SBA National Resource Center:
800-621-3141

SPINA BIFIDA ASSOCIATION

A Guide for Medical Professionals

Delivering the Diagnosis to Expectant Parents

It is never easy or pleasant to give expectant parents news that their pregnancy has a fetus affected by Spina Bifida (SB). In order to help you provide the best care possible for your patients, and to help expectant parents fully explore the options they have, the Spina Bifida Association (SBA) wants to ensure that you have the most current and accurate information about Spina Bifida.

What you may remember about SB does not necessarily reflect the advances in medical and psychosocial care over the last generation.

- It is currently the most common permanently disabling birth defect, that is compatible with an adult life expectancy.
- For prenatally diagnosed patients, motor function may be better if a baby with SB is delivered at 37 weeks by elective C-section before a trial of labor (Luthy et al, 1991). This practice is recommended by many SB clinics.
- Fewer than 5% of pregnancies affected by isolated neural tube defects will result in a stillborn child or preterm delivery.
- Because of new treatments, surgeries, and therapies, at least 75% of individuals with SB live into adulthood (Bowman & McClone, 2001).
- The majority of people living with SB are adults. Most of them have ventriculoperitoneal (VP) shunts, but their life expectancy is not yet known. While there are adults with SB living into their 60's and 70's, those individuals predate the development of the VP shunt.

What is Spina Bifida?

Spina Bifida is a general term used to describe a variety of different neural tube defects which include Spina Bifida Occulta (SBO), a closed neural tube defect, and Spina Bifida Cystica (open neural tube defect). The most common open neural tube defect is Myelomeningocele. When most people speak of Spina Bifida they are referring to Myelomeningocele.

What causes Spina Bifida?

Spina Bifida is caused by a complex interaction of both genetic and environmental influences that ultimately affect any given pregnancy. The genetic influences remain poorly understood. There are a number of environmental influences which have been identified, though the exact interactions of environmental factors with the genetic influences are also not well understood. The most common environmental factor studied has been folic acid (Hernandez-Diaz, Wetler, Walker & Mitchell, 2001). Other factors include: use of seizure medication during pregnancy (Hill, Wlodarczyk, Palacios, & Finnell, 2010), maternal hyperthermia during early gestation (Suarez 2004), and maternal obesity (Stothard, Tennant & Bell, 2009). It is known however, that addressing all of these environmental factors before conception does not eliminate all possible causes of SB.

Current recommendation by the Centers for Disease Control and Prevention (CDC) state that all women of childbearing age should take a daily multivitamin with folic acid (400 micrograms). Furthermore, women who have had a previous pregnancy affected by SB should

take 4 milligrams of folic acid at least one month prior to conception, and through the first trimester (CDC 1991; CDC 1992). Unfortunately, even if all women did this, approximately 30% of cases of SB would still occur.

Medical Issues

- Hydrocephalus- Almost all individuals with SB develop hydrocephalus due to the presence of the Arnold Chiari malformation. For some individuals it appears that they develop some degree of hydrocephalus in utero. For many individuals, hydrocephalus develops post-natally, after surgical closure of the back. In the past, most individuals with SB required placement of a VP shunt to treat the hydrocephalus; but VP shunts are associated with potential complications. Pediatric neurosurgeons are now looking at new treatment options that might eliminate the problems associated with VP shunts; and are questioning through research, whether all individuals who previously got VP shunts actually needed them.

- Mobility- This issue is dependent on the spinal level of the SB lesion. Most children are able to walk (though usually with the aid of orthopedic bracing and assistive devices such as crutches). Some are full-time walkers, but many walk short distances and choose a wheelchair or electric cart for long distances. It is generally only those with thoracic or high lumbar spinal lesions who end up using the wheelchair as their only method of mobility.

- Neurogenic Bowel and Bladder- This issue is common in SB because the nerves that control the bladder, anus and a small segment of the sigmoid colon are controlled by sacral nerve roots (S2-S4). For the majority, bowel and bladder continence is achieved by medical and/ or surgical management.

  In recent years, a number of treatments have been developed which allow individuals to achieve varying levels of functional continence. The value of functional continence is that it promotes skin health and facilitates inclusion of people with SB to a greater degree of societal participation.

- Intelligence and Learning Issues-Most people with SB have normal IQ scores but may have learning difficulties. These can often be addressed when parents and teachers understand the issues and work together.

