

## Birth defects - central nervous system

Birth defects of the central nervous system are called neural tube defects (NTDs). NTDs include conditions called spina bifida, anencephaly and encephalocele. They are all present at birth and are due to a problem with the development of the brain and/or spinal cord in the developing baby (fetus).

The brain and spinal cord of a growing fetus develop from a simple structure called the *neural tube*. The neural tube 'zips up' along its length to close and protect the brain and spinal cord. If the neural tube doesn't close at any part along its length, the baby will have a neural tube defect. The types of neural tube defects (spina bifida, anencephaly and encephalocele) are due to the place along the neural tube that hasn't closed, leaving parts of the brain and/or spinal cord exposed.

A range of genetic and environmental factors are thought to be responsible for NTDs, including the mother having not enough of the vitamin folate and some epilepsy medications. Taking folate (folic acid) before and during early pregnancy can significantly reduce the chance that a mother will have a baby with this kind of birth defect.

### The central nervous system (CNS)

The central nervous system consists of the brain and spinal cord. Both are wrapped in a thin lining called *meninges* and are surrounded by a fluid called *cerebrospinal fluid*. The brain is attached to the spinal cord by the brain stem, located at the base of the brain. The spinal cord runs the length of the backbone and is protected by the bones (vertebrae) of the spine. Nerves branch off from the spinal cord into the parts of the body.

### Development of the fetal CNS

The central nervous system of a growing fetus starts with a simple structure called the 'neural groove' that folds in to form the 'neural tube'. This then develops into the spinal cord and brain. By day 28 after conception, the neural tube should be closed and fused. If it doesn't close, the result is a neural tube defect.

In many cases, these defects can be diagnosed during pregnancy with ultrasound scans and, rarely, with other tests such as amniocentesis (analysing a sample of amniotic fluid).

### Risk factors and prevention of CNS birth defects

Neural tube defects are thought to be caused by a range of genetic and environmental factors working in combination. Some of these factors include:

- **The mother has a folate deficiency** – if the mother is lacking some nutrients, especially the B-group vitamin called folate (folic acid), the chance of having a baby with a NTD is increased. If folate is taken **before conception** and at least for the first four weeks of pregnancy, around seven out of 10 cases of NTDs can be prevented. You should talk to your doctor about how much folate you should take if you are thinking of becoming pregnant.
- **Genetics** – the exact genetic association is unclear, but a woman is at increased risk of having a baby with a neural tube defect if she has a close relative who has had a baby with the condition (a family history). A woman who has already given birth to a child with a neural tube defect is also at increased risk of having subsequent babies with a similar condition.
- Having a personal or family history of a NTD can influence the amount of folate needed to reduce the chance of having a baby with a neural tube defect. You should talk to your doctor about how much folate you should take if you are thinking of becoming pregnant.

- In some cases, however, there is evidence to suggest that some forms of neural tube defects are caused by specific genetic changes (mutations) that are not related to folate. In these cases, the neural tube defect is caused by the baby inheriting faulty gene copies from both parents. These faulty genes prevent the baby from making use of folate that is necessary to grow and develop in pregnancy (even if the folate is present in the right amount). In these cases, taking folate before and during pregnancy will not prevent the condition.
- **Medication** – particular medications used to treat and control epilepsy are thought to contribute to the risk of neural tube defect.

## Spina bifida

In a baby with spina bifida, the bones (*vertebrae*) of the spine fail to fuse. The spinal cord and nerves protrude (or 'pop out') through the gap that has been created due to a failure of closure of the neural tube. This can affect the nerves that spread from this area into the abdomen and legs. Spina bifida can occur anywhere along the length of the spine, but more commonly appears in the lower back.

Nine out of 10 affected babies also have a build-up of cerebrospinal fluid inside the brain. This condition is called hydrocephalus and is sometimes referred to as 'water on the brain'. The incidence of spina bifida in Victoria is around one in every 1,235 births.

Spina bifida can be mild, moderate or severe and is graded according to the degree of the defect into:

- **Occulta** – the bones (*vertebrae*) have not closed completely, but the spinal cord is unharmed. The characteristic soft lump may be missing, which is why this form of spina bifida is sometimes diagnosed later in life.
- **Meningocele** – the membrane (meninges) covering the spinal cord protrudes or bulges out through the gap in the spine.
- **Myelomeningocele** – the meninges, spinal cord and blood vessels protrude through the gap.

Spina bifida is incurable. The main form of treatment is surgery to seal the gap. If the baby has hydrocephalus, a shunt is inserted into the brain to drain the excess cerebrospinal fluid.

## Anencephaly

When the neural tube doesn't close at the head, this may cause the uppermost brain tissue, the meninges, top of the skull (*calvarium*) and the scalp to be partially or completely missing. For reasons unknown, anencephaly occurs twice as often in females as males. Around one in 10 affected babies is one of a pair of twins. In some cases, the baby has other problems such as congenital heart disease and cleft palate. The incidence of anencephaly in Victoria is around one in every 1,370 births.

A baby with anencephaly is not able to live. Most are stillborn or die within a few days of birth.

## Encephalocele

In this rare form of neural tube defect, the meninges and brain tissue bulge out through a gap in the skull. In severe cases, the brain is pushed out to such an extent that the bundle of brain tissue covered by the meninges (membranes) may be larger than the baby's head. Infection can occur if the membrane breaks and exposes the brain tissue. Other problems associated with encephalocele can include cleft lip or palate, additional fingers (*polydacty*) and abnormalities of the sex organs. The incidence of encephalocele in Victoria is around one in every 6,667 births.

If there is a significant amount of brain tissue that has been pushed out, surgery may not be possible. Treatment options include the use of a shunt to remove the fluid build-up (treat the hydrocephalus), if necessary. The baby will experience a range of difficulties that can include intellectual impairment, difficulty in controlling muscles (*spasticity*) and fits (*convulsions*). Physiotherapy and anticonvulsant medication may help.

## Where to get help

- Your doctor

- Paediatrician
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6200
- Genetic counselling services – available at most large public maternity hospitals
- Spina Bifida Foundation of Victoria Tel. (03) 9663 0075

## Things to remember

- The brain and spinal cord of a growing fetus develop from a simple structure called the neural tube.
- If the neural tube doesn't fuse together, the baby will have a neural tube defect.
- Types of neural tube defects include spina bifida, anencephaly and encephalocele.
- Taking the right amount of folate before and during early pregnancy can prevent seven out of 10 cases of neural tube defects.

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