

Spina Bifida and Hydrocephalus Explained

Introduction

Spina bifida comes from the word for 'split spine' in Latin. It is one of a class of serious birth defects, called neural tube defects (NTDs), which involve damage to the bony spine and the nervous tissue of the spinal cord. Some vertebrae of the spine don't close properly during development and the spinal cord's nerves don't develop normally. They are exposed and can be subjected to further damage. At birth, they protrude through the gap instead of growing normally down the bony spinal column. Nerve signals to most parts of the body located below the level of the 'split spine' are damaged and a wide range of muscles, organs and bodily functions are affected. The other main type of neural tube defect is anencephaly in which the brain and skull don't develop properly. All babies with anencephaly will either be stillborn or die soon after birth.

Varying effects

People are affected by spina bifida (SB) in a variety of ways, ranging from minor to severe:

- Legs and feet – a range of walking difficulties (through to an inability to walk); reduced sensation; proneness to burns and pressure sores.
- Bowel and bladder – some level of urinary and faecal incontinence; increased stress on the kidneys; some level of sexual dysfunction.
- Brain – in most cases, the baby has hydrocephalus (a build-up of cerebrospinal fluid in the brain) and the Arnold Chiari malformation (the brain stem physically 'jams into' the spinal cord). These abnormalities may cause many different brain function disabilities.

One in 1,000 pregnancies are affected

The risk of spina bifida is approximately one in every 1,000 pregnancies. It is caused by a combination of genetic and environmental factors, which are not yet fully understood. Inadequate metabolism of folate in early pregnancy is a significant factor in the occurrence of this condition. The number of babies born with spina bifida has dropped dramatically in recent years, due to improved ultrasounds and other tests, which detect the condition and provide the choice of pregnancy termination. There is no cure. However, the vitamin folate can prevent up to 70 per cent of spina bifida cases, if taken by the mother one month before conception and during the first three months of pregnancy.

What causes spina bifida

The brain and spinal cord of a foetus develop during the first 28 days of pregnancy. For reasons not yet fully understood, something sometimes goes wrong and spina bifida develops.

Spina bifida occurs when the spinal cord (nerve tissue which connects the brain to the rest of the body) does not form correctly at some point along its length.

In most cases, the meninges ('skins' that cover the spinal cord) are also damaged, leading to a form of spina bifida called meningocele.

In more severe cases, the vertebrae (small round bones which make up our spinal column or backbone), the meninges and the spinal cord are all damaged; this form of spina bifida is called myelomeningocele.

The most serious form is anencephaly (limited brain development) which babies do not survive.

These types of spina bifida are all types of a condition called neural tube defects (NTD).

We know that spina bifida has both genetic and environmental causes, but do not fully understand what these are, or how they operate together. It is very important that women considering pregnancy ask their doctor about prevention measures, especially if someone in her family, or her partner's, has spina bifida.

Detection

Well over 90 per cent of cases of spina bifida should be detected with a good quality ultrasound at 18 weeks. If present, specialist gynaecological care occurs until birth. If first detected at birth, there will be a large soft lump or lesion on the back. This lump contains spinal cord nerves and tissue. Exposed nerves must be surgically placed gently back under the skin within 24 hours.

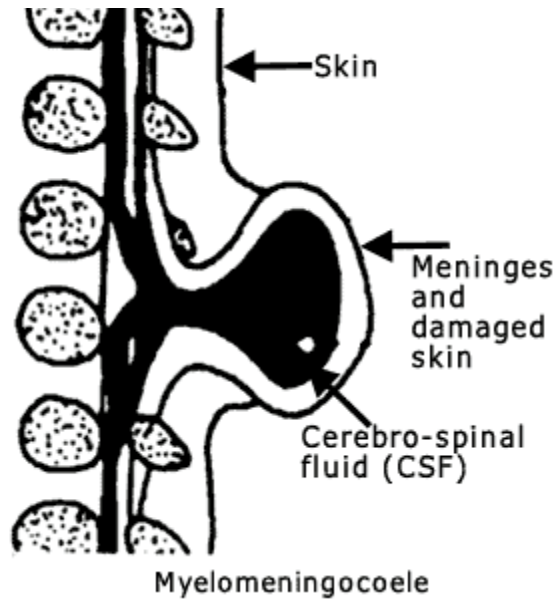
The spina bifida lesion

The nervous system of a growing foetus starts as a simple structure called the neural plate. This plate quickly becomes the baby's brain, and the spinal cord and neural tube that enclose it. By day 28, the neural tube should have closed and fused. If it doesn't close, the result is a neural tube defect. Spina bifida can occur at any place along the spine. However, surviving babies are generally affected lower down the spine because, at higher levels, the survival rate is low. The exact cause is not completely understood, but it appears that a combination of genetic and environmental factors is responsible. Inadequate metabolism of folate in early pregnancy is an important cause.

