

Produced by the Centre for Genetics Education. Internet: <http://www.genetics.edu.au>

Important points

- The neural tube is the embryonic structure that develops into the brain and spinal cord. Very early in a baby's development, a layer of cells folds over and 'zips up' to form the neural tube. If the neural tube fails to 'zip up' completely at some point, the spine or brain's development will be impacted, causing many babies to die and others to have problems with walking and with bowel and bladder control.
- Spina bifida and anencephaly are the most common forms of **neural tube defects** affecting about 1 in every 500 pregnancies and about 1 in every 600 births – the difference is due to the loss of pregnancies affected with neural tube defects.

Spina bifida

- Where the spinal cord and the protective sac that surrounds the cord (*meninges*) may protrude through the open part of the spine
- Symptoms vary depending on the position of the opening along the spine and on how much of the spinal cord, or the protective sac (*meninges*), protrudes through the spine
- The spinal abnormality may be so slight as to cause only minor effects; in other cases, surgery is possible but some degree of disability may remain
- Some affected babies, children and adults develop *hydrocephalus* or 'water on the brain' and the fluid needs to be drained through a special tube (called a shunt) that is surgically placed and runs under the skin, down into the chest or abdomen

Anencephaly

- When the neural tube fails to close at the head, the brain and the skull bones do not develop normally
- Infants born with this problem die at, or soon after, birth

Prevention

- In most women, it is possible to reduce the risk of neural tube defects by their taking the vitamin, folic acid, and other vitamins, in a specified dose at least one month prior to conception and continuing throughout the first few months of pregnancy (see Genetics Fact Sheet 19)
- If there is a family history of a neural tube defect, or the woman has epileptic seizures, she may need more folic acid (folate) than most women
- Genetic counselling (see Genetics Fact Sheet 3) can provide current information about the conditions, the preventive plan, assessment of their particular risk and discussion of the tests that are available to detect affected babies before birth (see Genetics Fact Sheets 17A & 17B)

What are neural tube defects?

The neural tube is the embryonic structure that develops into the brain and spinal cord. Very early in a baby's development, a layer of cells folds over and 'zips up' to form the neural tube.

If the neural tube fails to 'zip up' completely, this will have an impact on the development of the spine or brain.

Spina bifida and **anencephaly** are the most common problems with the development of the spine or brain; together they are called **neural tube defects (NTDs)**.

- NTDs occur very early in pregnancy, by the 28th day after conception. This is often before a woman knows she is pregnant
- NTDs cause many babies to die and others to have problems with walking and with bowel and bladder control. They affect about 1 in every 500 pregnancies and about 1 in every 600 births; the difference in these rates is due to the loss of pregnancies affected with NTDs
- Any population group can be affected

What causes neural tube defects?

While research is ongoing, it is evident that both heredity and certain factors in the prenatal environment act together to cause NTDs.

Anencephaly

Anencephaly occurs when the neural tube fails to close at the head. The brain and the skull bones do not develop normally.

- Infants born with this problem die at, or soon after birth.

Spina bifida

The term *spina bifida* comes from the Latin words 'spina' meaning spine and 'bifida' meaning split or divided.

The back-bone (*spine*) is made up of separate bones called *vertebrae*, which normally cover and protect the spinal cord.

- When the baby is developing, if the developing neural tube fails to close (usually at the base of the spine), the *vertebrae* will not completely fuse
- As a result, the spinal cord and the protective sac that surrounds the cord (*meninges*) may protrude through the open part of the spine, ie. *spina bifida*.

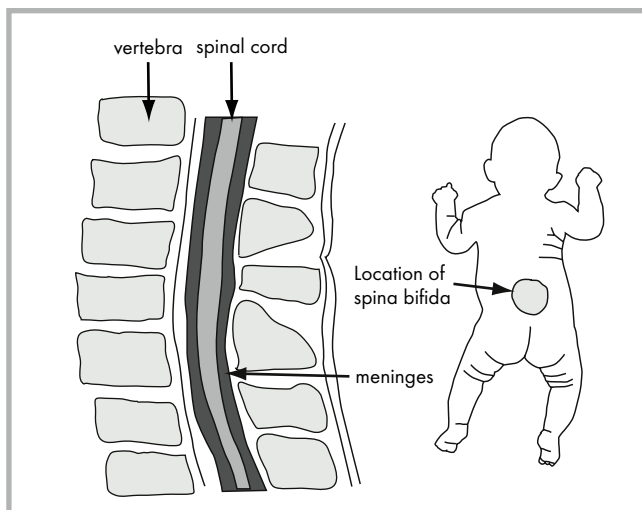


Figure 59.1: Spina bifida occulta where the outer parts of the vertebrae are not completely joined. The spinal cord and covering (meninges) are undamaged.

