Congenital Diaphragmatic Hernia

Recommendations of Practice

Background

- Congenital diaphragmatic hernia (CDH) is a rare disease with a prevalence ranging between 1-4 per 10,000 births
- CDH is a diaphragmatic defect which allows the abdominal contents to herniate into the thoracic cavity. It may be able to be corrected surgically after delivery. However, the result of herniation may lead to disturbed lung development during the critical embryonic period, and therefore may lead to pulmonary hypoplasia and pulmonary hypertension.
- CDH can be classified as isolated (50-60%) or complex (syndromic or non-isolated - 40-50%).
- Chromosomal anomalies are identified in 10-20% of the cases. The vast majority of the cases occur sporadically.

Objective

- To guide the accurate diagnosis and investigation of CDH
- To individualise the prognosis of the fetus and management of those presenting with a CDH.

Definition

- CDH is a developmental discontinuity of the diaphragm that allows the abdominal contents to herniate into the chest.

Differential Diagnosis

- Congenital pulmonary adenomatoid malformation (CPAM)
- Bronchopulmonary sequestration (BPS)
- Diaphragmatic eventration
- Bronchogenic cyst
- Oesophageal duplication cyst
- Teratoma.

Important History

- Family history of congenital abnormalities
- Aneuploidy screen.

Ultrasound

- 86% are left-sided, 13% are right sided and 2% are bilateral
- 70% are posterolateral, 25-30% are anterior. Rarely central.

Ultrasound appearance

- Left sided CDH:
    - Heterogeneous mass in chest, often results in right mediastinal shift
    - May have fluid-filled stomach in the chest cavity or absent stomach in the abdomen
        - Stomach can be seen behind the left cardiac atria or aside the left ventricle
    - The liver may be herniated
        - Liver in the chest can be located by following the course of the umbilical vein by using colour Doppler
    - Gallbladder and hepatic or umbilical veins may be abnormally located within the abdomen.
- Right sided CDH:
    - Presence of liver (homogenous mass) in the right chest, often results in left mediastinal shift
    - Mid-thoracic or posterior-thoracic position of the stomach
    - Bowel and gallbladder may herniate
    - Pleural fluid is often present
- Peristalsis of bowel in the chest helps to distinguish CDH from an intra-thoracic mass
- Complex:
    - Genetic and non-genetic syndromes:
        - Trisomy 18, tetrasomy 12p. Deletions of 2q, 4p and 8p
        - Smith-Lemli Opitz, CHARGE, Goldenhar, Beckwith-Wiedemann, Fryn and Noonan Syndromes.
    - Anomalies:
        - Cardiac, renal, central nervous system, and gastrointestinal
            - 28% of CDH has associated cardiac anomaly.
- Other associated findings:
    - Polyhydramnios may be present due to oesophageal compression
    - Hydrops can occur from compression of great vessels.
- Specific measurement to determine prognosis:
    - Observed/Expected Lung-to-head ratio (O/E LHR)
        - Validated method of assessing contralateral lung size as a predictor of mortality and short-term morbidity.
        - This may guide decision making when performed at 22-26 weeks.
        - Better prediction when measured 32-33 weeks and this will guide postnatal prognostic counselling.
    - 2D measurement of contralateral lung, measured in the 4 chamber view of thorax – longest axis and 90 degree to this or “trace method”
    - Head value is the fetal head circumference (HC)
    - Does not predict the status of the pulmonary vascular bed.
- The following calculators allow clinicians to calculate the O/E LHR:
    - http://www.perinatology.com/calculators/LHR.htm
    - https://totaltrial.eu/?id=6

Investigation

- Referral to regional Fetal Medicine Hub
- Fetal genetic studies – amniocentesis for microarray
- MRI: Assessment of fetal lung volume ratio
    - MRI O/E LHR is more accurate than ultrasound.

    - Aids clarity where antenatal imaging is suboptimal and can increase detection of liver herniation.
- Screening fetal echocardiogram

Prognosis:

- Prognostic factors:
    - Liver herniation – predicts a worse prognosis
    - Right vs. left sided lesion – right sided lesion associated with worse prognosis
    - Bilateral CDH: poor prognosis
    - Non- isolated: prognosis is usually poor
    - Observed to Expected Lung area to head circumference ratio (O/E LHR)
        - O/E LHR:
            - extreme <15%
            - Severe 15-25%
            - Moderate 26-35%, 35-44.9% with liver herniation
            - Mild 36-45%, >45% with liver herniation.
- In-utero fetal death rate is around 2% without demonstrable direct cause.

