

Neural tube defects: open spina bifida (also called spina bifida cystica)

Information for health professionals

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The aim of this information sheet is to support staff involved in counselling pregnant women and their partners when a suspected or confirmed diagnosis of open spina bifida has been made, following an ultrasound scan.

All diagnoses of the conditions must be recorded and audited to ensure the effectiveness of the screening programme.

1. Definition

The fetal nervous system starts as a single structure called the neural plate. By the 28th day after conception the neural plate should fold over and close to form the neural 'tube'. Failure of the complete closure of the neural tube results in an NTD, of which there are several types: anencephaly, spina bifida, and encephalocele.

There are two main types of spina bifida: spina bifida occulta and spina bifida cystica. There are two forms of spina bifida cystica, meningocele, and myelomeningocele (these are also called open spina bifida). This information sheet explains myelomeningocele which accounts for approximately 75% of all cases.

Myelomeningocele is the most serious form of spina bifida, it accounts for 75% of cases. Here the bones of the spine (vertebrae) do not form properly. This results in a small sac 'herniating' through the opening in the spine. A myelomeningocele can occur anywhere along the spinal cord. It is most common in the lower back (lumbar and sacral areas). The meningeal sac is covered with a membrane and contains cerebrospinal fluid (CSF). The sac may also contain portions of the spinal cord and nerves which may become damaged. Therefore, the higher the myelomeningocele is on the baby's back, the more loss of function occurs.

Meningocele is a rare type of spina bifida in which only the membranes protecting the spinal cord herniate through the vertebrae.

Between 70–80% of children with open spina bifida will also develop hydrocephalus – which is a collection of too much cerebrospinal fluid (CSF) in the brain (Rintoul et al. 2002).

The condition appears to be caused by a combination of genetic factors and environmental influences. The specific genes and environmental factors are not completely understood.

2. Prevalence

Open spina bifida occurs in approximately 6 per 10,000 births (Boyd et al. 2011).

3. Screening and diagnosis

Most cases of open spina bifida are detected at the 18⁺⁰–20⁺⁶ weeks Fetal Anomaly ultrasound scan. A second scan may be needed to confirm the diagnosis.

4. Treatment

The management of open spina bifida can only be finalised after birth when the type and size of the lesion is assessed. The treatment will depend upon the position and the severity of the lesion. Most cases of open spina bifida require an operation to repair the neural tube after birth. It is likely that some damage to the baby's nervous system will have already taken place and surgery will not be able to correct all problems.

If the child develops hydrocephalus, this needs to be treated with surgery as increased pressure induced by the excessive fluid can cause brain damage. Surgery involves draining the fluid into the baby's abdomen (a ventriculo-peritoneal shunt). This often needs to be replaced as the child grows because of problems with blockage and infection.

5. Prognosis

The prognosis for babies born with spina bifida depends on the location, size and extent of the defect and the presence of hydrocephalus.

The range of disability caused by open spina bifida is variable and ranges from apparent normality to very severe disability. This is dependent on the extent of the original neurological deficit and the outcome of the CSF shunt. Children with spina bifida frequently have problems controlling their bladder and bowels. In more serious cases the baby may have problems walking or be unable to walk. Hydrocephalus can cause learning difficulties.

Generally, abnormalities higher on the spine produce a greater risk of paralysis and other debilitating complications.

6. Recurrence

If a woman has had a baby with spina bifida, there is a 3–4% chance of having another baby with this condition (Cowchock et al. 1980).

Many cases of spina bifida are isolated. However, prenatal investigations are sometimes offered to exclude chromosomal abnormalities as up to 10% of NTDs are associated with chromosomal abnormalities (Sepulveda et al. 2009).

When NTDs occur as part of a genetic syndrome the recurrence risks are much higher (up to 25%).

7. Prevention

All women are advised to take a supplement of 400 micrograms of folic acid for at least three months before pregnancy and up to the end of the 12th week of pregnancy (NICE CG62 2008).

Women who have a family history of NTDs are advised to take a higher dose of 5mg of folic acid, prescribed by their GP, as this has been shown to reduce their chances of having another baby with an NTD (NICE Public Health Guidance 11 2008).

Couples who have a family history of NTDs should be given the opportunity to see a genetic counsellor to discuss risks to future pregnancies.

8. Referral pathway

Following diagnosis of open spina bifida, referral should be made to a specialist in fetal medicine for a second opinion and further information.

This will involve careful assessment of the fetus to identify any additional abnormalities and also assess the level of the spina bifida. Where appropriate, the offer of karyotyping (by chorionic villus sampling (CVS) or amniocentesis)¹ to exclude a chromosomal abnormality should be discussed. Depending on the ultrasound findings, additional information from a paediatric team with expertise in the care and management of children with spina bifida should be offered.

A termination of pregnancy should be offered following appropriate counselling. Women should be offered the opportunity to discuss the possible implications of continuing or ending their pregnancy.

Some women choose to continue the pregnancy and these parents will need ongoing care and support.

Women who elect to continue their pregnancy should be referred antenatally to the Paediatric team who will care for their baby. Ongoing antenatal care involves ultrasound scans to monitor the fetus.

The NHS FASP has produced a care pathway for neural tube defects and for prenatal investigation. They are both available here: www.fetalanomaly.screening.nhs.uk/timelines.

9. Further information, charities and support organisations

Antenatal Results and Choices (ARC)

Email: info@arc-uk.org

Helpline: 0845 017 2290

Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides information and support to parents before, during and after antenatal screening and diagnostic tests, especially those parents making difficult decisions about testing, or about continuing or ending a pregnancy after a diagnosis. ARC offers ongoing support whatever decisions are made.

¹More information on CVS and amniocentesis can be found in the following leaflets: *Chorionic villus sampling (CVS) – information for parents*, *Amniocentesis test – information for parents*, *Chorionic Villus Sampling (CVS) and Amniocentesis – for health professionals*. These are available here: www.fetalanomaly.screening.nhs.uk/publicationsandleaflets.

SHINE, previously the Association for Spina Bifida and Hydrocephalus (ASBAH)

Information line: 0173 355 5988

Email: info@shinecharity.org.uk

Website: www.shinecharity.org.uk

SHINE is the leading UK registered charity providing information and advice about spina bifida and hydrocephalus to individuals, families and carers. SHINE offers services to those affected by, or with an interest in, spina bifida or other neural tube defects – from before birth (with advice and support, whether or not the pregnancy continues).

References

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This information has been produced on behalf of the NHS Fetal Anomaly Screening Programme for the NHS in England. There may be differences in clinical practice in other UK countries. The leaflets have been developed through consultation with the NHS Fetal Anomaly Screening Programme expert groups.

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