Spina Bifida

U.S. DEPARTMENT OF HEALTH
AND HUMAN SERVICES
National Institutes of Health

Spina Bifida

What is spina bifida?

Spina bifida is a birth defect that mainly affects the spine. Normally in the first month of pregnancy, a special set of cells forms the "neural tube." The top of the tube becomes the brain and the remainder becomes the spinal cord and structures around it. In spina bifida, the neural tube doesn't close all the way and some of the bones of the spine don't close in the back.

Often, abnormalities of the brain (such as hydrocephalus, described below) accompany abnormalities of the spine because the neural tube closes first in the middle and then closure proceeds both upward and downward—meaning that if something happens that prevents normal formation of the spine, it may also prevent normal formation of the part of the brain that is forming (closing) at the same time.

The term neural tube defect describes a group of conditions, including spina bifida, that occur when the neural tube does not close all the way.

Each year approximately 1,400 babies born in the United States have spina bifida, according to the U.S. Centers for Disease Control and Prevention. The exact cause of spina bifida is unknown. There is no cure but most people with spina bifida lead long and productive lives.

Scientists suspect genetic, nutritional, and environmental factors all play a role in spina bifida. People with spina bifida have different abilities and medical issues.

What are the types of spina bifida?

There are four types of spina bifida: occulta, closed neural tube defects, meningocele, and myelomeningocele. The symptoms of spina bifida vary from person to person, depending on the type and level of involvement.

- **Occulta** is the mildest and most common form in which one or more bones of the spinal column (called vertebrae) are malformed. The name "occulta," which means "hidden," indicates that a layer of skin covers the opening in the bones of the spine. It is often found by accident on an x-ray or similar test. This form of spina bifida very rarely causes disabilities or symptoms.
- **Closed neural tube defects** are a diverse group of defects in which the spine may have malformations of fat, bone, or the membranes (called the meninges) that cover the spinal cord. Many of these neural tube defects require surgery in childhood. People with this type of spina bifida may have weakness of the legs and trouble with bowel and bladder control. These issues may change or progress as children grow. It is important to have close follow-up with doctors to minimize these changes as much as possible.
- **Meningocele** occurs when a sac of spinal fluid pokes through the spine. This fluid is normally only around the brain and spine, but a problem with the bony covering over

the spine allows it to poke out in this case. The malformation contains no nerves and may or may not be covered by a layer of skin. Individuals with meningocele may have minor symptoms.

- **Myelomeningocele** is the most severe form of spina bifida. A portion of the spinal cord or nerves is exposed in a sac through an opening in the spine that may or may not be covered by the meninges. The opening can be closed by surgeons while the baby is in utero or shortly after the baby is born. Most people with myelomeningocele have changes in their brain structure, leg weakness, and bladder and bowel dysfunction. Myelomeningocele is often called a snowflake condition because no two people with the condition are the same. Typically, the lower in the spine the opening occurs relates to less symptoms in the person. People with myelomeningocele require close follow-up with physicians throughout their childhood and lifespan to maximize their function and prevent complications such as kidney failure.

How does folic acid help?

Folic acid, also called folate, is an important vitamin for the development of a healthy fetus. Although taking this vitamin cannot guarantee having a healthy baby, it can help. Studies show that women of childbearing age who add folic acid to their diets can significantly reduce the risk of having a child with a neural tube defect. Therefore, it is recommended that all women of childbearing age take a daily vitamin supplement with 400 micrograms of folic acid before and during early pregnancy.

Foods high in folic acid include dark green vegetables, egg yolks, and some fruits. Many foods—such as some breakfast cereals, enriched breads, flours, pastas, rice, and other grain products—are now fortified with folic acid. Many multivitamins contain the recommended dosage of folic acid as well.

Women who already have a child with spina bifida, who have spina bifida themselves, or who have already had a pregnancy affected by any neural tube defect are at greater risk of having another child with a neural tube defect. These women should take a higher prescription dose of folic acid before and early in pregnancy.

Spina bifida complications

Spina bifida's impact is determined by the type of defect and in the case of myelomeningocele and closed neural tube defects the size and location of the malformation.