Babies with spina bifida are born with a lesion (lump) where the spinal cord has not properly formed.

This lesion is closed by a neurosurgeon, usually immediately after birth, to return the damaged spinal cord to the body in as normal a position as possible. This protects it from further damage or infection. It is not a cure; it is the crucial first step in a complex management process.

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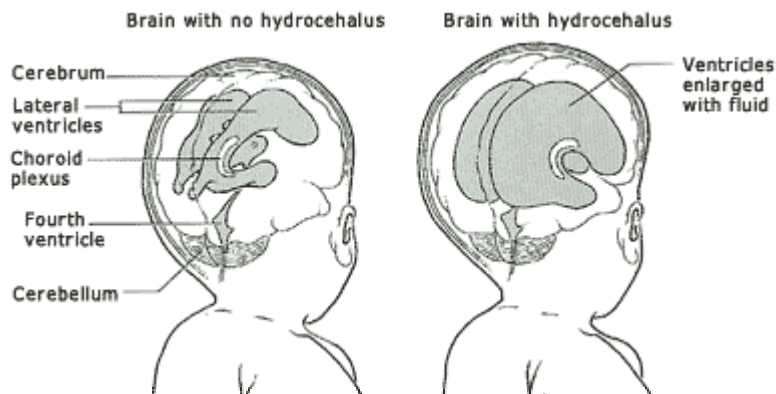
Hydrocephalus & the Chiari Malformation

The Arnold Chiari Malformation occurs when the lower part of the brain is positioned abnormally, resting further down into the spinal column than it should be, causing several problems. This is a very commonly associated condition of spina bifida.

The main problem the Chiari Malformation causes is hydrocephalus (the inability of the body to drain the spinal fluid produced in the brain). It does so by blocking the normal flow of cerebrospinal fluid or CSF (the fluid in the brain and spinal cord). It accumulates as it cannot be drained away.

Often a baby will be born with hydrocephalus, with the brain's ventricles already enlarged with fluid. In other cases, the problem begins after the lesion on a newborn's back is closed by surgery.

Over 80% of all people with spina bifida will need a special device called a 'shunt' implanted, to help the fluid drain away. Because so many people with spina bifida have hydrocephalus too, spina bifida is often called spina bifida hydrocephalus or SBH.

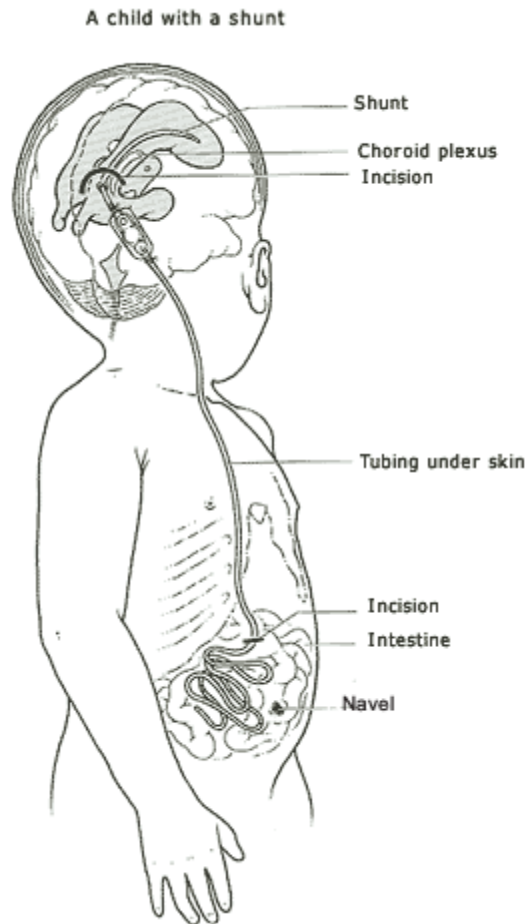


A Brain without hydrocephalus compared to a brain with hydrocephalus

Spina Bifida and Hydrocephalus Explained

Shunts

A shunt is a long narrow tube with a one-way valve. Inserted under the skin, it simply and effectively drains the spinal fluid to a convenient place - usually the abdomen. It also equalises pressure within the body.



