NHS

Screening Programmes

Fetal Anomaly

Congenital diaphragmatic hernia (CDH)

Information for health professionals

Royal College of Nursing
The voice of nursing in the UK

BMFMS

Publication date: April 2012
Review date: April 2013
Version 2

Congenital diaphragmatic hernia (CDH) Information for health professionals

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 congenital diaphragmatic hernia (CDH) 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

CDH is the protrusion of some of the abdominal or retroperitoneal structures through a defect in the diaphragm into the chest cavity. Typically this occurs at about 10–12 weeks gestation. Lung growth is impaired to a variable degree by the reduced intrathoracic space (pulmonary hypoplasia) and the heart is displaced (mediastinal shift), causing a degree of compression of the opposite (contralateral) lung. The pulmonary vasculature is also abnormal and can lead to a failure of normal cardiovascular adaptation to extrauterine life at birth. This results in pulmonary hypertension with shunting of blood away from the lungs (through the patent foramen ovale and ductus arteriosus) leading to reduced tissue oxygen delivery (hypoxia).

The majority of cases of CDH are sporadic and the cause is unknown. 80% are left sided.

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.

Most cases are isolated but up to 40% of CDH babies have other abnormalities. These most commonly affect the developing heart, kidneys and intestinal systems. Up to 20% are chromosomal abnormalities.

2. Prevalence

CDH occurs in approximately 4 in every 10,000 births (Boyd et al. 2011). Males are more commonly affected than females with a ratio of 1.3:1.

3. Screening and diagnosis

Many cases of CDH are diagnosed on the 18^{+0}–20^{+6} weeks Fetal Anomaly ultrasound scan.

Some may be diagnosed during a third trimester ultrasound scan or occasionally as early as the dating scan. However, undiagnosed cases still occur and present at birth, or very soon after, depending on the severity of the hernia.

4. Treatment

The primary aim of newborn management of CDH is to achieve adequate gas exchange, employing a ventilation strategy that protects the fragile hypoplastic lungs. This may involve conventional or oscillatory ventilation. Cardiovascular support is often required, and specific therapies to treat pulmonary hypertension may be considered. Surgery will only be undertaken when the baby is sufficiently stable, showing signs of reducing ventilation requirements and resolving pulmonary hypertension (West et al. 1992). This may take several days. Surgery to relocate the abdominal structures back into the abdomen and repair the defect in the diaphragm takes about one to two hours and requires general anaesthesia. Large defects require a prosthetic patch for repair.

After surgery, the baby will remain on a ventilator and will continue to require intensive care. The length of time needed on a ventilator after surgery depends on the degree of pulmonary hypoplasia and severity of pulmonary hypertension. It can take some time for the abdominal contents to recover after surgery and a period of parenteral (intravenous) nutrition is often required.

A baby who is failing conventional therapy should be considered for extracorporeal membrane oxygenation (ECMO) support. This option will be discussed with parents by senior medical staff in such an event and a decision will be made as to its suitability. ECMO is only available in a number of hospitals in the UK and would likely require transfer of the baby.

5. Prognosis

Survival for babies born with CDH is approximately 50%, depending on the severity of the CDH and associated abnormalities. Antenatal ultrasound scanning can look for other anomalies and is used to calculate prognostic parameters such as the lung to head ratio (LHR) of the opposite lung to estimate fetal lung size, along with the position of the fetal liver. Ultrasound scan can be supplemented by fetal MRI studies. Results of these investigations can aid in prenatal counselling.

The efficacy of fetal therapy (endoscopic tracheal plugging when antenatal prognostic tests suggest a poor outcome) is currently being evaluated in a randomised trial in Europe.

Postnatal outcome in CDH has been shown to be related to defect size (including whether the liver is in the chest), lung to head ratio (LHR) and birth weight, Apgar score at 5 minutes and the presence or absence of major associated structural or chromosomal anomalies.

6. Recurrence

For isolated CDH, the risk of recurrence for siblings is 1–2% (Norio et al. 1984). For syndromic CDH, the specific recurrence risk is dependent on the associated condition.

7. Prevention

There is no known way to prevent this condition.

8. Referral pathway

Following diagnosis of CDH, referral should be made to a specialist in fetal medicine for a second opinion and further information. This will include confirmation of the diagnosis, further prognostic evaluation (possibly including LHR) and multidisciplinary counselling. Where appropriate, karyotyping will be offered (usually by amniocentesis) to exclude a chromosomal abnormality.[1]

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. Paediatric referral in the antenatal period will be appropriate for women who elect to continue their pregnancy.

9. Further information, charities and support organisations

This information booklet has been produced by the NHS FASP and is based on the leaflet developed by CDH UK. CDH UK is a registered charity offering support to families affected by CDH. The complete leaflet can be found on the CDH UK website or by contacting them directly.

A more extensive list of support organisations is available on the website www.fetalanomaly.screening.nhs.uk.

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.

[1]More 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.

CDH UK – The Congenital Diaphragmatic Hernia Charity
Freephone: 0800 731 6991
Email support@cdhuk.co.uk
Website: www.cdhuk.co.uk

CDH UK provide information, support and advice to families, friends and health professionals who are affected by or involved in the treatment of congenital diaphragmatic hernia. They also raise awareness of the condition and encourage research and study into causes, prevention and treatments.

References

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Peetsold MG, Heij HA, Kneepkens CMF, Nagelkerke AF, Huisman J, Gemke RJ. The long-term follow-up of patients with a congenital diaphragmatic hernia: a broad spectrum of morbidity. *Pediatric SurgInt* 2009; 25: 1-17.

Norio R, Kaariainen H, Rapola J, Herva R, Kekomaki M. Familial congenital defects; aspects of etiology, prenatal diagnosis, and treatment. *Ann Med Genet* 1984; 17: 471-48

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.fetalanomaly.screening.nhs.uk.

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