid to their diets, women of childbearing age greatly reduce the risk of having a child with a neural tube defect such as spina bifida. Therefore, it is recommended that all women of childbearing age consume 400 micrograms of folic acid daily. Foods that are high in folic acid include dark green vegetables, egg yolks, and some fruits. Many foods -- including some breakfast cereals,

enriched breads, flours, pastas, rice, and other grain products -- now have added folic acid. A lot of multivitamins contain the recommended dosage of folic acid as well.

Women who already have a child with spina bifida, have spina bifida themselves, or have already had a pregnancy affected by any neural tube defect have a greater chance than other women of having a child with spina bifida or another neural tube defect. These women may need more folic acid before they become pregnant.

What Kind of Complications Can Spina Bifida Cause?

Complications caused by spina bifida can range from minor physical problems to severe physical and mental disabilities. However, most people with spina bifida have normal intelligence.

How severe the complications are depend on the size and location of the malformation, whether or not skin covers it, whether or not spinal nerves are exposed, and which spinal nerves are involved. Most of the time, all the nerves that are located below the malformation are affected. This means that the higher the malformation is on the child's back, the greater the amount of nerve damage and loss of muscle function and sensation.

In addition to loss of feeling and paralysis, another complication associated with spina bifida is Chiari II malformation -- a rare condition (but common in children with myelomeningocele) in which the brainstem and the cerebellum, or rear portion of the brain, push downward into the spinal canal or neck area. This condition can lead to squeezing of the spinal cord and cause a variety of symptoms including difficulties with eating, swallowing, and breathing; choking; and arm stiffness.

Chiari II malformation may also cause a condition called Hydrocephalus. This means there is an abnormal buildup of cerebrospinal fluid in the brain. (Cerebrospinal fluid is a clear liquid that surrounds the brain and spinal cord). The buildup of fluid puts damaging pressure on the brain. Hydrocephalus is commonly treated by surgically implanting a shunt -- a hollow tube -- in the brain to drain the excess fluid into the abdomen. Some newborns with myelomeningocele may develop meningitis, an infection in the meninges. Meningitis may cause brain injury and can be life-threatening.

Children with both myelomeningocele and hydrocephalus may have learning disabilities, such as difficulty paying attention, problems with language and reading, and trouble learning math.
Additional problems such as latex allergies, skin problems, digestive conditions, and depression may occur as children with spina bifida get older.

How Is Spina Bifida Treated?

There is no cure for spina bifida. The nerve tissue that is damaged or lost cannot be repaired or replaced. However, there are certain treatments. Treatment depends on the type and severity of the disorder. Generally, children born with the mild form of spina bifida need no immediate treatment, although some may require surgery as they grow.

The most important goals in treating myelomeningocele are to prevent infection from developing through the exposed nerves and tissue of the defect on the spine, and to protect the exposed nerves and structures from additional damage. Typically, a child born with spina bifida will have surgery to close the defect and prevent infection or further damage within the first few days of life.

Doctors have recently begun performing fetal surgery to treat myelomeningocele. Fetal surgery, which takes place before birth, is performed in utero (within the uterus). This kind of surgery involves opening the mother's abdomen and uterus and sewing shut the opening over the developing baby's spinal cord.
Some doctors believe the earlier the defect is corrected, the better it will be for the baby. Although the procedure cannot bring back lost nerve function, it may prevent additional loss from happening. However, the surgery is considered experimental and there are risks for the unborn child as well as for the mother.
If the surgery causes the baby to be born too early, there can be complications like organs that aren't mature, brain bleeding, and death. Risks for the mother include infection, blood loss, gestational diabetes, and weight gain due to bed rest.

Still, the benefits of fetal surgery are promising. Surgery may help reduce exposure of the delicate spinal nerve tissue and bones to the environment inside the uterus - including the amniotic fluid, which is considered toxic to the exposed areas.

Another benefit that doctors have discovered is that the procedure positively affects the way the brain develops in the uterus. Certain complications -- such as Chiari II with associated hydrocephalus -- are able to correct themselves when surgery is performed. This can reduce, and sometimes, eliminate the need for surgery after birth to implant a shunt to drain excess brain fluid.

Many children with myelomeningocele develop a condition called progressive tethering, or tethered cord syndrome. With this condition the spinal cord becomes attached to an immovable structure -- such as overlying membranes and vertebrae -- causing the spinal cord to become abnormally stretched and the vertebrae to lengthen with growth and movement. This condition can cause a loss of muscle function in the legs, bowel, and bladder. Early surgery on the spinal cord may allow the child to regain a normal level of functioning and prevent further nerve deterioration.

Some children will need additional surgeries to manage problems with the feet, hips, or spine. Individuals with hydrocephalus (excess brain fluid) will usually need additional surgeries over time to replace the shunt (drain), which can be outgrown or become clogged.

Some individuals with spina bifida need medical devices such as braces, crutches, or wheelchairs. The location of the malformation on the spine often indicates the type of devices needed. Children with a defect high up on the spine and more paralysis will often require a wheelchair, while those with a defect lower on the spine may be able to use crutches, bladder tubes, leg braces, or walkers.

Treatment for paralysis and bladder and bowel problems usually begins soon after birth, and may include special exercises for the legs and feet to help prepare the child for walking with braces or crutches when he or she is older.

What Does the Future Hold for Children with Spina Bifida?

Children with spina bifida can lead relatively active lives. How the child will progress in life depends on the number and severity of abnormalities and associated complications. Most children with the disorder have normal intelligence and can walk, usually with assistive devices. If learning problems develop, early educational intervention is helpful.

What Research Is Being Done to Find a Cure or Additional Treatments for Spina Bifida?

Within the Federal Government, the National Institute of Neurological Disorders and Stroke (NINDS), a component of the National Institutes of Health (NIH), supports and conducts research on brain and nervous system disorders, including spina bifida.

In one study supported by NINDS, scientists are looking at the hereditary basis of neural tube defects. The goal of this research is to find the genetic factors that make some children more susceptible to neural tube defects than others. Lessons learned from this research will fill in gaps of knowledge about the causes of neural tube defects and may lead to ways to prevent these disorders. These researchers are also studying the role genes play in the process of neural tube closure, which will provide information on the human nervous system during development, which may result in improved clinical care, treatment, and genetic counseling.

Other scientists are studying genetic risk factors for spina bifida, especially those that diminish or lessen the function of folic acid in the mother during pregnancy, possibly leading to spina bifida in the fetus. This study will shed light on how folic acid prevents spina bifida and may lead to improved forms of folate supplements.

NINDS also supports and conducts a wide range of basic research studies to understand how the brain and nervous system develop. These studies help scientists to better understand neural tube defects, such as spina bifida, and offer hope for new avenues of treatment for and prevention of these disorders as well as other birth defects.

Another component of the NIH, the National Institute of Child Health and Human Development (NICHD), is conducting a large five-year study to determine if fetal surgery to correct spina bifida in the womb is safer

and more effective than the traditional surgery -- which takes place a few days after birth. Researchers hope this study, called the Management of Myelomeningocele Study, or MOMS, will help them to know which procedure, pre-birth or post-birth, is best for the baby.

Spina Bifida Resources

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