Spina Bifida (SB)

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Definition

Spina Bifida (SB) is one of the "neural tube defects", a malformation of the vertebrae or backbones, which surround and protect the spinal cord, resulting in problems with the brain and spinal cord. The degree of disability resulting from this malformation varies according to the level/location on the spine, the extent of the vertebral bony opening, and the exposure of the spinal cord or brain. If just the membranous covering (the meninges) of the spinal cord is protruding through the vertebral bones, and the spinal cord is not entrapped, (a meningocele), the individual may have few symptoms. If however, both the membranous sac and a malformed cord are protruding (a meningomyelocele), a complex array of symptoms will be present.

Introduction

Malformations of the brain and/or spinal cord resulting in spina bifida occur very early in the pregnancy. We start out somewhat 2 dimensional/flat and by day 26 our "edges" come together to form the neural tube, which goes on to become the brain and spinal cord. If the "edges" don't come together well, a neural tube defect results. We don't fully understand why neural tube defects occur, but there seem to be both genetic and environmental risks

One child in every 1000 live births in the US each year is affected. Some children are born with a hidden spina bifida (spina bifida occulta) where there is a failed fusion of the vertebral bones surrounding the spinal cord. You might be able to detect tufts of hair, birthmarks, or cysts on the skin over the area of the defect. Usually these children appear to be perfectly normal. Later in life however, it is possible to develop bowel, bladder and musculoskeletal difficulties.

The next level of malformation is the meningocele or a protrusion through the vertebral/bony defect of a cystic sac full of cerebral spinal fluid/CSF (the fluid that acts as shock protection around the cord and brain). Sometimes this bulging sac results in hydrocephalus ("water on the brain" which is actually increased CSF pressure on the brain). In this situation a shunt or small tube will be inserted into the brain to drain the excess fluid into other parts of the body where it can be absorbed and not put undue pressure on the newly developing brain. Children with this condition have varying degrees of disability dependent on the level or location of the malformation on the spine, and on the severity of the defect.

When the spinal cord itself protrudes through the vertebral defect, a myelomeningocele, also called a meningomyelocele results. Almost all children with this defect will require a shunt for hydrocephalus. All will have significant developmental and medical issues. You will get to know "the team": pediatricians (in charge of all the specialists!), neurosurgeons (in charge of shunts), urologists (in charge of the bladder), orthopedists (in charge of the legs and hips), geneticists (in charge of helping you understand what we know about the causes of this malformation and your risks of having another child with spina bifida), psychologists (in charge of keeping you sane when the going gets tough), and many wonderful nurses and therapists and social workers. Sometimes it can feel overwhelming, but you and they all want the same thing: what's best for your child! And things do get easier over time as you learn, and as things settle down into a routine.

Diagnosis

Significant spina bifida is obvious at birth. There will be a flurry of tests with ultrasounds, CT scans, and MRI scans. Then there will be surgery to close the defect in the spine, and probably to place a shunt. Then teaching will begin to

prepare you to work with your child in the day to day care he or she will need. Meanwhile the team will be watching closely to see the extent of neurologic involvement. This information is important to determine what interventions will be most helpful for you and your child, and to guide information for you about what you might be able to expect. You can expect and should ask for recommendations to get your child into an early intervention program (to keep an eye on and encourage his or her development in spite of the disability.) You should also ask to have a social worker/case manager to assist you with the myriad of appointments and mounds of red tape. And don't forget to ask for a psychologist to help sort out some of your grief and stress, and some of the parenting issues.

Treatment

Because spina bifida is not curable, we treat according to the problems that the child shows us. The goal of treatment is to be sure that your child achieves the maximum possible level of motor, intellectual, and social functioning. The following section will introduce you to common associated problems that children with spina bifida can have and some of the current treatments.

Toileting issues – this is usually one of the first hurdles for parents in dealing with their child's day to day care. Both the brain and spinal cord are involved in both urinating and bowel movements. Therefore, both are affected in children with spina bifida, and both need to be addressed from birth for the child's good health. You will not hurt your baby with the clean intermittent catheterization techniques that you will learn to help with urination. And you will be taught techniques to help avoid constipation. It will all become a "normal" part of daily hygiene Your child will learn these same techniques as she or he gets older. Usually a physical or occupational therapist can assist with supportive equipment needs. Sometimes medications are helpful if catheterization alone does not keep your older child dry. At your pediatrician's office be sure that your child's blood pressure is checked routinely, especially if there have been a lot of bladder/kidney problems. If the readings are persistently elevated, see your child's urologist.

