

# Prenatal diagnosis and prognostication of congenital diaphragmatic hernia

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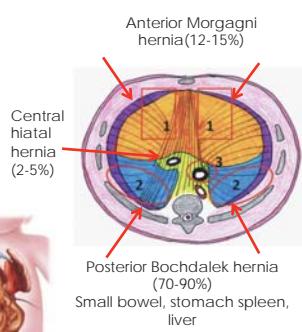
## Outline

- 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



<sup>1</sup>Russo et al. Prenatal Diagnosis. 2018;38:629-637

## Associated abnormalities

### Genetic abnormalities<sup>1</sup>

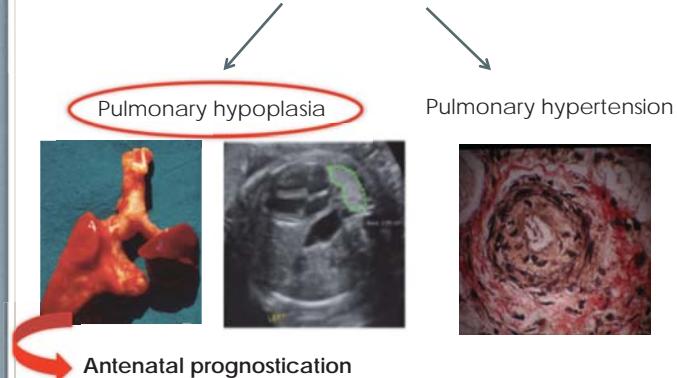
Common anomalies	
1. Anaploids	CDH, cardiac defects, increased limb, central nervous system anomalies, craniofacial anomalies, neural tube defects, skin pigmentation, hydramnios, polyhydramnios. <sup>2</sup>
2. Polyploid (48, 51, 54, 57, 69, 72, 84, 96, 108, 120)	Tetralogy 21, 18, 13, 22, 16
3. Trisomies (21, 22, 23, 23, 23, 23, 23)	CDH, cardiac defects, craniofacial anomalies, growth restriction <sup>3</sup>
4. Monosomy 21	CDH, cardiac defects, growth restriction, pulmonary hypoplasia, fetal hydrops, diaphragmatic and tracheobronchial tubercles and a single umbilical artery <sup>4</sup>
5. Partial trisomy 4p (Wolf-Hirschhorn syndrome)	CDH, facial dysmorphisms, microcephaly, cardiac defects, growth restriction <sup>5</sup>
6. Deletions	Chromosome 16p11.2, chromosome 14q11.2, 14q31.1, 14q32.2 <sup>6</sup>
7. Microdeletion syndromes	14q32.2 deletion, <sup>7</sup> 15q11.2 deletion <sup>8</sup>
8. 22q11.2 deletion	CDH, microcephaly, craniofacial anomalies, postural instability, hemispheric calcification, developmental delay <sup>9</sup>
9. 12p11.21/HOX11 and other genes, 12q13.13	CDH, microcephaly, amniotic fluid, renal hypoplasia <sup>10</sup>
10. 12q13.13/Mesomeric syndrome	CDH, conotruncal anomalies, microcephaly, facial anomalies <sup>11</sup>
11. 1p36.11/Microdeletion with linear skin defects	CDH, facial anomalies, hypoplasia, dermal aplasia, vesicle <sup>12</sup>
12. POCN, Xq22.3 Goltz syndrome	CDH, facial synkinesis, hyperplasia, digital anomalies <sup>13</sup>
13. PRKAG2/12p11.21 Conotruncal syndrome	CDH, nephromegaly, pectus excavatum, growth delay, male pseudogynecomastia <sup>14</sup>
14. NTG, 13q14.1 Derin-Doran syndrome	CDH, nephromegaly, pectus excavatum, growth delay, male pseudogynecomastia <sup>15</sup>
15. Syndromes with unknown genes	
16. Prune belly syndrome	Left-sided CDH, pulmonary hypoplasia, hypoplasia of the distal phalanges and ribs, cryptorchidism, undescended testes, hydronephrosis, and ureteral stenosis, cardiac defects, renal dysplasia and perinecystic infiltration <sup>16</sup>
17. Genomic imprinting	CDH, amniotic fluid, renal hypoplasia <sup>17</sup>
18. Cerebral palsy	Anomalous CDH, amniotic fluid, intra-abdominal, ectopic cord, and congenital heart defects as vertebral spinal defects and ventriculo-uterine fistulae <sup>18</sup>

