

This fact sheet describes neural tube defects and includes a discussion of the symptoms, causes and available testing.

In summary

- Neural tube defects (NTDs) occur early in a baby's development. The neural tube normally develops into the brain and spinal cord. In NTDs, the neural tube fails to 'zip up'.
- A NTD can impact the way the brain develops, causing problems with walking, bowel and bladder control and is often so severe the baby does not survive.
- NTDs include spina bifida and anencephaly. Approximately 1 in 1000 pregnancies is affected by a NTD.
- NTDs are caused by a combination of genetic and environmental factors acting together.

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. Approximately 1 in 1000 pregnancies are affected by a NTD.
- The chance of having a NTD is higher in indigenous mothers (approximately 1 in 600) and in multiple pregnancies, such as twin pregnancies (approximately 1 in 440).
- Any population group can be affected.

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*.

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.

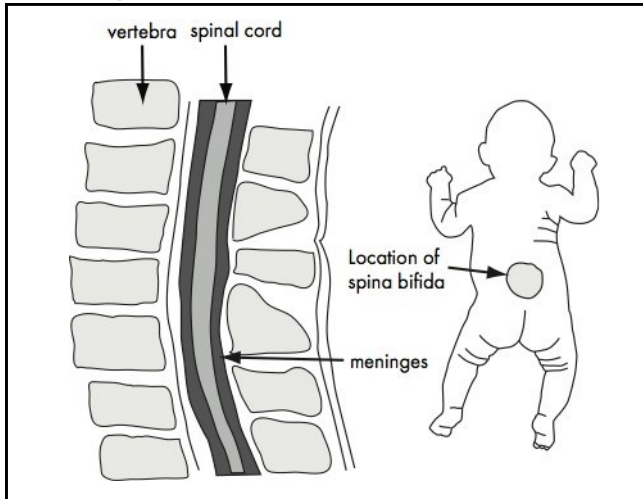


Figure 60.1: Spina bifida occulta where the outer parts of the vertebrae are not completely joined. The spinal cord and covering (meninges) are undamaged.

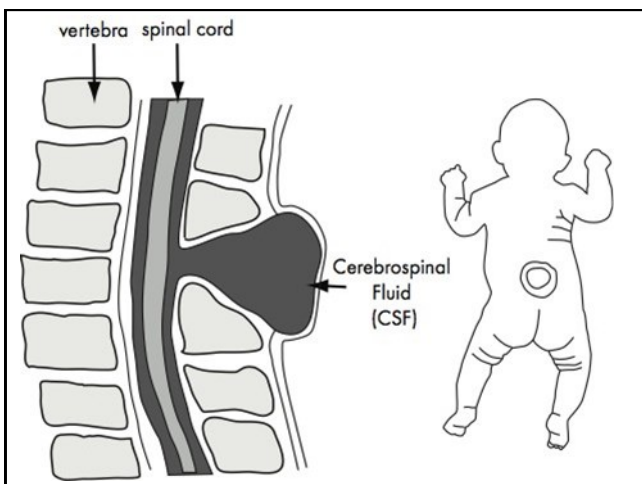


Figure 60.2: Meningocele where the outer part of the vertebrae are split and the spinal cord is normal. Only the covering of the spinal cord (meninges) is damaged and pushed out through the opening.

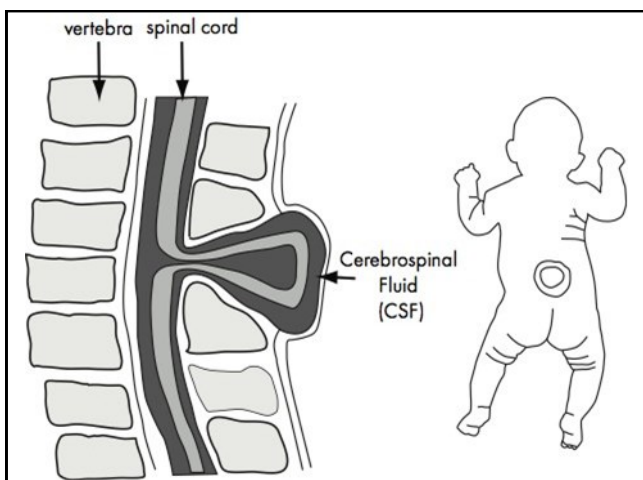


Figure 60.3: Myelomeningocele where the outer part of the vertebrae is split with the spinal cord and the meninges damaged and protruding from the opening.