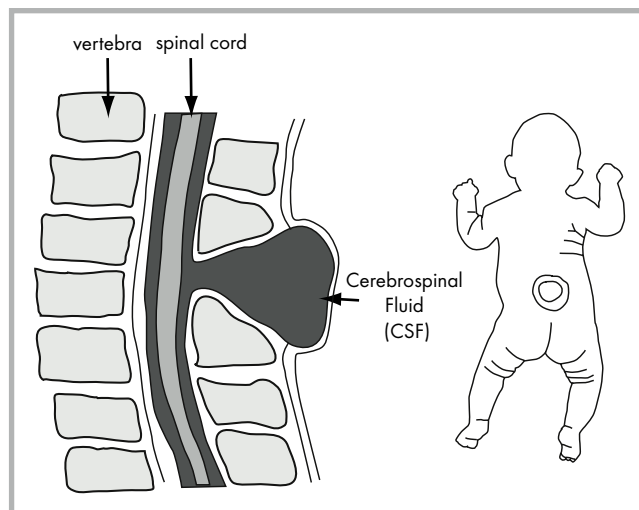


Figure 59.2: Meningocele where the outer part of the vertebrae are split and the spinal cord is normal. Only the covering of the spinal cord (meninges) is damaged and pushed out through the opening.

Symptoms associated with spina bifida vary depending on the position of the opening along the spine and on how much of the spinal cord, or the protective sac (meninges), protrudes through the vertebrae.

- If only the sac protrudes, the condition is less severe than if the cord itself and the associated nerves protrude and are damaged
- The condition tends to have a more severe effect when the opening is higher up the spine

Spina bifida has been grouped into different categories according to the location and severity of the abnormality:

- **Occulta** is where the outer parts of the vertebrae is not completely joined. The spinal cord and covering (meninges) are undamaged. There are often tufts of hair found at the site of the abnormality (Figure 59.1).
- **Meningocele** is where the outer parts of the vertebrae are split and the spinal cord is normal. Only the covering of the spinal cord (meninges) is damaged and pushed out through the opening (Figure 59.2).
- **Myelomeningocele** where the outer parts of the vertebrae is split and the spinal cord and the meninges are damaged and protruding from the opening (Figure 59.3).

Can spina bifida be treated?

The spinal abnormality may be slight, causing only minor effects. In this situation, treatment may not be required.

In most cases of spina bifida, the opening and protrusion of the spinal cord can be surgically corrected but some degree of disability may remain, affecting the lower limbs as well as bladder and bowel function.

Some individuals with spina bifida may develop *hydrocephalus* or 'water on the brain'. This happens when spinal fluid collects in and around the brain, causing the head to become enlarged. The fluid can be drained through a special tube (called a shunt) that is surgically placed and runs under the skin, down into the chest or abdomen. Such treatment helps to reduce the build-up of pressure

inside the skull caused by fluid, and to minimise the chance of intellectual impairment occurring.

Can neural tube defects be prevented by taking the vitamin folate?

Every woman has a chance of having a child with an NTD. Most of the time this chance is small.

In most women, it is possible to reduce the chance of NTDs occurring in a baby, by taking the vitamin, folic acid (folate), and other vitamins, in a specified dose.

It is recommended that they take these vitamins at least one month prior to conception and continue to do so throughout the first few months of pregnancy (see Genetics Fact Sheet 19).

If there is a family history of an NTD, or the woman has epileptic seizures, she may need more folate than most women.

Advice on this preventive treatment should be discussed with a woman's medical practitioner or a genetic counsellor.

What is the pattern of inheritance of neural tube defects in families?

