Spina bifida

Summary

- Spina bifida refers to a range of birth defects that affect the spinal cord.
- In spina bifida, some vertebrae of the spine aren’t closed, leaving the spinal cord nerves exposed and damaged.
- The recommended dose of folic acid, taken daily one month before conception and during the first three months of pregnancy, will greatly reduce your chances of having a child with a neural tube defect.

Spina bifida is Latin for ‘split spine’. It is one of a class of serious birth defects called neural tube defects (NTD). It is an abnormality of the folding of the posterior surface of the embryo, which normally forms the vertebral column with its muscles and the spinal cord and the spinal nerves.

Because of this abnormality, the growing embryo does not develop normally and the spinal cord and nerves are exposed on the surface of the back, instead of being inside a canal of bone surrounded by muscle. This means that the spinal cord and nerves can be easily damaged.

Almost always, the nerves supplying the parts of the body located below the level of the exposed area do not function properly, leading to a range of motor and sensory problems, and disturbance of bodily functions, such as bowel and bladder.

The other main type of NTD is anencephaly in which the brain and upper part of the skull are not developed properly. All babies with anencephaly will either be stillborn or die soon after birth.

Pregnant woman or women planning to become pregnant should take folic acid regularly to reduce the risk of the fetus developing spina bifida.

Most cases of spina bifida are detected before birth. Spina bifida cannot be cured, but a range of treatments and management options is available.

Diagnosis of spina bifida

Approximately 90 per cent of cases of spina bifida are detected with an ultrasound scan before 18 weeks of pregnancy. Other tests used to diagnose spina bifida are maternal blood tests which measure alpha-fetoprotein (AFP), and magnetic resonance imaging (MRI) scans.

If spina bifida is present, specialist obstetric care and support will be provided. Consultation with an expert paediatrician is available at both the Royal Children's Hospital and Monash Children's Hospital.

In open spina bifida where the cord and nerves are exposed (called spina bifida aperta), it is important to close the defect within the first few days of life to avoid infection, excess drainage of cerebrospinal fluid and further damage to the spinal cord and nerves.

Occasionally, spina bifida is not detected until birth when a large soft lump or skin covered lesion on the baby’s back is noticed. This lump contains spinal cord, nerves and often fatty tissue (called a lipomeningocele). The need for surgery in this situation is not urgent, because the spinal cord and nerves are not exposed.

Symptoms of spina bifida
The effects of spina bifida vary according to the type, location and severity of the condition. It can be located in the neck, chest or lumbar spinal region. The low thoracic upper lumbar lesions (in the mid-back area) generally produce a greater degree of paralysis and other debilitating complications.

Problems associated with spina bifida include:

- reduced sensation in the lower body, legs and feet, leading to the possibility of burns and pressure sores
- a degree of paralysis of the lower body and legs, causing walking difficulties or inability to walk
- different degrees and types of urinary incontinence
- different degrees and types of faecal bowel incontinence
- some sexual dysfunction, particularly related to penile erection and ejaculation
- deformities of the spine – commonly scoliosis, where the spine bends into an ‘S’ shape
- cord tethering, where the spinal cord sticks to the area of the original lesion and becomes stretched
- Arnold Chiari malformation – an abnormality of the back of the brain and upper spinal cord which can cause disturbance of breathing, swallowing, eye movement and fluid flow leading to hydrocephalus
- learning difficulties.

Spina bifida and hydrocephalus

The brain and spinal cord are bathed in and nourished by cerebrospinal fluid. Most people with spina bifida have the Arnold Chiari malformation and about 80 per cent have abnormality of cerebrospinal fluid flow causing hydrocephalus (Latin for water on the brain).

Hydrocephalus may be managed early with a shunt if adequate absorption of fluid does not occur. This shunt drains the fluid away from the brain to parts of the body where no damage can be done. The brain looks structurally different in people with spina bifida, but it can function normally. It is not uncommon, however, to have some brain function disability.

Causes of spina bifida

Neural tube defects (both anencephaly and spina bifida) are caused by genetic and environmental factors that are not yet fully understood. The risk of these conditions is approximately one in every 800 pregnancies. Inadequate intake of folate by the mother in early pregnancy is a significant factor in the occurrence of spina bifida.

The number of babies born with spina bifida in Australia has dropped dramatically in recent years due to greater awareness and intake of folate by women prior to and in the early stages of pregnancy.

Improved ultrasound and other tests that detect spina bifida and provide the choice of pregnancy termination have also reduced its occurrence.

High-risk groups

People whose children are at high risk of spina bifida include those who have a:

- previous child with a neural tube defect (NTD)
- family history of NTDS on one or both sides
- close relative with an NTD
- close relative with a child with an NTD.

The children of women taking some anti-epileptic medications (such as valproic acid) also have an increased risk of spina bifida.
Folate can prevent spina bifida

Folate (folic acid) is a B-group vitamin. The recommended dose of folate, taken daily one month before conception and each day during the first three months of pregnancy, can prevent most neural tube defects.

The National Health and Medical Research Council recommends that all women planning a pregnancy or likely to become pregnant should take 0.5 mg of folic acid daily. People in high-risk groups should take a higher dose.

Good sources of folate include:

- folate supplements
- foods naturally rich in folate – asparagus, spinach, oranges, bananas and legumes
- foods fortified with folate, such as some breakfast cereals and bread. Look for the ANZFA Folate Enriched logo on the packet.

Treatment for spina bifida

There is no cure for spina bifida. Treatment options include:

- Surgery – may be used to close the lesion and reduce the risk of infection.
- Shunt insertion – hydrocephalus is treated with the insertion of a tube, called a shunt, into the ventricles in the brain where the spinal fluid is produced, allowing excess cerebrospinal fluid to drain out of the brain via another tube into the abdomen or the heart.
- Orthopaedic surgery – children with spina bifida usually undergo operations on their legs and feet to improve their mobility.
- Mobility aids – walking aids or wheelchairs are commonly used.
- Diet and enemas – are used to manage faecal incontinence.
- Bladder surgery – can increase bladder size and tighten muscles.
- Self-catheterisation and continence pads – may be required to manage urinary incontinence. Sometimes faecal or urinary bags are necessary.
- Regular monitoring of kidney, bladder, shunt and spine functions.

Where to get help

- Your doctor
- Spina Bifida Clinic at the Royal Children’s Hospital Tel. (03) 9345 5898
- Fetal Diagnostic Clinic Monash Health Tel. (03) 9594 2343
- Fetal Management Unit Royal Women’s Hospital Tel. (03) 8345 2000

Things to remember

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