

Spina bifida

Patient Information Series – What you should know, what you should ask.

What is spina bifida?

Spina bifida is a defect of the spine exposing the contents of the spinal cord (the nerves going from the brain to various parts of the body). There are two kinds of spina bifida; an open defect is when there is no skin covering the hole in the spine; a closed defect is, instead, covered completely by the skin.

How does spina bifida happen?

Spina bifida is a failure of closure of the spine during the first month of pregnancy. In most cases of open spina bifida, there is a fluid-filled sac bulging out. It contains deformed nerves and the sac around them called meninges. That sac is called a myelomeningocele. Less frequently, the spine is open in the back and is not covered by meninges; this is called a myelocele.

The cause for the spina bifida is variable with many factors often contributing to the anomaly. A lack of folic acid increases the risk of spina bifida. It is important when you plan to have a baby to increase your intake of folic acid before conception.

A problem with the number of chromosomes (where our genetic make-up is stored) or changes within the chromosomes are more frequent in babies with spina bifida. Other genetic problems can be the reason for the spina bifida. Most often, no explanation is found on why this is happening.

Should I have more tests done?

The two main categories of anomalies associated with spina bifida are anomalies of the brain and foot deformities. In almost all cases of open spina bifida, a typical change in the back of the brain is found (called Arnold-Chiari malformation). Many babies will also have extra fluid inside their brain. The feet will often be in a clubbed position. Some babies will also have other anomalies in their body which can make things more challenging. Because of this, a specialized ultrasound is recommended to thoroughly assess the baby.

As some babies will have anomalies in their chromosomes, testing them with an amniocentesis is often offered. When an amniocentesis is done, a needle is inserted in your abdomen to collect some fluid from around the baby and test it.

In rare cases, the neural tube defect is part of a genetic disease, such as Meckel syndrome, Jarcho-Levin syndrome, and Currarino syndrome. These disease are more likely if there are other problems detected on the ultrasound. Many women will receive genetic counseling and decide with a specialized doctor if these possibilities should be tested by an amniocentesis.

What does it mean for my baby after it is born?

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The outcome is extremely variable, depending where and how much of the spine is affected and on whether or not other anomalies are seen. Although most babies survive, some will not. Those who survive may present problems such as paralysis of limbs, loss of bladder or bowel control, sexual dysfunction and learning disabilities. Severe excessive amount of fluid in the brain, clubfoot and curved spine are associated with more problems after birth.

After birth, your baby will need surgery to repair the defect and long term follow-up to help with the various issues associated with the changes in the spine and the nerves. Many will require multiple surgeries as they grow up.

Will it happen again?

The risk of having another child with spina bifida or similar types of problems is around 2-4%. A higher dose of folic acid vitamin before trying to become pregnant can decrease that risk. Rarely, the risk will be much higher because certain genetic causes were found. A doctor with expertise in genetics can help you assessing your specific risk.

What other questions should I ask?

- Is the spina bifida open or closed?
- Are other malformations present (such as Arnold-Chiari, clubfoot, scoliosis)?
- What genetic testing is available?
- How often will I have ultrasound examinations done?
- Is surgery during the pregnancy an option?
- Where is surgery after pregnancy available for the baby?
- Where should I deliver?
- Where will the baby receive the best care after delivery?
- Can I meet in advance the team of doctors that will be looking after my baby following delivery?

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