

What are the risk figures?

Accurate diagnosis is essential, and these risks should be discussed with your doctor or genetic counsellor.

Average Couple (Australian population)
The risk is 1 in 750

If you have one child or pregnancy
with a neural tube defect
The risk is 1 in 50

If you have two children or pregnancies
with a neural tube defect
The risk is 1 in 10

If you or your partner have a neural tube defect
The risk is 1 in 25

If you or your partner have a brother
or a sister with a neural tube defect
The risk is 1 in 100

Who to Contact

You can discuss any of these issues with your doctor. Your local doctor can also refer you to a genetic counselling service.

We would encourage you to discuss these tests with a genetic counsellor, in the context of your risks and personal beliefs. Victorian Clinical Genetics Services offers an information and counselling service.

For further information contact:

Victorian Clinical Genetics Services

4th Floor, Royal Children's Hospital,
Flemington Road, Parkville, Victoria 3052
phone: (03) 8341 6201
website: vcgs.org.au

Maternal Serum Screening, Victorian Clinical Genetics Services

phone: 1300 934 355
website: vcgs.org.au

Other useful telephone numbers:

1. Spina Bifida Clinics:

Department of Developmental Medicine (for children)
Royal Children's Hospital, Flemington Road,
Parkville, Victoria 3052
phone: (03) 9345 5898

Monash Neurology (for adults)
Monash Medical Centre
246 Clayton Road, Clayton, Victoria 3168
phone: (03) 9594 2240

2. Spina Bifida Foundation Victoria:

phone: (03) 9663 0075
website: www.sbfv.org.au

3. Yooralla:

phone: (03) 9666 4500
website: www.yooralla.com.au

Neural Tube Defects

This Pamphlet Provides Information About Neural Tube Defects Such As:

Spina Bifida
Anencephaly
Encephalocele



What are neural tube defects?

The 'neural tube' is a tube-shaped structure, which forms during early development of the embryo, to enclose the spinal cord and brain. Neural tube defects result from a failure of closure of the developing spine or skull. This may lead to damage and poor formation of a baby's spinal cord or brain. The cause of neural tube defects is uncertain. At present we believe it is due to a combination of genetic and environmental factors.

A baby born with a neural tube defect usually has one of three problems:

- 1. Anencephaly**, in which the brain and skull do not form properly.
- 2. Spina bifida**, in which the skin and bone have not closed over the developing spinal cord so that part of the spinal column, or backbone, is open. The severity varies from a small defect covered by skin to a large opening. Part of the spinal cord or its coverings may protrude through the opening and lie in a sac on the baby's back and are technically called a myelocele or meningocele.

Spina bifida occulta, is usually a minor deficiency in the spine, which occurs in about 10% of the population. It generally does not cause problems. Persons with this type of spina bifida occulta are at no greater risk of having a child with a neural tube defect than the average couple.
- 3. Encephalocele**, where there is a defect in the skull and part of the brain and/ or its lining protrude.

What kind of problems do neural tube defects cause?

Babies born with anencephaly are always stillborn or die shortly after birth.

The problems caused by spina bifida vary depending on the size of the opening, its location on the back and the amount of damage to the spinal cord and brain. If the defect is small, low down and covered by skin, there may be no problems or mild difficulties with leg weakness or reduced sensation. More often the defect is large, higher up the spine and covered only by a thin membrane. This leads to more severe problems. These may include lack of bowel and bladder control, lack of feeling in the legs (paraplegia), mis-shaped feet, dislocated hips and spinal curvature.

Hydrocephalus, enlargement of the ventricles, the hollow, fluid filled spaces within the brain, often occurs because the improperly formed spinal cord does not allow normal drainage of spinal fluid. In some cases this can be treated by draining excess fluid through a tube (shunt).

Severe spina bifida may not be compatible with life.

The effects of an encephalocele depend on its size. If large, it is often not compatible with life and the unborn baby dies in the uterus or soon after birth. Smaller encephaloceles may be associated with intellectual and physical disability.

How often do neural tube defects happen?

In Australia approximately 1 in 750 pregnancies has a neural tube defect. Spina bifida and anencephaly are the most common neural tube defects.

There is an increased risk in families where a neural tube defect has occurred. The reason for the increased risk is not fully understood but genetic factors are important.

Can I reduce this risk?

YES

Recently it has been shown that women at increased risk (refer to blue box) of having a baby with a neural tube defect can reduce the risk by taking a folic acid (or folate) tablet for at least one month prior to conception and continuing for the first three months of pregnancy. One 5 mg tablet per day is the recommended dose and is a safe amount to take. These supplements are available over the counter from a pharmacy.

This has been shown to reduce the likelihood of having a baby with a neural tube defect.

All other women, whose risk is low, only need to take a smaller dose of folic acid for at least two months prior to conception, continuing until three months pregnant. One 0.5mg tablet per day is the recommended dose.

Are there tests which can find neural tube defects during pregnancy?

The Maternal Serum Screening Test in the second trimester is a blood test for pregnant women to find if they may be at an increased risk of having a baby with neural tube defects and other conditions. This test is performed between 15 and 20 weeks of pregnancy. By itself, this test cannot diagnose neural tube defects, rather, it identifies women who should be offered further testing. A doctor who specialises in fetal ultrasound can diagnose neural tube defects.

Ultrasound scanning is a test using sound waves, which are bounced off the baby to give a shadow picture. The best time is between 18 and 20 weeks of pregnancy, when a doctor, specialising in fetal ultrasound, can diagnose most cases of spina bifida and anencephaly. Women at increased risk of having a child with neural tube defects may have a vaginal ultrasound at 11 weeks as anencephaly can be diagnosed by this time. Ultrasound carries no risk to the pregnancy.

If good ultrasound views of the baby are not obtained then the patient should be referred to a specialist obstetric ultrasound practice.