Spina bifida complications may include:

- **Abnormal sensation or paralysis**, which mostly occurs with closed neural tube defects and myelomeningocele. People with these conditions typically have some degree of leg and core muscle weakness and loss of feeling in the groin and feet or legs. The sensation can be more significant on one side of the body. Typically, the lower in the spine where the condition occurs results in less weakness and loss of feeling. The strength and feeling do not improve with age due to nerve damage. People with these types of spina bifida may lose strength and sensation as they grow and mobility can become more

difficult with age. People with these conditions may walk independently or use some combination of leg braces, walkers, crutches, or wheelchairs. As they age, they may require more of these supports.

- **Chiari II malformation**, in which parts of the brain called the brain stem and the cerebellum (hindbrain) protrude downward into the spinal canal or neck area. It is almost always seen on advanced imagining of the brain in people with myelomeningocele, but it rarely causes symptoms. When it does, this condition can press on the spinal cord and cause a variety of symptoms including difficulty breathing, swallowing, and arm weakness. Surgery is sometimes required to reduce pressure in this area.
- **Blockage of cerebrospinal fluid, causing a condition called hydrocephalus**. Hydrocephalus is the abnormal buildup of the fluid that surrounds the brain. Most people with myelomeningocele have this condition, which is not seen in the other types of spina bifida. This buildup can put damaging pressure on the brain. Hydrocephalus is commonly treated by surgically implanting a shunt—a hollow tube—in the brain which allows drainage of the excess fluid into the abdomen where it is absorbed by the body. The tube is tunneled under the skin and not very noticeable to others. Another treatment option is an endoscopic third ventriculostomy or ETV, a procedure that creates a new path for the fluid to flow.

- **Meningitis**, an infection in the meninges covering the brain. It can sometimes be associated with shunts. Meningitis may cause brain injury and can be life-threatening.
- **Tethered cord syndrome** can occur with all forms of spina bifida, although it is very rare in individuals with spina bifida occulta. Usually the spinal cord and nerves float freely. A tethered cord means that there is some type of tissue attached to and pulling the cord down. This can cause damage to the nerves and decrease feeling and strength, as well as problems with bowel and bladder control. It is surgically treated if a person has symptoms.
- **Bowel and bladder incontinence** affect most individuals with myelomeningocele and closed neural tube defects. The nerves at the very bottom of the spine control bowel and bladder function and don't usually work properly in people with these types of spina bifida. Most people with myelomeningocele and some types of closed neural tube defects need a regimen or other assistance to drain their bladders periodically or to have regularly scheduled bowel movements.
- **Learning disabilities**, including difficulty paying attention, understanding concepts, impaired motor skills, impaired memory, and difficulty with organization and problem solving are commonly seen in children with myelomeningocele. People with strength lower down in their legs tend to have less difficulty than those

with more leg weakness. Evaluation for an individualized education plan is recommended for all children with myelomeningocele.

- **Other complications** such as skin ulcers, low bone mineral density, impaired male fertility, obesity, and kidney failure can be seen in people with myelomeningocele and neural tube defects as they age. Additionally, people with myelomeningocele are at risk for precocious puberty (when changes to that of an adult occur too soon), sleep apnea, and depression.

How is spina bifida diagnosed?

In most cases, spina bifida is diagnosed before birth (prenatal). However, some mild cases may go unnoticed until after birth (postnatal). Very mild forms of spinal bifida are found when doing tests for other conditions or may never be detected.

Prenatal Diagnosis

The most common screening methods used to look for spina bifida during pregnancy are maternal serum alpha fetoprotein (MSAFP) screening and fetal ultrasound. A doctor can also perform an amniocentesis test.

- **Maternal serum alpha fetoprotein (MSAFP) screen**. At 16 to 18 weeks of pregnancy, a sample of the mother's blood is taken to measure the level of a protein called *alpha-fetoprotein* (AFP), which is made naturally by the fetus and placenta. During pregnancy, a small amount of AFP normally crosses the placenta and enters

the mother's bloodstream. Abnormally high levels of AFP may indicate that the fetus has spina bifida or other neural tube defect. This test is not specific for spina bifida and cannot definitively determine that there is a problem with the fetus. This means that a high AFP level alone is not enough to be sure the fetus has a neural tube defect. If a high level of AFP is detected, the doctor may request additional testing, such as an ultrasound or amniocentesis.