Postnatal:

- Survival and morbidity:
    - Survival is improved by antenatal detection
        - Severe lung hypoplasia: the vast majority die despite intensive care
        - Moderate lung hypoplasia: 40-60% survival
            - 30% require oxygen therapy for at least one month after birth
        - Mild lung hypoplasia: 60-90% survival
    - Survivors may suffer from chronic lung disease, persistent pulmonary hypertension, gastro-oesophageal reflux, feeding problems or thoracic deformations.

Treatment

- Multidisciplinary counselling
    - MFM specialist, neonatologist, paediatric surgeon, geneticist, paediatric intensivist
    - Include the Perinatal Palliative Care team if a severe prognosis or for planned palliative care
- Fetal treatment
    - In utero therapy is offered in Brisbane Australia for those who meet criteria. This may improve survival for those with severe CDH. Note: the use of FETO for moderate CDH is currently in equipoise. Please see document for more detail regarding access to FETO for severe cases: https://www.healthpoint.co.nz/public/wahi-rua-new-zealand-maternal-fetal-medicine/?solo=otherList&index=7
- Followup:
    - Fortnight scan for fetal growth and to exclude polyhydramnios.
- Timing and mode of delivery:
    - Timing: to consider delivery between 38 to 39 weeks gestation.
    - Mode of delivery: caesarean for standard obstetric indications only.
- Perinatal Palliative Care:
    - Perinatal palliative care can be provided currently in Auckland, Middlemore, Wellington and Christchurch

    - It is highly recommended for those with a risk of poor prognosis eg severe lung hypoplasia moderate lung hypoplasia, liver herniation, bilateral CDH, and low LHR.
- Introduction to perinatal palliative is recommended at approximately 24 weeks gestation and would continue to birth and after birth depending on the neonatal symptoms and family distress.

Recurrence risk

- Isolated: 1-2%
- Multiple congenital anomalies of unknown aetiology: <5%
- Referral to genetic counsellor for genetic work-up for recurrence risk of specific chromosomal abnormalities and syndromes.

This Recommendation of Practice was updated in March 2023 by Dr Jaynaya Marlow with input from members of Wāhi Rua NZMFM Network.

The most up to date version of this Recommendation of Practice can be found on Healthpoint Wāhi Rua: New Zealand Maternal Fetal Medicine Network (NZMFM) webpages: https://www.healthpoint.co.nz/public/wahi-rua-new-zealand-maternal-fetal-medicine/

References

- Bouchghoul H et al. Optimal gestational age at delivery in isolated left-sided congenital diaphragmatic hernia. Ultrasound Obstet Gynecol 2021;57: 968 – 973
- Deprest J, Brady P, Nicolaides K, Benachi A, Berg C, Vermeesch J, Gardener G, Gratacos E. Prenatal management of the fetus with isolated congenital diaphragmatic hernia in the era of the TOTAL trial. Seminars in Fetal & Neonatal Medicine 2014. 19: 338-348
- Graham G, Devine P. Antenatal diagnosis of congenital diaphragmatic hernia. Seminar Perinatology 2005; 29:69
- Lally K, Engle W. Post-discharge follow-up of infants with congenital diaphragmatic hernia. Pediatrics 2008; 121:627-632
- Marlow J and Thomas J. A review of congenital diaphragmatic hernia. AJUM 2013 16-21
- Stevens T, van Wijngaarden E, Ackerman K. Timing of delivery and survival rates for infants with prenatal diagnoses of congenital diaphragmatic hernia. Pediatrics 2009; 123:494
- Stoll C, Alembik Y, Dott B, Roth M. Associated malformations in cases with congenital diaphragmatic hernia. Genetic Counselling. 2008: 19(3):331-339
- Yamamoto M, Ville Y. Fetal lung assessment in congenital diaphragmatic hernia: evidence for growth. Ultrasound Obstetric Gynecol. 2010; 35:522-524
- http://www.totaltrial.eu