NHS

Screening Programmes

Fetal Anomaly

Alobar holoprosencephaly (HPE) information for health professionals

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Aim

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Definition, causes and risk factors

HPE is a severe brain abnormality caused by incomplete cleavage of the embryonic forebrain/failure of the prosencephalon to cleave into the cerebral and lateral hemispheres. This process usually takes place during the third week of fetal embryonic life.

It may present as an isolated malformation of the central nervous system or as part of a syndrome. It is characterised by a spectrum of cerebral abnormalities resulting from incomplete cleavage of the forebrain. The cause of HPE is unknown. Often, no specific cause can be identified. Suggested risk factors include maternal diabetes, infections during pregnancy (syphilis, toxoplasmosis, rubella, herpes, cytomegalovirus), and various drugs taken during pregnancy such as alcohol, aspirin, lithium, thorazine, anticonvulsants, hormones and retinoic acid. Women with previous pregnancy loss and first trimester bleeding are also more likely to have a fetus or child diagnosed with HPE.

Although some neonates with this condition have normal chromosomes, specific chromosomal abnormalities have been identified in others.

Screening and diagnosis

Antenatal diagnosis of HPE is usually made during the 18^{+0} – 20^{+6} weeks' fetal anomaly ultrasound scan but can occasionally be detected ultrasonographically from as early as 12 weeks' gestation. HPE is common in chromosome anomalies, particularly trisomies 13, 15, 18, 21 and triploidy.

Neonates and children diagnosed with HPE may have microcephaly, hydrocephalus, varying degrees of learning difficulties, epilepsy, endocrine abnormalities, or abnormalities of other organ systems such as cardiac, skeletal, genitourinary and gastrointestinal.

The NHS Fetal Anomaly Screening Programme will be audited against a 80% detection rate of all cases of HPE at the 18^{+0} – 20^{+6} weeks' fetal anomaly ultrasound scan.

Prevalence

It is estimated that HPE affects 1 in 5,000-10,000 live births. Since many pregnancies with an affected fetus end in early miscarriage, the frequency among all pregnancies may be as high as 1 in 200-250.

Treatment

Given the known poor prognosis, women are offered a choice of continuing with the pregnancy with the support of the hospital team, or termination of the pregnancy.

Some women may choose to terminate the pregnancy. For those who choose to continue, treatment is based on management of the condition of the baby and support for the parents.

Prognosis

Current studies indicate that only 3% of all fetuses with HPE survive to delivery and the vast majority of these infants do not survive past the first six months of life.

Prognosis is dependent upon the degree of fusion and malformation of the brain, as well as other health complications that may be present. The more severe forms of HPE are usually fatal, though some babies may live several months or years. This disorder consists of a spectrum of defects, malformations and associated abnormalities. The level of disability is based upon the degree to which the brain is affected.

Prevention

Little is known about preventing this condition, though a genetic diagnosis would enable future pre-natal genetic counselling.

Referral pathway

Following diagnosis of holoprosencephaly alobar at the 18^{+0} – 20^{+6} weeks' fetal anomaly ultrasound scan, a second medical opinion should be sought. Prenatally diagnosed cases of HPE should be referred to a tertiary centre or FMU and a consultation with a specialist genetics counsellor may be offered.

A termination of pregnancy should be offered to allow women the option of not continuing with their pregnancy. Paediatric referral in the antenatal period will be appropriate for women who elect to continue their pregnancy.

Further information, charities and support groups

Antenatal Results and Choices (ARC)
73 Charlotte Street
London
W1T 4PN

Tel: 0207 631 0285
Email: info@arc-uk.org
Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides impartial information and individual support to parents whether they are going through antenatal screening or whose unborn baby has been diagnosed with an abnormality.

Contact a Family (CAFAMILY)
209 to 211 City Road
London
EC1V 1JN

Tel: 0808 808 3555
Email: info@cafamily.org.uk
Website: www.cafamily.org.uk

Contact a Family is a charity which provides support, advice and information for families with disabled children, no matter what their condition or disability.

DIPEx
PO Box 428
Witney
Oxon
OX28 9EU

Email: info@healthtalkonline.org
Website: www.healthtalkonline.org &
www.youthhealthtalk.org

DIPEx has created a unique database of personal and patient experiences through in-depth qualitative research into over 40 different illnesses and health conditions. The results of their research are published on two websites which are aimed at patients, their carers, family and friends, doctors, nurses and other health professionals. Their target is to complete at least 100 conditions within the next 5 – 10 years.

Further information, charities and support groups continued...

The Miscarriage Association
c/o Clayton Hospital
Northgate
Wakefield
West Yorkshire
WF1 3JS

Tel: 01924 200799
Email: info@miscarriageassociation.org.uk
Website: www.miscarriageassociation.org.uk

Miscarriage can be a very distressing, frightening and lonely experience. The Miscarriage Association provides support and information.

Stillbirth and Neonatal Death Society (SANDS)
28 Portland Place
London
W1B 1LY

Tel: 0207 436 5881
Email: helpline@uk-sands.org
Website: www.uk-sands.org

SANDS support anyone affected by the death of a baby and promotes research to reduce the loss of babies' lives.

This information has been produced on behalf of the NHS Fetal Anomaly Screening Programme. 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.fetalanomaly.screening.nhs.uk

If you have any comments on this booklet or enquiries for the Programme please contact us at the address below:

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