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 that occurs in about 1 in 1500 births, where the contents of the spinal cord (the nerves going from the brain to various parts of the body) are exposed. Spina bifida is a neural tube defect and occurs when the developing spine does not close completely, resulting in exposure of the spinal cord contents to the amniotic fluid. It is usually seen in the lower back and the sacral region but can also involve the upper back or the neck. 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 but the bony elements are underdeveloped or not present.

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 of the backbone. 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. Folic acid is a water-soluble vitamin which is commonly found in green vegetables. 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. Such babies usually have other abnormalities as well in other organ systems. Other genetic problems can be the reason for the spina bifida.

Intake of certain medicines such as Sodium Valproate and Carbamazepine during the early period of pregnancy increases the risk of spina bifida in the baby. These drugs are commonly used for treatment of epilepsy and at times certain mental health conditions like bipolar disorder.

Other risk factors include maternal diabetes and obesity.

Most often, no explanation is found for this condition. However, a detailed evaluation is undertaken to explain its occurrence.

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.

In continuing pregnancies, it is important to monitor the fluid in the tubes of the brain, curvature of the spine, lower limbs and the overall growth pattern of the baby. At times, the head of the baby enlarges due to blocked drainage of fluid in the brain resulting in a condition called hydrocephalus.

Your caregiver may refer you to genetic counselling and genetic testing. This may include tests like amniocentesis or chorionic villus sampling (CVS) to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing. Such testing can provide essential information regarding your individual case. 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 diseases are more likely if there are other problems detected on the ultrasound. Many women will receive genetic counselling 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?

The outcome is extremely variable, depending on where and how much of the spine is affected and on whether 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 amounts of fluid in the brain, clubfoot and curved spine are associated with more problems after birth.

If your baby is diagnosed with spina bifida, you would be referred to a specialised unit who will be responsible in holistic postnatal management.

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. The initial surgery is performed within 48 hours of life to prevent further damage to the nerves and infection. The surgeon will reposit the neural tissue back into the spinal column and close the defect with muscle and skin. The defect can be sealed but the preceding nerve damage and the consequent bladder, bowel and muscular dysfunction will persist.

Many will require multiple surgeries as they grow up, including corrective surgeries for club feet and even hydrocephalus.

Long term management also includes physiotherapy to reduce the stiffness of joints and mitigate muscle weakness. Urinary and bowel incontinence can be seen in some cases which may require specialist care.

Will it happen again?

The risk of having another child with spina bifida or similar types of problems is around 2-4%. Usually prenatal vitamins have 400 micrograms (400ug) of folic acid. But a higher dose of folic acid 4 milligrams (4mg) is recommended for reducing the risk of spina bifida in the next pregnancy. Women on drugs for epilepsy and obesity are also advised to take a higher dose of folic acid. It is usually advised to take folic acid for 3 months preconceptionally and continue further in the first trimester to reduce the risk of spina bifida, by about 75%.

Rarely, the risk will be much higher because certain genetic causes were found. A doctor with expertise in genetics can help you assess 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?
- Can the pregnancy be terminated?
- 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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