  Despite having normal intelligence, many adults with SB still struggle to live independently. Research suggests that there are likely multiple reasons for this, including the occurrence and type(s) of learning issues present, family and individual beliefs and expectations, and potential lack of exposure to peer and societal experiences compared to their non-disabled peers.

Quality of Life in Adulthood

Research over the last decade has demonstrated that self-perceived quality of life for persons with SB does not appear to be influenced primarily by medical issues (though medical issues may influence quality of life at times of medical crisis or stress). Rather, studies have shown that for most individuals with SB, quality of life is more strongly associated with the individual's beliefs and experiences, and that the family also influences the individual's beliefs in his or her capabilities. (Sawin & Bellin, 2010). Thus, lack of independence in persons with SB today appears to be more strongly influenced by learning issues that are inadequately addressed, and by negative attitudes and societal limits.

When Delivering the Diagnosis:

- Acknowledge your level of knowledge about SB. If you don't know, tell the patient you will find the information, or refer them to someone who does know. Choose your words carefully. This will likely be remembered as the worst day of their lives and your compassionate words and approach will echo in their memories.

- Avoid giving a definitive prognosis. There is a broad range of outcomes, and even with the most detailed information available, an accurate prognosis is difficult to provide. Giving parents the potential best and worst case scenarios will help the family begin to adjust to the shock of the situation, and allow them to process the information, so they can make the decision that is best for them.

- Be careful not to infer blame; it is no one's fault. While we know a number of factors that might be associated with SB, we really don't understand all the genetic and environmental factors and interactions that cause SB.

Families will be weighing all of their options: Termination or continuing the pregnancy. For some people, adoption may also be an option. For those who wish to continue the pregnancy, the option of fetal surgery exists, although in-utero repair is not a cure for SB.

Fetal Surgery

Fetal surgery as a treatment option began in the late 1990's and showed some initial promise related to some of the medical issues seen in SB, particularly the incidence of Arnold-Chiari malformation and hydrocephalus. The NIH- funded Management of Myelomeningocele Study (MoMS) began in 2003 and ended December 2010. The first results of that study were published in 2011 (Adzick & Spong, 2011). Outcomes were evaluated for 158 subjects at 12 months of follow-up. Findings indicated a reduced rate of VP shunt placement in the treatment group compared to the control group (40% versus 82%); and a decreased rate of hindbrain herniation (64% versus 96%). In addition, some improvement in motor outcomes was noted at 30 months (Adzick & Spong, 2011). However, there were noted potential complications, and the original article did not address all issues affecting individuals with SB, mothers, nor any longitudinal data on health outcomes. While the study shows promise for fetal surgery as a treatment option, some cautions remain, including the concern that fetal surgery may pose risks for subsequent pregnancies due to uterine damage. This is currently being studied.

SBA's Professional Advisory Council provided an additional response to this publication on SBA's Web site under their Published Research section.

In light of the complexity of an SB diagnosis, it is important that families have access to people who can provide accurate information about what to expect over the long-term in order to make an informed decision.

Referrals

- As soon as possible, refer the patient to a pediatric neurosurgeon, and/or to a SB Clinic to get information on longitudinal care issues.
- Ensure that the people you refer to have the most current diagnostic information (fetal ultrasounds or MRI scans). That will ensure that the consultants are able to give the most accurate information about what to expect in the future should families decide to continue with the pregnancy.
- Families would also benefit from referral to a geneticist or genetic counselor.
- Pregnant women should be referred quickly to a Perinatologist or Maternal-Fetal Medicine specialist. Families who wish to consider abortion will need to discuss that option fairly quickly and should discuss medical issues related to the procedure with their health providers.
- Women who wish to consider the option of fetal surgery will need immediate referral to a center experienced in this intervention. Fetal surgery is done prior to 25 weeks gestation. It is important to note that not all women with an affected fetus are candidates for the procedure.
- Let families know about Spina Bifida Association (SBA), which has many resources that are useful and informative to families. There is an information sheet available to expectant couples on the SBA website. The expectant mother or family can also contact the SBA National Resource Center with any questions. SBA can also help link families to a local chapter that can also provide resources and support.

For More Information

SBA offers a wide variety of in-depth health information sheets which cover all aspects of Spina Bifida care. They are available for download at www.spinabifidaassociation.org.

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This information does not constitute medical advice for any individual. As specific cases may vary from the general information presented here, SBA advises readers to consult a qualified medical or other professional on an individual basis.