- **Ultrasound**. A fetal ultrasound uses high-frequency sound waves to create a picture of the developing baby inside the womb. It is highly accurate in diagnosing some birth defects during pregnancy, including spina bifida. Fetal ultrasound can be performed during the first trimester (usually between 11-14 weeks) and the second trimester (usually at 18-22 weeks), and diagnosis is more accurate during the second trimester.
- **Amniocentesis**. In this test, a doctor removes a sample of the amniotic fluid that surrounds the fetus and tests it for protein levels that may indicate a neural tube defect and genetic disorders.

Postnatal Diagnosis

Closed neural tube defects are often recognized at birth due to an abnormal fatty mass, tuft or clump of hair, or a small dimple or birthmark on the skin at the site of the spinal malformation. Spina bifida occulta is usually found when x-rays are done for another reason.

In rare cases, myelomeningocele and meningocele are not diagnosed during routine prenatal tests. The baby will be diagnosed when they are born with a bubble on their back. Babies with myelomeningocele and closed neural tube defects may have muscle weakness in their feet, hips, and legs that result in joint deformities first noticed at birth. Mild cases of spina bifida (occulta, closed neural tube defects) not diagnosed during prenatal testing may be detected postnatally using ultrasound or X-ray imaging to look at the spine.

Doctors may use magnetic resonance imaging (MRI) or a computed tomography (CT) scan to get a clearer view of the spinal cord and vertebrae. To evaluate for hydrocephalus, the doctor will request a head ultrasound, CT or MRI to look for extra cerebrospinal fluid inside the brain.

How is spina bifida treated?

Treatment depends on the type of spina bifida a person has. Myelomeningocele and meningocele require a surgery to close the bubble shortly after birth to prevent infection such as meningitis. Most people with myelomeningocele have hydrocephalus and most of them will need a shunt placed as an infant. Children with a closed neural tube defect may need surgery to prevent further complications such as weakness and bowel and bladder function. Generally, people with spina bifida occulta will not need any treatment.

Prenatal Surgery

Prenatal (before birth) surgery involves opening the mother's abdomen and uterus (or womb) and sewing shut the abnormal opening over the developing baby's spinal cord. This is thought to protect the baby's spinal cord from ongoing damage in the uterus. The Management of Myelomeningocele Study (MOMS) showed that prenatal surgery to close the defect in the spinal cord improved outcomes compared to children who had postnatal surgery for spina bifida. Data from the 2012 study showed that prenatal surgery reduced the need to drain fluid from the brain, improved mobility, and increased the chances that a child will be able to walk independently early on. This study was funded by the National Institutes of Health's *Eunice Kennedy Shriver* National Institute of Child Health and Human Development.

The procedure does not restore lost neurological function that happened before the surgery, but may prevent additional damage from occurring during the rest of the pregnancy. Although prenatal surgery poses some risk to the fetus as well as to the mother, the benefits are promising and are still being studied.

Postnatal Surgery

In treating myelomeningocele and meningocele, the key priorities are to prevent infection from developing in the exposed nerves and spinal cord through the spinal defect, and to protect the exposed nerves and spinal cord from additional trauma. Therefore, a child born with these types of spina bifida who has not undergone prenatal surgery will have surgery to close the defect and

minimize the risk of infection or further trauma within the first few days of life.

Treatments for Complications

Some children with myelomeningocele and closed neural tube defects will need surgery to improve the alignment of their feet, legs, or spine. Children with myelomeningocele usually have hydrocephalus and may require surgery to help drain fluid in the brain, such as the placement of a shunt or ETV. Multiple surgeries may be required to replace the shunt, which may become clogged, infected, or disconnected.

Some individuals with myelomeningocele or closed neural tube defects require assistive devices for mobility such as braces, walkers, crutches, or wheelchairs. The location of the defect on the spine often determines the type of assistive devices needed. Children with a defect high on the spine will have little movement of the legs and will use a wheelchair for mobility. Children with a defect lower on the spine usually have more strength in the legs. They may be able to walk independently, or they may use crutches, leg braces, walkers, and wheelchairs depending on the activity. Children with myelomeningocele usually have some degree of delayed mobility, so they are referred to physical therapists early on to maximize their strength and function.

