

Neural tube defects (NTDs): anencephaly

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 anencephaly 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 a neural tube defect (NTD) of which there are several types: anencephaly, spina bifida and encephalocele.

Anencephaly is a congenital malformation characterised by the total or partial absence of the cranial vault and covering skin. As a result the brain is missing or reduced to a small mass and there are significant abnormalities of the face and neck.

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

Anencephaly occurs in approximately 6 per 10,000 births in England (Boyd et al. 2011).

3. Screening and diagnosis

Anencephaly can be detected prenatally using ultrasound, frequently as early as 11 to 14 weeks gestation. The diagnosis cannot usually be made before 11 weeks because skull ossification is not complete. This abnormality is easily visible at the detailed Fetal Anomaly ultrasound scan between 18⁺⁰–20⁺⁶ weeks.

A repeat scan may be offered to confirm the diagnosis.

4. Treatment

There is no cure or treatment for anencephaly.

5. Prognosis

The condition is not compatible with life and the majority of babies with anencephaly are either stillborn or die within a few hours of delivery. Occasionally, babies can live up to a few days but this is rare.

6. Recurrence

The recurrence rate for women who have had an affected pregnancy is 1 in 50 (2%) (Czeizel A. and Munkacsy J., 1984).

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 their future children.

8. Referral pathway

Following diagnosis of anencephaly by ultrasound scan, a second medical opinion should be sought from a specialist in fetal medicine.

After confirmation of the diagnosis, the woman should be informed that this is a lethal abnormality.

A termination of pregnancy should be offered to allow women the option of not continuing with their pregnancy following appropriate counselling.

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

Work with the bereavement service may help the family prepare for a stillbirth or early neonatal death.

Women may be referred to their local bereavement service. Palliative care should be discussed and women may be referred to the children's palliative care team.

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

9. Further information, charities and support organisations

Anencephaly.co.uk

Email: richard@anencephaly.co.uk

Email: joanne@anencephaly.co.uk

Website: www.anencephaly.co.uk

Anencephaly.co.uk's aim is to provide a place for people in the UK to talk to other families within the UK who have had experiences with anencephaly.

Antenatal Results and Choices (ARC)

Email: info@arc-uk.org

Helpline: 0845 077 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.

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

Boyd PA, Tonks AM, Rankin J, Rounding C, Wellesley D, Draper ES. Monitoring the prenatal detection of structural fetal congenital anomalies in England and Wales: register-based study. *Journal of Medical Screening* 2011; 18(1): 2.

NICE. Antenatal care: routine care for the healthy pregnant woman, Clinical Guidelines CG62. Issued March 2008.

NICE. Improving the nutrition of pregnant and breastfeeding mothers and children in low-income households. NICE public health guidance 11. Issued March 2008.

Czeizel A, Metneld J. Recurrence risk after neural tube defects in a genetic counselling clinic. *J Med Genet* 1984; 21: 413–416.

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.

All of our publications can be found online at: www.fetalanomalyscreening.nhs.uk.

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