

Congenital Diaphragmatic Hernia



This guideline was updated in December 2015 by Dr Joana DeSousa, with input from members of the New Zealand Maternal Fetal Medicine Network.



Background

Congenital diaphragmatic hernia (CDH) is a rare disease with a prevalence ranges between 1 and 4 per 10,000 births. It is a diaphragmatic defect which allows abdominal content to herniate into the chest. It can 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.

It 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, most commonly T13, 18 and T21. It may also occur in rare syndromes such as Fryn's and Pallister Killian syndrome. Vast majority of the cases occurs sporadically.

Objective

To guide the accurate diagnosis, investigation, to define individual prognosis of the fetus and management of women presenting with congenital diaphragmatic hernia.

Definition

It is a developmental discontinuity of the diaphragm that allows abdominal content (may include stomach, bowel and liver) to herniate into the chest.

Different Diagnosis

- Congenital pulmonary adenomatoid malformation (CPAM)
- Bronchopulmonary sequestration (BPS)

- 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.

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 just absent stomach in the abdomen)
 - Liver may be herniated. (Liver in the chest can be located by following the course of the intrahepatic by using colour Doppler ultrasound)
 - Gallbladder and hepatic or umbilical veins may be abnormally located within the abdomen
 - Peristalsis of bowel in the chest helps to distinguish CDH from an intra-thoracic mass





- **Right sided CDH:**
 - Presence of liver (homogenous mass) in the right chest, often resulted in left mediastinal shift
 - Bowel and gallbladder may hernia
 - Pleural fluid is often present
- **Complex:**
 - Other abnormalities associated with other syndromes, such as cardiac defects (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:**
 - Lung area to head circumference ratio (LHR)
 - 2D perpendicular linear measurement of contralateral lung, measured in the 4 chamber view of thorax (in mm²) – longest axis and 90 degree to this or
 - “trace method”
 - Divided by head circumference (in mm).

Note:

<http://www.perinatology.com/calculators/LHR.htm> allows clinicians to input measurements and to calculate LHR and observed/expected LHR.



Investigation

- Referral to Fetal medicine unit
- Fetal genetic studies – amniocentesis:
 - Karyotype if isolated
 - Microarray if multiple abnormalities
- MRI: Does not improve assessment of lung hypoplasia
 - Have a role in the quantitative assessment of liver herniation
- Screening fetal echocardiogram

Prognosis

Prognostic factors:

- Liver herniation – reliable predictor of postnatal survival
- Fetal lung volume – optimum equation has not been determined
- Right vs. left sided lesion – right sided lesion associated with worse prognosis
- Lung area to head circumference ratio (LHR or Observed/Expected LHR)
 - more indicative of morbidity than mortality
 - LHR: Poor prognostic group <1 (poorer if liver is herniated)
 - Moderate 1.0 – 1.4
 - Better prognosis > 1.4
 - O/E LHR: extreme $<15\%$
 - Severe 15-25%
 - Moderate 26-35%
 - Mild 36-45%



- In-utero death rate is around 2% without demonstrable direct cause.
- When the condition is not isolated, prognosis is usually poor.

Postnatal:

Survival and morbidity:

- Severe lung hypoplasia: 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, neonatal surgeon, geneticist, paediatric intensivist.

Fetal treatment

Currently in-utero therapy is not available in New Zealand

Follow-up

Fortnight scan for fetal growth and to exclude polyhydramnios

Timing and mode of delivery

- Timing: to consider delivery by 40 weeks gestation
- Mode of delivery: caesarean for standard obstetric indications only



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

References

- 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
- <http://www.totaltrial.eu>
- Graham G, Devine P. Antenatal diagnosis of congenital diaphragmatic hernia. *Seminar Perinatology* 2005; 29:69
- 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
- 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
- Lally K, Engle W. Post-discharge follow-up of infants with congenital diaphragmatic hernia. *Pediatrics* 2008; 121:627-632