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

- **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 NTD (*Figure 60.1*).
- **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 60.2*).
- **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 60.3*).

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. This treatment helps to reduce the build-up of pressure inside the skull caused by fluid, and to minimise the chance of intellectual impairment occurring.

WHAT CAUSES NEURAL TUBE DEFECTS?

While research is ongoing, it is evident that both genetic and environmental factors act together to cause NTDs. We don't know all the factors involved yet, and the way these factors interact is still not well understood.

Women have an increased likelihood of having a baby with a neural tube defect if:

- They have had a baby with spina bifida, anencephaly or another neural tube defect
- They or their partner were born with a neural tube defect
- They or their partner have a close relative who was born with a neural tube defect
- They take certain anti-epileptic and other medications (which may affect absorption or metabolism of folate)
- They have insulin-dependent diabetes
- They are obese (BMI>30).

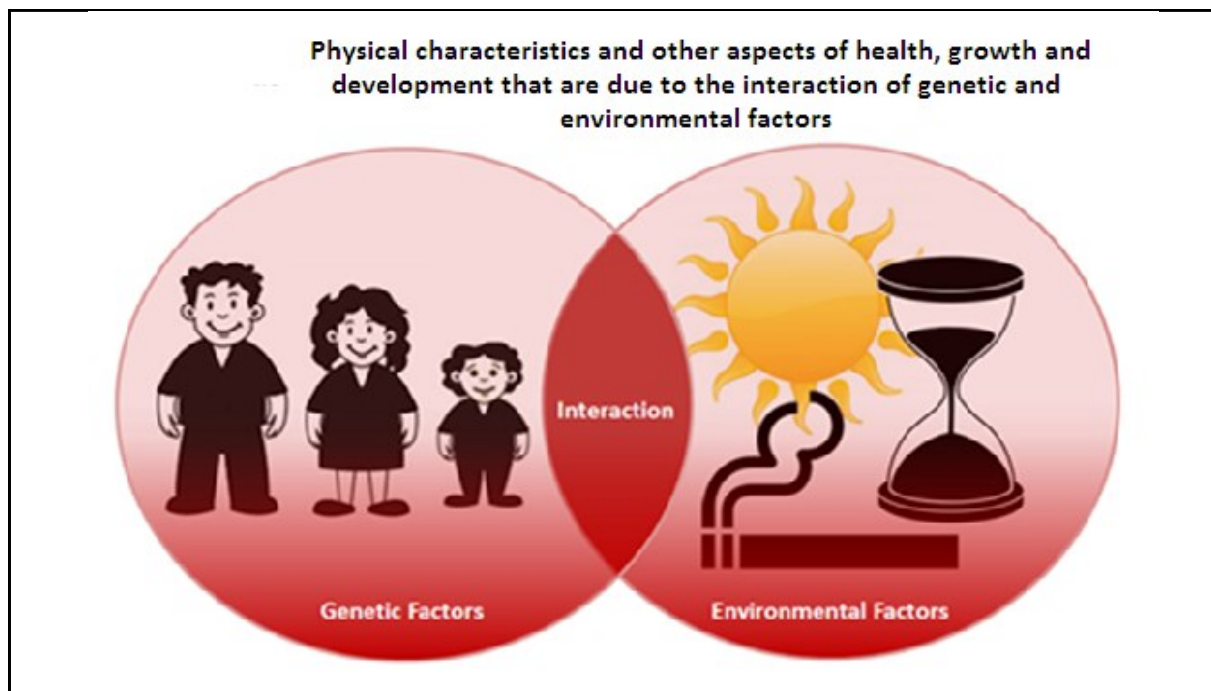


Figure 60.4. A diagrammatic representation of the interaction between genetic and environmental factors

HOW ARE NEURAL TUBE DEFECTS INHERITED?

