NHS

Screening Programmes

Fetal Anomaly

Abdominal wall defects: exomphalos (omphalocele)

Information for health professionals

Royal College of Nursing
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BMFMS

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

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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 exomphalos 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

Exomphalos (omphalocele) is a type of abdominal wall defect.

Abdominal wall defects occur when a fetus's abdominal wall does not develop fully while in utero. This results in the intestine developing outside the abdomen.

Early in development, the intestines develop inside the umbilical cord and then move inside the abdomen by 12 weeks of pregnancy. If the abdominal contents protrude into the base of the umbilical cord and are covered by a peritoneal membrane, this is called an exomphalos. Non-rotation of the intestines is commonly seen.

The exomphalos can either be large or small. A small exomphalos may contain only tissue left over from structures in the unborn fetus's digestive tract.

The cause of exomphalos is not always known. Familial occurrence has been described.

Up to 80% of babies with exomphalos have other serious abnormalities such as heart defects and chromosomal abnormalities (Groves et al. 2006). Some of these abnormalities can be diagnosed by ultrasound scan while others can only be diagnosed as a result of invasive chromosome testing.

Congenital heart defects	1 in 5 babies with exomphalos has a cardiac defect. These can be serious. Up to 80% of these babies do not survive. Cardiac defects are especially common in babies with chromosome abnormalities.
Chromosome abnormalities	Exomphalos can be associated with congenital abnormalities. The incidence varies (Groves et al. 2006).
Syndromes	Up to 1 in 10 babies with exomphalos can have a specific syndrome (or collection of abnormalities).

Up to 40% of prenatally diagnosed apparently isolated cases are found to have associated anomalies after delivery (Cohen-Overbeek et al. 2010).

2. Prevalence

Exomphalos occurs in approximately 4 in every 10,000 births (Boyd et al. 2011).

3. Screening and diagnosis

Exomphalos is usually detected at the 18^{+0}–20^{+6} weeks Fetal Anomaly ultrasound scan.

It can be diagnosed by ultrasound earlier in pregnancy however the condition is not usually diagnosed before 11 weeks. This is due to the physiological herniation of the bowel into the umbilical cord during early fetal development.

Karyotyping by amniocentesis or chorionic villus sampling (CVS) is usually recommended due to the strong association with chromosomal abnormalities.[1]

4. Treatment

The management of exomphalos can only be finalised after birth and the medical team have assessed the baby for other associated abnormalities.

Once the baby is in a stable condition, the surgical team will decide on one of a number of approaches.

If there is only a small degree of bowel herniation, it is most likely that surgery will be performed to return the bowel to the baby's abdomen and the defect closed. This is called a primary repair.

Surgery is more challenging if the exomphalos is large (in which case it may contain liver, bowel and other organs) or if the abdominal cavity is small. In this case, a primary repair is not possible. The surgical team will then decide on the most appropriate treatment.

5. Prognosis

Prognosis and survival depends on the size of the defect, the presence of other abnormalities and the associated complications. The mortality rate for uncomplicated isolated exomphalos is 10% but rises to more than 80% in cases with other major congenital anomalies (Cohen-Overbeek et al. 2010).

6. Recurrence

This abnormality occurs sporadically. There is a low risk of recurrence in future pregnancies when the condition is not part of a syndrome. If exomphalos is associated with a syndrome, there may be a higher recurrence risk.

[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.

7. Prevention

There is no known way to prevent this condition from happening.

8. Referral pathway

Following diagnosis of exomphalos, 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. Where appropriate, the offer of karyotyping (by chorionic villus sampling (CVS) or amniocentesis) to exclude a chromosomal abnormality should be discussed. Some cases may benefit from referral to Clinical Genetics. Antenatal care should be individualised to suit the needs of the woman.

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. Ongoing antenatal care involves regular ultrasound scans to monitor the fetus. Referral to Paediatric Surgery should be made to discuss the surgical implications.

The choice of timing, mode and place of delivery will be discussed. There is no clear consensus as to the optimal method of delivery. Vaginal deliveries are considered unless there is an obstetric contraindication. The size of the exomphalos may influence the mode of delivery. Clear plans should be in place should the woman go into labour prematurely.

9. Further information, charities and support organisations

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.

GEEPS
Email: geeps@btinternet.com
Website: www.geeps.co.uk

GEEPS is an international network of families and friends of children born with abdominal wall defects. GEEPS is run by the families and friends of affected children and is a non-profit-making network. The aim of GEEPS is to support families through the shock of diagnosis and beyond in the hope that some of the stress can be relieved by sharing thoughts and fears with other parents who have been in a similar situation.

References

Groves R, Sunderajan L, Khan AR, Parikh D, Brain J, Samuel M. Congenital anomalies are commonly associated with exomphalos minor. *Journal of Pediatric Surgery* 2006; 41(2): 358-361.

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.

Cohen-Overbeek TE, Tong WH, Hatzman TR, Wilms JF, Govaerts CF, Galjaard JH, Steegers EAP, Hop WCJ, Wladimiroff JW, Tibboel D. Omphalocele: comparison of outcome following prenatal and postnatal diagnosis. *Ultrasound Obstet Gynecol* 2010; 36: 687-692.

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.

Please email any comments or feedback to: enquiries@ansnsc.co.uk.