

About Spina Bifida

Spina Bifida

Spina bifida is a complicated birth defect which has occurred in human beings for thousands of years. In the United States, spina bifida is the second most common birth defect and affects about one out of every one thousand pregnancies. In some babies, for reasons that appear to be a combination of environmental and genetic factors, normal development is interrupted anywhere from the brain to the end of the spinal cord and the birth defect called spina bifida results (Figure 1). This problem occurs very early in pregnancy, probably in the third or fourth week following fertilization. It is possible that only the bones of the spinal column will be incompletely developed and that the nerves beneath will be normal. This condition, called spina bifida occulta, does not cause neurologic problems such as paralysis or weakness and is not medically significant. When the spinal cord itself is not fully formed however, the nerves do not develop as they should and the baby will have myelomeningocele, the most severe form of spina bifida (Figure 2a),(Figure 2b). It is this type of spina bifida that is discussed on this website.

Causes of Spina Bifida

At this time the cause or causes of spina bifida are not well understood. In a small number of women it appears to be caused by certain medications, most often antiseizure drugs. In some cases it has occurred in the family before, but this is relatively uncommon. In the majority of cases the cause of the spina bifida is never determined. However, it is now known that taking folic acid, a B vitamin, before a woman becomes pregnant will reduce the chances that the baby will have spina bifida and related conditions of the brain and spinal cord.

Prenatal Detection

There are several tests available to pregnant women which can be used to detect spina bifida before the baby is born. The most commonly used test is the maternal serum alpha fetoprotein (MSAFP) blood test which is done on the mother's blood around the 16th week of pregnancy. This test checks the level of protein (alpha fetoprotein) that leaks out of the developing brain and spinal cord into the fluid surrounding the baby. This protein is normally found in amniotic fluid, but the levels found are higher if the baby has spina bifida. This test is not specific for spina bifida, however. An error in the dates of the pregnancy, multiple babies, as well as other birth defects can all cause an abnormal reading. If the alpha fetoprotein level is abnormal, additional testing is recommended.

Main Effects on the Body

- Effects on the Spinal Cord

Spina bifida can adversely affect many body systems including the nervous system, the bones

and muscles as well as the kidneys and bladder. The point along the spinal cord where the undeveloped area occurs, is called the "level" of the spina bifida (Figure 3). The higher up the spinal column the "level" occurs, the greater the effect on normal nerve function. Some individuals with low levels of spina bifida can walk with little or no assistance where those with higher levels will require braces and in cases of very high levels, wheelchairs, to get around.

- Effects on the Brain

Most individuals with spina bifida will have differences in the development of the brain itself. In those with spina bifida, the brain is usually positioned further down into the upper spinal column than it should be. This change in position is part of a condition called the Chiari II malformation. The brain tissue displaced into the upper spinal canal blocks the normal flow of cerebrospinal fluid, also known as CSF. This leads to a build-up of fluid within the cavities or "ventricles" of the brain. This condition is called hydrocephalus (Figure 4). In 80 percent to 90 percent of individuals with spina bifida, the hydrocephalus can only be treated by the insertion of a drainage tube called a "shunt" (Figure 5). There are no medications that can treat hydrocephalus effectively. The shunt is placed under the skin from the head to the abdominal cavity where the excess CSF is absorbed back into the body. Shunts are not a perfect solution to the management of hydrocephalus as they can break, become clogged or infected and sometimes need to be replaced when the child grows. When a shunt fails it usually needs to be replaced. In most cases shunts are required for life.

- Effects on the Bowel and Bladder

Bowel and bladder function are controlled by nerves which come from the lowest levels of the spinal column. Therefore, almost all individuals with spina bifida will have trouble controlling their bowel and bladder function (Figure 6). Close attention to emptying the bladder, promptly treating urinary tract infections and maintaining kidney function is essential to preserving the health of individuals with spina bifida.

- Effects on the Muscles and Bones

The effect of spina bifida on the muscles and bones is complex and varies significantly depending on the level of the defect in the spinal cord. Some degree of paralysis occurs with most cases of spina bifida (Table 1). Higher levels cause more severe loss of muscle function. Because the muscles of the body support the bones and keep them balanced, loss of muscle function results in a variety of problems including dislocated joints, misshapen bones, dislocated hips and curvature of the spine. Problems caused by loss of muscle function are managed with a combination of bracing, physical therapy and surgery.

- Other Medical Problems

Individuals with spina bifida are also at a higher risk for several medical conditions found in the general population. These problems include fractures, seizures, lazy eye, early puberty, and allergy to latex (natural rubber).

Important Non-Medical Issues Affecting Individuals with Spina Bifida

Spina bifida can affect educational, social, and psychological development.

Educational issues: The intelligence of individuals with spina bifida varies, just as it does with people in general. IQ scores of those with spina bifida cluster in the 70 - 90 range as opposed to

the 100 range as they do in those without this birth defect. Affected individuals often have poor short term memories and poor organizational skills. Some children with spina bifida do well in a regular classroom where others do best in a special educational setting.

Social development: In many cases, infants and children with spina bifida require early and frequent hospitalization. This can interrupt normal social development. The challenge is to balance medical needs with the need to let a child develop into a confident, self-sufficient and independent adult.

Psychological development: Children with special needs of any type often rebel against their disability when they realize it cannot be wished away. They may become depressed, defiant or withdrawn. Early attention to these issues through peer support groups and/or counseling is often critical to healthy psychological development.

Life Expectancy of Children Born with Spina Bifida

This is a difficult question to answer since the treatment for people with spina bifida has been improving steadily for the past several decades. In general, individuals who get early treatment and continue to have regular medical check-ups with specialists familiar with the medical problems of individuals with spina bifida do much better than those who do not. When there are no severe problems with hydrocephalus, unusual problems from the Chiari II malformation or significant kidney damage, the vast majority of people with spina bifida will live into adulthood and have normal or near normal life expectancies.

Treatments Available for Children and Adults with Spina Bifida

Because spina bifida affects so many body systems it is important that professionals from many areas be consulted to provide up-to-date, comprehensive medical, psychological and social evaluation, support and treatment. There are many spina bifida clinics throughout the country which bring the appropriate specialists together to provide the necessary care.

The search for more effective treatments for individuals with spina bifida is continuing. For the past several years several medical centers around the country have been operating on babies with spina bifida to close the open defect in the spine while they are still in their mothers' womb. Preliminary results are encouraging, but at this time it is not known if surgery before or after birth is best.