Hydrocephalus – as discussed above, your child may need a shunt to keep some of the excess CSF pressure off the brain. For the first few years your child's pediatrician and neurosurgeon will be measuring the head size regularly to make sure your child's shunt is draining well. They will ask you to bring your child in if he or she becomes lethargic, has headaches, vomiting, or irritability, since these can be signs of fluid/pressure increasing in the brain. Of course you would know to bring your child in immediately if she or he had a seizure for the first time. New onset of "crossed" eyes, fever, longer or more frequent naps, a change in personality or school performance or increased weakness of the arms or legs can also be subtle cues of a malfunctioning shunt. If your child does have a shunt be sure to ask about antibiotics before any dental procedures that may result in bleeding. Be aware that your child may have a hypersensitivity to loud noises if shunted.

One of the first "advocacy" issues for your child will be to make sure all contact with your child is "latex-free". More than half of the children with spina bifida has an allergy to latex. Catheterizations should be performed with non-latex catheters and non-latex gloves. Latex toys such as rubber balls and balloons should be avoided. Watch too for the latex in bandaids and ace wraps.

Just as the nerves from the spinal cord carry messages to the muscles about motor/movement, so too nerves from the spinal cord carry messages to the skin and "insides" about sensory/sensation. Both motor and sensory functions below the level of your child's malformation/lesion are affected.

Motor losses may include paralysis, weakness, or spasms of the legs (spasticity). Many infants are delayed in their motor milestones: rolling over, sitting, crawling, and walking. The ability to walk is not only determined by the level of the lesion and associated weakness, but also by your child's intellectual/cognitive functioning, the involvement of the parents, and the therapy program.

Sensory losses are also important in your child's development and overall health. Senses include tactile as well as positional and pressure. If your child cannot feel a fullness in his or her rectum, they will have a harder time making it to the potty on time. If he has a hard time sensing where he is in space, balance for sitting and walking will be more difficult. If she does not feel the numbness and pain of sitting too long in one position, she will develop pressure sores (decubiti). Even fractures may not be felt. These last two problems are good reasons to do daily skin checks.

What will help with motor and sensory losses? They cannot be regained, but you and your child will learn to work around these losses with physical and occupational therapy.

In the meantime recognize that your child doesn't feel pain below the level of the lesion in the same way you do. Be careful to avoid tight-fitting shoes or braces. Check to be certain bath water is not too hot. Watch to be certain that a leg isn't resting against a heater or cigarette. Be sure there are frequent re-positionings. Check skin daily.

Mobility is a major concern for parents. Will he or she walk? As a rule (but there are exceptions to every rule, and rules are made to be broken!), children with sacral level lesions learn to walk well by 3 years old, sometimes requiring ankle supports. Children with low-lumbar lesions usually require crutches and bracing. Those with thoracic and high-lumbar level lesions have the greatest weakness and the hardest time walking. Even ambulatory children with level L3-4 lesions usually prefer wheelchairs by adolescence. Remember the goal is mobility, not necessarily walking. Be realistic about your child's desire to move, explore, and keep up with friends. Sometimes this is best accomplished with a wheelchair.

Obviously, another major concern of parents is will he be mentally retarded? This is another example of rules meant to be broken. Most children have an IQ in the average range. Those with hydrocephalus tend to have an IQ in the low average or below range. Complications with shunt malfunctions, and infections obviously add insult to injury and may limit intellectual function. What is important to look for and have your child tested for are learning disabilities, poor memory skills, organizational and attention deficits, processing problems, impaired speed of motor responses, and poor eye-hand coordination. These can stymie learning if not recognized and addressed. Therefore the school is responsible for testing and adapting the classroom to meet your child's specific learning needs. Ask for your child to have neuropsychological testing to assess his academic strength and weaknesses. This is where your advocacy skills are critical.

Visual problems are present in at least 20% of children with this disability. Most commonly the problem is strabismus (or a wandering or crossed eyes). If your child still has this problem by nine months of age, be sure to consult an ophthalmologist.

Remember we said above that the orthopedist would be part of your child's team? Here's why: because of total or partial paralysis, the muscles are imbalanced and mobility is impaired, sometimes even before your child is born. So children with spina bifida commonly are born with a clubfoot, or develop dislocated hips, or scoliosis of their spines. A good physical therapist will keep you informed of how the muscles and joints are doing, help you with positioning your child, and make sure the muscles don't tighten the joints into contractures. You will also learn about standing frames and parapodiums to assist your child to stand. You can keep your child's pediatrician informed. Be sure your child's pediatrician checks for scoliosis annually from birth through adolescence. Usually you will see the orthopedist if your child has a problem like these and/or if bracing and/or casting are indicated.