Largest CDH series (n=256 CDH), chromosomal & subchromosome abnormalities in 6.3%<sup>2</sup>

<sup>1</sup>Russo et al. Prenatal Diagnosis. 2018;38:629-637

<sup>2</sup> Yu et al. J Med Genet. 2012; 49: 650-59

## Neonatal morbidity & mortality



## Prenatal diagnosis

60% prenatal detection rate

### 4-chamber view

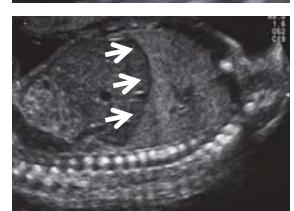
- Axial (single ribs)
- Echogenicity
- Anechoic/cystic structures
- Mediastinal shift



### Differential diagnosis of lung lesions

Solid lesions	Cystic lesions
Cystic adenomatoid malformation (microcystic)	Cystic adenomatoid malformation (macrocystic)
Hemochorionic separation	Bronchogenic cyst
Mediastinal teratoma	Mediastinal emphysema
Rhabdomyoma	Congenital diaphragmatic hernia
Right sided congenital diaphragmatic hernia	Pericardial and pleural effusions
Tracheal/laryngeal stenosis	Congenital lobar emphysema

Bush et al. Prenat Diagn. 2008



Diaphragm: Hypoechoic/dome-shaped

## Mediastinal shift



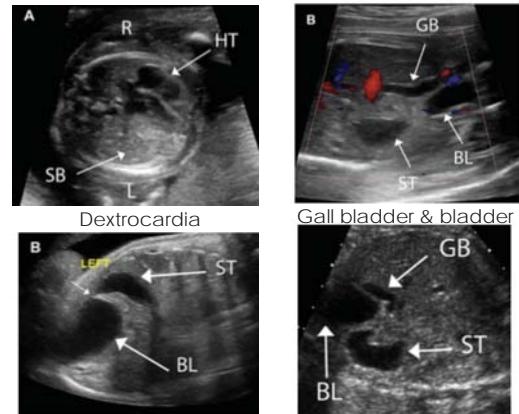
## Abnormal/mixed echogenicity



## Anechoic/cystic structures (Left CDH- Stomach up)



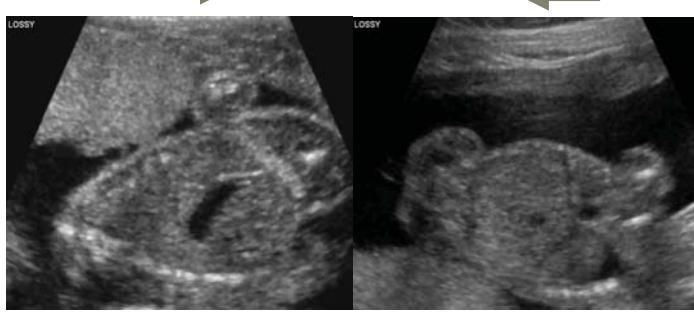
## Left CDH- Stomach down



Morgan T et al. J Ultrasound Med 2016

### Normal fetal breathing

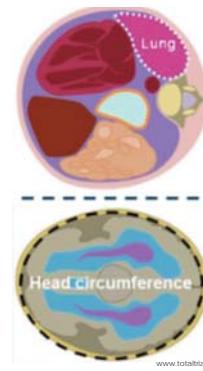
### Paradoxical Respiration



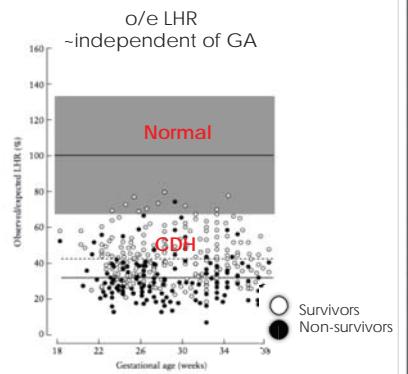
Normal Diaphragm

Left CDH

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



Lung-to-head ratio (LHR)



Ultrasound Obstet Gynecol 2012; 39: 2–6.