An example of a shunt in place

Modern shunts work very reliably and blockage and infection problems have been minimised. However, shunts will block at some stage and require replacement or revision in another operation.

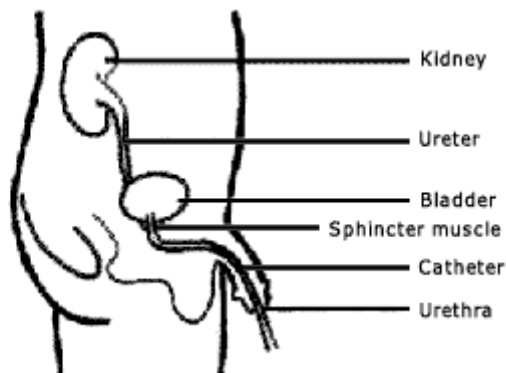
Increased head size, fixed staring, irritability, vomiting, sleepiness or a bulging fontanelle (soft spot in a baby's skull) are all signs of possible shunt problems in newborns. For older people, seizures, continence changes, headaches, poor performance or emotional changes are the usual signs.

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Bladder problems

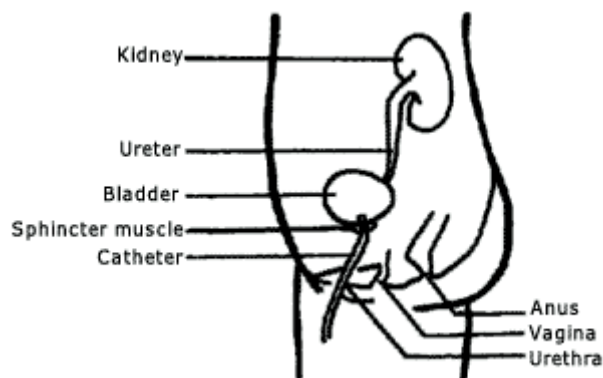
People with spina bifida usually have normal kidneys (which clean the blood and flush away the waste as water). The nerve damage caused by spina bifida means, for almost all persons, that some degree of urinary incontinence is present. It has many forms, however the inability to stay dry and increased pressures in the urinary system are major problems. This is a complex and difficult area to manage, and surgery, drugs and alternative toileting techniques are all involved in its management.

Children with spina bifida need special toilet training, especially to use a catheter, a device a lot like a straw. At first parents, and then later the children themselves, learn how to insert the catheter through the urethra (bladder opening) and into the bladder, to empty it at regular intervals each day.



Catheter for a male

Clean intermittent catheterisation has effectively managed incontinence in the majority of cases and enabled social continence to be achieved.

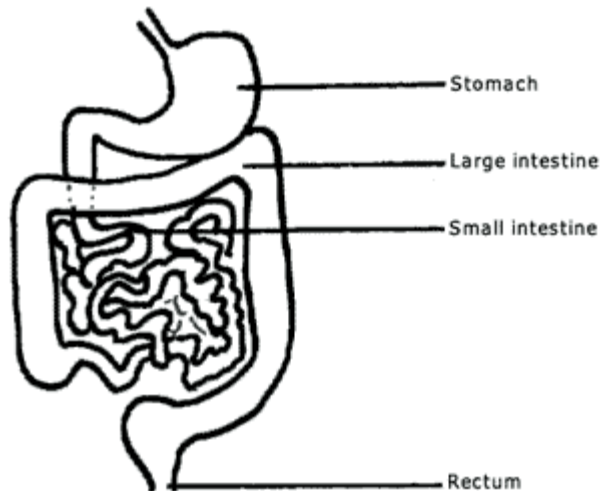


Catheter for a female

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Bowel problems

The food we eat passes from the stomach into the small intestine as liquid stool. From there, it enters the large intestine or colon or bowel where water is absorbed, and from there it passes through the rectum and anus as solid stool or faeces when we go to the toilet.



The Bowel

For most people with SBH, this function is impaired, again due to incomplete nerve signals. The results are:

- they may not know when it is time to go to the toilet
- they often have limited control over when their bowel will empty, and
- they must work hard to prevent constipation (the stool moving too slowly through the colon, losing too much moisture, and becoming too hard)

Normal toilet training is not possible for children with SBH. It is important for people with SBH to avoid constipation. With diet management and with careful long term training utilising what sensation exists, over time, social continence is achieved.