Be sure your pediatrician is checking your child's length and weight. Children with spina bifida are at increased risk for obesity. Not only is this unhealthy, but it makes both self-esteem and mobility even more difficult. It can also lead to problems with decubiti and with fitting braces. Your child is less active and

expends fewer calories. Do not overfeed. Be sure your child is drinking lots of fluids to decrease constipation and bladder/kidney infections. Lots of fiber helps too with issues of constipation. Ask your doctor to refer you to a nutritionist to help you keep your child's diet healthy. Your goal is to avoid obesity. Short stature is common in children with spina bifida.

A possible complication of spina bifida is a symptomatic Arnold Chiari II malformation. Most children with spina bifida have this malformation, but it rarely becomes symptomatic, causing compression of the spinal cord. In this abnormality the lower parts of the brain (brainstem and part of the cerebellum) are displaced downward toward the neck. This becomes symptomatic only if it begins to cause compression on the lower parts of the brain and the spinal cord. Signs of compression include poor feeding and prolonged feeding times, difficulty swallowing or breathing, choking, hoarseness, snoring, a croupy cough, or breath-holding spells. These symptoms should be brought to the attention of your pediatrician and/or neurosurgeon. Most children with spina bifida never develop this complication.

You may not be ready for the next topic, but believe me your child will be, and sooner than you think! Yes, sexuality! If you and your child have done a good job with achieving independence, a sense of capability in school and at home, and good self-esteem with supportive peer relationships, then the obvious next step is dealing with sexual changes and feelings -- and questions. Be prepared!

Once again there are exceptions to every rule. As a rule, about 75% of boys/men with spina bifida can have erections. Many however, have retrograde ejaculations, meaning that the semen is not expelled from the penis, but "backtracks" to the bladder. There are lots of options for obtaining erections, but fertility is not so easily predictable in males.

Girls/women on the other hand, have normal fertility and an approximate 4% risk of having a child with a neural tube defect (spina bifida, encephalocele, or anencephaly). Be sure your daughter has access to a genetic counselor to clarify her risks. The risk is the same as the general population for sexually transmitted diseases, and pregnancy. Females with spina bifida may experience less sensation and fewer if any orgasms. Lubrication is also diminished. Birth control is a difficult topic. Because of the increased risks of blood clots, hormonal contraceptives can be risky. Urinary tract infections are more likely, and IUDs can increase the risk of pelvic infections. Latex condoms and diaphragms are out because of latex sensitivities. Try to help your daughter find the resources she needs to make wise choices.

Emergency Situations – What can go wrong?

After all of that what else could possibly go wrong? Well don't worry, we've talked about most of it.

We have already discussed signs and symptoms of shunt malfunction and/or infection. Shunt problems are emergencies, as are any changes in neurologic ability or function. (Sometimes the problem is at the "tail" end with a tethered cord: look for things like back or abdominal pain that begins abruptly or during sleep, increased sensitivity to local pressure on the spinal column, or change in toileting skills, or sensation.)

Seizures in and of themselves are not emergencies if they have been previously diagnosed, and if you have been instructed on what to do. If however, your child is having a first seizure or a prolonged seizure (greater than 5 minutes by the clock), it is considered an emergency.

Your child is at increased risk of an accident or injury not only because of decreased motor and sensory ability, but possibly because of slower mental processing, or poor attention, or poor judgment. Like any child, you cannot protect them from themselves at all times. If there's a threat of loss of life or limb because of the accident, it's an emergency.

Conclusion

Spina bifida is one form of neural tube defect, resulting in a vertebral bony defect with either the meninges, or the meninges and spinal cord protruding through the defect. This malformation can have significant impact on growth, development and lifelong adaptations. Depression, alcohol and/or drug abuse, obesity, skin breakdown, osteoporosis, kidney failure, underachievement at school and in adult life can all become secondary disabilities. However, with early intervention, education of the child and the parents, and good community support, the prognosis for these children is very good. Many will go on to college, to jobs, and to happy, satisfyingly full lives. Support from "the team" and the school is essential to the mental, emotional and physical health of the individual and the family.

References

Primary Care of the Child with a Chronic Condition by PL Jackson and JA Vessey

Children with Disabilities by ML Batshaw, MD

Spina Bifida Association of America
1-800-621-3141

National Organization for Rare Diseases
1-800-999-6673

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