Treatment for bladder and bowel dysfunction typically begins soon after birth. Children with myelomeningocele and some closed neural tube defects have damage to the lowest spinal nerves which control typical bowel

and bladder function. Some children may be able to urinate typically, but most will need to drain their bladders with a catheter or thin tube 4-6 times a day to remain dry in between and to prevent kidney damage. Kidneys are monitored closely so that medications or surgeries can be performed to prevent renal failure. To prevent bowel accidents many people with myelomeningocele and closed neural type defects will use rectal medications or large volume enemas to have planned bowel movements. Close follow-up with a spina bifida specialty clinic is recommended to develop a safe bowel and bladder program.

Treatment for progressive tethering of the spinal cord (called tethered cord syndrome) can be treated with surgery to help prevent further neurological deterioration.

What research is being done?

The National Institute of Neurological Disorders and Stroke (NINDS), one of the National Institutes of Health (NIH), is the primary federal supporter of research on brain and nervous system disorders. NINDS conducts research in its laboratories at the NIH in Bethesda, Maryland, and supports research through grants to major medical institutions across the country. In addition to NINDS, other NIH Institutes support research on spina bifida and neural tube defects.

Genetic studies. In one study supported by NINDS, scientists are looking at the hereditary basis of neural tube defects and hope to find the genetic factors that make some children more likely to have a neural tube defect.

These researchers are also studying gene expression during the process of normal neural tube closure, which will provide information on the human nervous system during development. Findings may lead to ways to prevent these disorders.

In addition, NINDS-supported scientists are working to identify, characterize, and evaluate genes involved in neural tube defects. The goal is to understand the genetics of neural tube closure and to develop information that will lead to improved clinical care, treatment, and genetic counseling.

Other scientists are studying genetic risk factors for spina bifida, especially those that reduce the effectiveness of folic acid in preventing spina bifida. This study will shed light on how folic acid prevents spina bifida and may lead to improved forms of folate supplements.

Developmental studies. NINDS supports and conducts a wide range of basic research studies to understand how the brain and nervous system develop. These studies contribute to a greater understanding of neural tube defects such as spina bifida and offer hope for new ways to treat and potentially prevent these disorders as well as other birth defects.

Surgery. Results of the Management of Myelomeningocele Study (MOMS) (see **Prenatal Surgery**) showed significant benefit to the developing baby. Because fetal surgery has shown promise, NINDS-funded researchers are also developing new methods, such as stem cell patches and tissue engineering, to add to the prenatal repair of spinal defects.

Other NIH research efforts. More information about research on spina bifida supported by NINDS and other NIH Institutes and Centers can be found using NIH RePORTER (www.projectreporter.nih.gov), a searchable database of current and past research projects supported by NIH and other federal agencies. RePORTER also includes links to publications and resources from these projects. Enter "spina bifida" to start your search.

How can I help with research?

Consider joining a clinical study. Both healthy individuals and those with a disease or condition can participate in medical research studies (sometimes called clinical trials or protocols) to help researchers better understand a disease and perhaps develop new treatments. For information about clinical studies on disorders including spina bifida and how to participate in one, please contact the NIH's Patient Recruitment and Public Liaison office at 800-411-1222 or visit the Clinicaltrials.gov website at https://www.clinicaltrials.gov.

Where can I get more information?

For more information on neurological disorders or research programs funded by the National Institute of Neurological Disorders and Stroke, contact the Institute's Brain Resources and Information Network (BRAIN) at:

BRAIN
P.O. Box 5801
Bethesda, MD 20824
800-352-9424
http://www.ninds.nih.gov

Information also is available from the following organizations:

Eunice Kennedy Shriver **National Institute of Child Health and Human Development**
Information Resource Center
P.O. Box 3006
Rockville, MD 20847
800-370-2943 or 888-320-6942 (TTY)
http://www.nichd.nih.gov

Spina Bifida Association
1600 Wilson Boulevard, Suite 800
800-621-3141
https://www.spinabifidaassociation.org

March of Dimes
1550 Crystal Drive, Suite 1300
Arlington, VA 22202
888-663-4637
https://www.marchofdimes.org

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Prepared by:
Office of Neuroscience Communications and Engagement
National Institute of Neurological Disorders and Stroke
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NIH Publication No. 21-NS-309 January 2021