As it is difficult for anyone to become fully independent if they have bowel accidents, this aspect of SBH requires enormous commitment and care.

Lower limb paralysis

The extent of the lesion provides a good estimate of the extent of lower limb functioning. (see diagram and chart below)

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Of course, every individual is different, and each person with SBH is affected slightly differently.

Because nerves are affected at and below the point at which malformation occurs, the higher up the spine it occurs, the greater the paralysis (immobility) of the lower limbs will be.

Most people with SBH will need leg braces or more help to walk. Braces can help to support and protect weak muscles or joints.

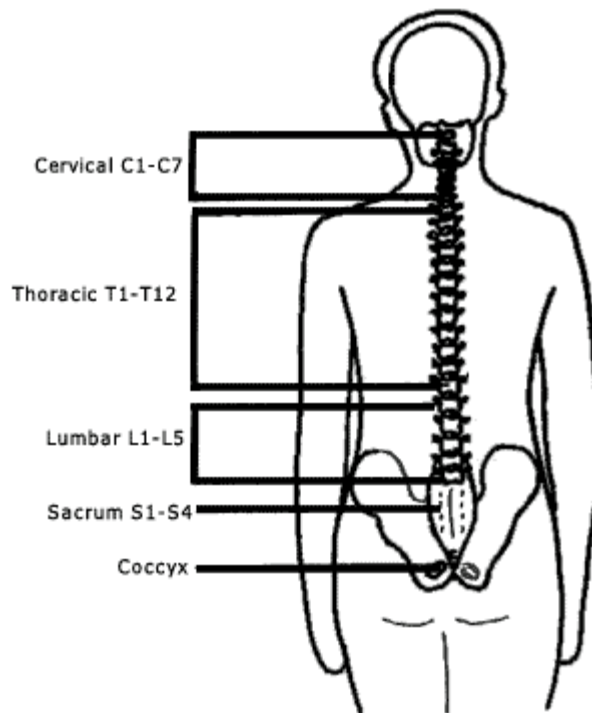
Sometimes people will use braces or crutches for short distances, and use a wheelchair when they are more likely to get tired, or to leave their hands free.

Most individuals will need one or more orthopaedic (bone or joint or soft tissue) operations over the course of their life to assist in maximising function.

The level of paralysis should not change as the person with SBH gets older. A range of problems, however, commonly occur throughout life. These include:

- scoliosis (spinal curving due to muscle imbalances) which requires surgery.
- tethered cord (the scar tissue where the lesion has been repaired "sticks", not allowing the spinal cord to move) which causes a range of problems and also requires surgery.

Lesion level and its effect on mobility



The Vertebrae

Lesion Level Effect on Mobility

T12 & above With braces, can only walk short distances; with walker or crutches, slightly longer. Will mostly use a wheelchair, even in childhood.

L1 - L3 Leg braces with a waistband; will use crutches. Wheelchair for distances

L4 Will usually need braces, perhaps above the knee; crutches or cane; wheelchairs when older

L5 - S Short leg braces; may need crutches or cane.

Allergies, skin care

People with spina bifida appear to have a higher than average risk of developing an allergy to latex (natural rubber). As with many allergic reactions, symptoms are sometimes limited to itchy eyes, runny nose or skin rashes. Severe allergic reactions, however, can be life threatening as the ability to breathe is affected, so people with spina bifida should avoid exposure to latex in any form.

It is important for people with SBH and their carers to treat this potential allergy as a serious risk. Many activities of daily life, from visiting the dentist, washing dishes, to playing with toys brings us into contact with latex.

Doctors and surgeons should wear non-latex gloves and non-latex catheters should be used.

Because people with SBH have impaired sensation below the level of the spinal lesion, they need to take special care to avoid extremes of temperature, to prevent frost bite and prevent burns from every day sources such as hot water, heaters, hot bitumen and even sunburn.

The skin of people with SBH heals more slowly than is usually the case, and skin can easily be damaged from something as simple as crawling on the floor, sitting too long in one position, or from badly fitting foot braces.

Pressure sores can easily form and these are extremely serious for a person with SBH. Even a small sore will require 2 or 3 months careful rehabilitation.

Unattended, these sores will require lengthy hospitalisation and extensive plastic surgery, as deeper level skin tissue and muscle becomes affected.

It is important that careful attention to skin care becomes part of the daily routine.

Seizures, eye problems, fractures

People with SBH have a higher than average risk of epilepsy (seizures or convulsions). This should be monitored closely.