For the majority of cases of spina bifida or anencephaly, both genetic factors and environmental factors are involved in the cause of the condition. The pattern of inheritance is not clear-cut and is described as multifactorial (see Genetics Fact Sheet 11).

In the majority of cases, the non-genetic factor contributing to the NTD is the lack of folate in the baby's environment.

Genetic factors are involved however as some women are at increased risk for having a baby with an NTD.

If a woman has had one baby with an NTD, the chance of having another affected baby is about 1 in 20.

- About half of this risk will be for anencephaly
- A woman is at equal risk for either spina bifida or anencephaly in future pregnancies, regardless of whichever of these neural tube defects occurred in a previous pregnancy
- A woman is also at risk for having a baby with an NTD if there is a close relative who has had a baby with either spina bifida or anencephaly, or is taking certain drugs to control epilepsy

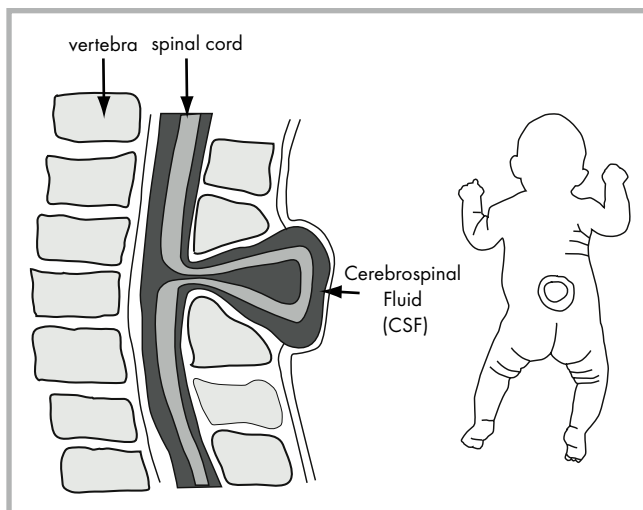


Figure 59.3: Myelomeningocele where the outer part of the vertebrae is split with the spinal cord and the meninges damaged and protruding from the opening.

What can be done if a woman thinks she is at increased risk for having a baby with a neural tube defect?

If a woman thinks that she may have an increased risk of having a child with a neural tube defect, she needs to discuss her family history or medications with her doctor. If she is at increased risk, she will need to take folic acid tablets to increase the level of folate over and above that which she could get from her diet.

Genetic counselling (see Genetics Fact Sheet 3) can provide current information about the conditions, the preventive plan, assessment of their particular risk and discussion of the tests that are available to detect affected babies before birth (see Genetics Fact Sheets 17A & 17B).

Other Genetics Fact Sheets referred to in this Fact Sheet: 3, 11, 17A, 17B, 19

Information in this Fact Sheet is sourced from:

- Barlow-Stewart K, Emery J and Metcalfe S. (2007). Testing and pregnancy. In: *Genetics in family medicine; the Australian handbook for general practitioners*. Canberra: Biotechnology Australia, Commonwealth Department of Industry, Tourism and Resources
- Eichholzer M, Tonz O, Zimmermann R. (2006). Folic acid: a public-health challenge. *Lancet*. 22;367(9519):1352-61
- Harper P. (2004). *Practical Genetic Counselling*. London; Arnold
- National Organisation for Rare Disorder (NORD) [online]. Available from: <http://www.rarediseases.org/>. [Accessed June 2007]
- Padmanabhan R. (2006). Etiology, pathogenesis and prevention of neural tube defects. *Congenit Anom (Kyoto)*, 46(2):55-67
- Spina Bifida Group of NSW [online]. Available from: <http://www.northcott.com.au>. [Accessed June 2007]
- Spina Bifida & Hydrocephalus Association of South Australia [online]. Available from: <http://www.spinabifida.asn.au/facts.html>. [Accessed June 2007]

Edit history

June 2007 (8th Ed)

Author/s: A/Prof Kristine Barlow-Stewart

Acknowledgements this edition: Gayathri Parasivam

Previous editions: 2004, 2002, 2000, 1998, 1996, 1994, 1993

Acknowledgements previous editions: Mona Saleh; Bronwyn Butler; Dr Deborah Kennedy; Dr Barbara Field