For the majority of NTDs, both genetic factors and environmental factors are involved in the cause of the condition. The combination of genetics and external factors is called multifactorial inheritance.

We can see that genetic factors are involved as some women who have a family history of NTDs are more likely to have a baby with a NTD.

- If a woman has had a previous baby with a NTD, the chance of having another affected baby is about 1 in 30. About half of this will be for anencephaly.
- A woman has an equal chance for either spina bifida or anencephaly in future pregnancies, regardless of whichever of these neural tube defects occurred in a previous pregnancy.
- If a woman has a sibling or a parent that had a baby with a NTD, the chance of having an affected baby herself is about 1 in 220.

Multifactorial inheritance refers to the pattern of inheritance of certain conditions due to a combination of both genetic and other factors that may include internal factors such as ageing, and exposure to external environmental factors such as diet, lifestyle, and exposure to chemicals or other toxins (*Figure 60. 4*).

Multifactorial conditions do not always develop despite the presence of a genetic mutation which increases the person's risk. For example, not all women who have a mutation in a folate metabolism gene will have a baby with a NTD. The mutation alone is not completely **penetrant**.

The reason for this **incomplete penetrance** of the condition is most likely due to the interaction between the information in the gene mutation with the information in one or more other genes and with other 'environmental' factors.

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

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

Studies have shown that 4 to 7 out of 10 (40% to 70%) cases of neural tube defects can be prevented by increasing the mother's intake of folate before and during early pregnancy.

Many pregnancies are unplanned, so all women should make sure that they have a folate-rich diet, or take a daily folate tablet.

A folate-rich diet with a wide variety of vegetables, fruits, legumes, whole grains and cereals is a healthy way of eating for everyone. Eating this group of foods can help prevent heart disease, some cancers, diabetes and other health problems. It can be difficult to obtain enough folate from your diet so many women choose to take a daily tablet.

In most women, it is possible to reduce the chance of NTDs occurring in a baby by taking folate in a specified dose.

HOW MUCH FOLATE IS NEEDED?

- Most women need 0.4mg (milligrams) of folate every day. Sometimes tablets are labelled as 400 micrograms, 400 mcg or 400 ug.
- Achieving this amount of folate can be through the food you eat. You may want to talk to your doctor or nutritionist to make sure you are getting enough folate through your food.
- You can take a daily folate tablet that you can buy at your chemist, health food store or supermarket.
- Women who take a 0.4mg folate tablet each day and eat a folate rich diet are not likely to exceed a safe dose of folate. Too much folate can mask vitamin B12 deficiency but this is rare and can be checked by your doctor.

Should all women have 0.4 mg folate each day?

In general, yes. However some women are at higher chance than others of having a baby with a neural tube defect.

These women should have more folate in their system and therefore may need to take 5mg of folate every day.

Women in this group should talk to their doctor or genetic counsellor before pregnancy for advice about the amount of folate they should take.

A note of caution about taking folate

Women who take medicines to control epilepsy, seizures or psychiatric disorders should take folate only as advised by their doctors. Folate can interfere with the anti-seizure activity of some medications.

Folate is included in most multi-vitamin and mineral tablets but often at a lower dose than 0.4mg. Multi-vitamin and mineral tablets should not be taken in high doses to get the right amount of folate because high doses of some vitamins, such as vitamin A, can cause other problems with a developing baby.

IS THERE ANY TESTING AVAILABLE FOR NEURAL TUBE DEFECTS?

Ultrasound, performed around 8-12 weeks of pregnancy, or at 18-20 weeks can often detect anencephaly and spina bifida. You may wish to discuss testing options with your doctor, midwife or genetic counsellor.

References

Abeywardana S & Sullivan EA 2008. Neural tube defects in Australia. An epidemiological report. Cat. no. PER 45. Sydney: AIHW National Perinatal Statistics Unit.

AIHW National Perinatal Statistics Unit 2011. Neural tube defects in Australia: prevalence before mandatory folic acid fortification. Cat. no. PER 53. Canberra: AIHW. Viewed 16 December 2015. <<http://www.aihw.gov.au/publication-detail/?id=10737420864>>.