In addition, it is common for young children to have strabismus (lazy eye), and it is more common for children with SBH.

If strabismus does not correct itself within a baby's first six months, an eye specialist should be consulted, as failure to treat the problem could lead to a permanent vision impairment.

Strabismus can also be a symptom of shunt failure, or hydrocephalic pressure on the optic (eye) nerve.

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Persons with SBH are also prone to leg fractures and may not, due to impaired sensation, identify immediately that one has occurred.

Education, sport and exercise

In general, the average IQ score for people with SBH is about 10% lower than the general population average.

Some people with SBH have an intellectual disability while others succeed in tertiary education. The range of achievement of individuals is very diverse.

Most children with SBH today attend mainstream school, however, most will have some learning difficulties, caused by the two partners of spina bifida - hydrocephalus and the Arnold Chiari malformation.

These difficulties might be in one or more of the following areas:

language, memory and learning, visuo-motor integration skills, planning and organisational skills. The companion publication to this one, *Educating the Student with Spina Bifida and Hydrocephalus* explains this important area in detail.

It is vitally important that these 'hidden disabilities' are identified and evaluated in individuals, to allow effective intervention. Specific learning difficulties must be managed effectively to ensure learning does occur, and eventually employment success and independence are achieved.

Because people with paralysis will be less active than mobile people, there is a risk of obesity (being overweight), as well as high blood pressure, atherosclerosis (heart disease) and osteoporosis (weak bones).

It is essential that people with SBH be encouraged from a very early age to develop regular exercise habits.

Independence and social development

Children born with spina bifida are children first, with a disability second.

To grow into responsible adults they must learn what every other child needs to learn; to make choices and decisions, to accept responsibility for their actions, to get along with other children and so on.

Their social development will be interrupted by frequent visits to hospitals and specialists, and they will often have mobility problems which prevent them from joining in activities at school, as well as specific learning difficulties.

The chances of a person with SBH leading a normal, rewarding and fulfilling life will depend on how those around them strike a balance between being over-protective, and asking more of them than they are able to give.

It is important to the development of all children that they be given opportunities to succeed, and to grow in self-confidence. It is normal for growing people to experience self-doubt or fears about how well they fit in with their peer group, and it is desirable that children with spina bifida not only socialise with children without disabilities, but that they also have a chance to socialise and talk to other children with disabilities to share their experiences.

Puberty, sexual issues

Precocious puberty (early sexual maturity) is more common in children with spina bifida, with some girls commencing puberty as early as 8 or 9 years of age.

Because the long bones in the arms and legs stop growing during puberty, this means that affected individuals may be shorter than they could have been.

Treatment to delay the onset of puberty may be required.

Damage to the lower part of the spinal cord means that some sexual function is lost.

Females may not have sensation in the clitoris and therefore not achieve orgasm with intercourse. However, they can have increased sensitivity in other areas and can have satisfying sexual lives.

Women with spina bifida are normally fertile, but may have a level of disability which makes it difficult for them to carry a pregnancy for nine months. Despite the problems, many women with SBH successfully bear children. It is important for women with SBH to seek medical advice when they are planning a family.

Some males with SBH are able to sustain an erection, though ejaculation (discharge of sperm) does not always occur, and males may have difficulty in fathering a child. Like females with SBH, males with SBH are able to manage a healthy sex life.

Like everyone else, people with SBH should be responsible for protecting themselves and their partners against AIDS and other sexually transmitted diseases, or against unwanted pregnancy. As there is a high risk of people with SBH having allergy to latex, they should seek advice before using condoms or other contraceptive devices.

Preventing spina bifida

As mentioned previously, we know that spina bifida is caused by both genetic and environmental factors, but we do not fully understand the mechanisms at work.

We do know that women who take folate (a common vitamin found in green leafy vegetables and in grains) one month prior to, and three months into their pregnancy, will reduce their chance of having a baby with spina bifida by 70%.

The National Health and Medical Research Council now officially recommends 500mcg of folate supplementation for women seeking to have a family.

The amount of folate supplementation needed for women in the 'high risk' groups - women with spina bifida or who have a close relative with spina bifida - is much higher.

In addition, there are two very effective ways to detect if the unborn child has spina bifida:

- specialist ultrasound at certain stages of pregnancy, and
- a simple chemical test called the Alpha Feta Protein (AFP) test.

It is very important that specialist medical advice is sought on all the above matters from expert doctors.