

Prenatal diagnosis and prognostication of congenital diaphragmatic hernia

Nimrah Abbasi MD
Maternal-Fetal Medicine, Mount Sinai Hospital



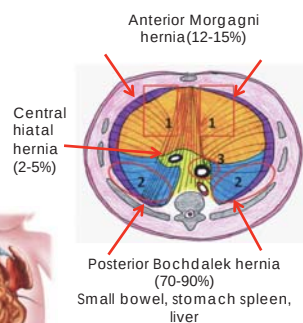
Outline

- Background
- Associated anomalies & genetic syndromes
- Prenatal diagnosis
- Prognostic indicators
 - Lung area (o/e LHR)
 - Intra-thoracic liver herniation
 - Role of MRI

Background

1/2500-1/5000 (*hidden mortality)

- ~85% left sided
- 13% right sided
- 2 % bilateral
- Rarely: eventration/ complete diaphragmatic agenesis

¹Russo *et al.* Prenatal Diagnosis. 2018;38:629–637

Associated abnormalities

Genetic abnormalities¹

[illegible]

Largest CDH series (n=256 CDH), chromosome & subchromosome abnormalities in 6.3%²

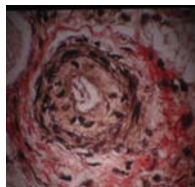
- Isolated ~ 60-70%, survival ~60%
- Associated structural and genetic abnormalities in 30-40%, survival ~15%¹
 - Cardiovascular (25-50%)
 - GU (5-10%)
 - CNS (1-10%)
 - MSK (1-15%)
 - GI (2-10%)
 - Chest (BPS/CCAM 2-5%)

¹Russo et al. Prenatal Diagnosis. 2018;38:629–637
²Yu et al. J Med Genet 2012; 49: 650-59

Neonatal morbidity & mortality

Pulmonary hypoplasia

Pulmonary hypertension



Antenatal prognostication

Prenatal diagnosis

60% prenatal detection rate

4-chamber view

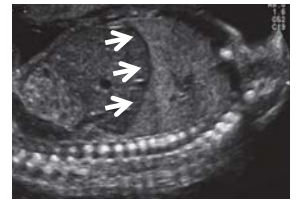
- Axial (single ribs)
- Echogenicity
- Anechoic/cystic structures
- Mediastinal shift



Differential diagnosis of lung lesions

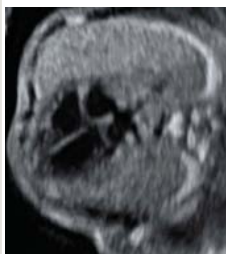
Solid lesions	Cystic lesions
Cystic adenomatoid malformation (microcystic)	Cystic adenomatoid malformation (macrocystic)
Bronchopulmonary sequestration	Bronchogenic cyst
Mediastinal teratoma	Mediastinal neuroblastoma
Rhabdomyoma	Congenital diaphragmatic hernia
Right sided congenital diaphragmatic hernia	Pericardial and pleural effusions
Tracheo/laryngeal atresia	Congenital lobar emphysema

Bush et al. Prenat Diagn. 2008



Diaphragm- Hypoechoic/dome-shaped

Mediastinal shift



Normal



Left CDH



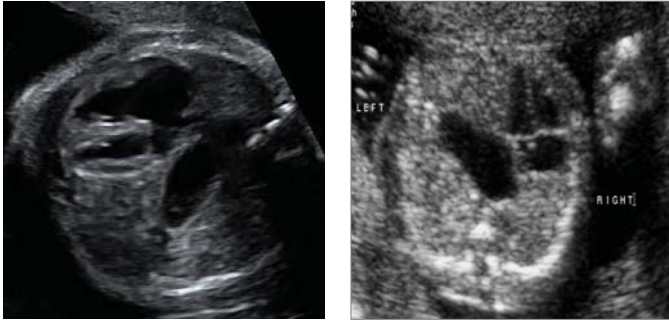
Right CDH

Abnormal/mixed echogenicity



Peristalsis in thorax

Anechoic/cystic structures (Left CDH- Stomach up)



Left CDH- Stomach down



Dextrocardia



Gall bladder & bladder



Stomach & bladder

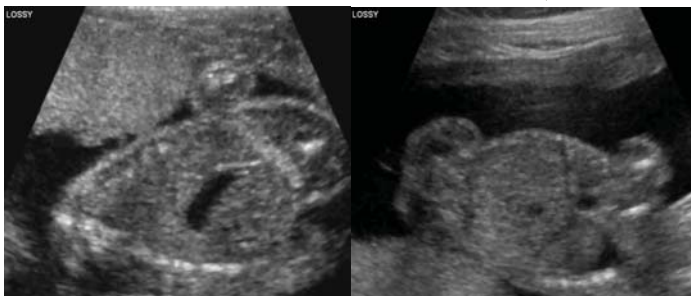


Gall bladder, stomach & bladder

Morgan T et al. J Ultrasound Med 2016

Normal fetal
breathing

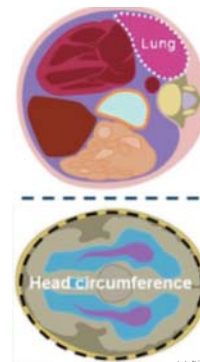
Paradoxical
Respiration



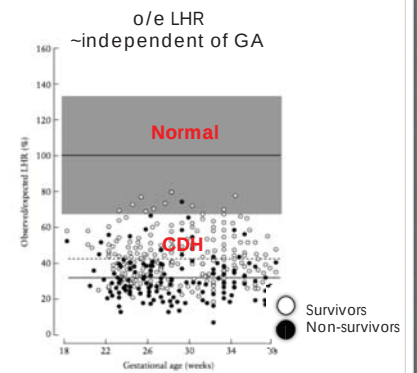
Normal
Diaphragm

Left CDH

observed-to-expected LHR (o/e LHR)



Lung-to-head ratio (LHR)



Ultrasound Obstet Gynecol 2012; 39: 2-6.

