Prenatal ultrasound diagnosis of congenital thoracic malformations

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Introduction

• Congenital thoracic malformations (CTMs) - a spectrum of lesions
• Originate in the embryonic period; a potential influence on pulmonary structure and function
• 1/10,000-1/35,000 pregnancies*
• Some regress or even disappear during pregnancy, but are usually still visible on postnatal CT
• Mostly detected on routine second trimester screening (GA 18-20wks)
• Varying clinical presentation and severity - severe lesions with nonimmune fetal hydrops → postnatal complications → incidental findings in adult age

* Witlox et al., Prenat Diagn 2011
Introduction - 2

• Early first-trimester US - confirming fetal viability, accurate date, number and size of fetuses, nuchal translucency, major fetal anomalies
• Second trimester US - fetal anatomy, structural anomalies
• With improving equipment and user skill, there is a shift of the second trimester US to the earliest possible GA
• Detection rate of fetal malformations at GA 12–14 wks lower than 15–22 wks (38% vs. 47%)*
• BUT - reassurance if normal; enables early TOP with lethal malformations

* Saltvedt et al., Obstet Gynaecol 2006
Introduction - 3

• Our Pediatric Pulmonary Institute follows an increasing number of patients with CTMs
• A true increased incidence or better sensitivity of the prenatal US?
• The extent and nature of CTMs that could be detected by earlier US is underreported
Aims

• Part 1:
  - To evaluate prenatal US findings of CTMs between 2001-2017
  - To estimate changes in detection rates of CTMs

• Part 2: To describe the spectrum and rate of CTMs diagnosed by early prenatal US (GA<16 wks)
Methods

- A single center retrospective study
- Computerized database of all fetal US, 2001-2017 in a single community-based clinic (AL-KOL, Haifa, Prof. Bronshtein)
- Examinations performed as a second opinion were excluded
- CTMs were identified, as well as additional lesions – a single lesion/ multiple lesions/ hydrops
- **Part 2** – analysis of CTMs detected <16wks, including outcome (when available)
Results – part 1

- A total of 34,716 prenatal US → 49 CTMs
- Median GA 15.4wks (11.6-23.9) in 2001-2007; 15.7wks (12-33.6) weeks in 2007-2017
- 6 types of lesions:
  - **CPE** – congenital pleural effusion
  - **CPAM** - congenital pulmonary airway malformation
  - **CDH** – congenital diaphragmatic hernia
  - **PH** - pulmonary hypoplasia
  - **BPS** – bronchopulmonary sequestration
  - **CLE** – congenital lobar emphysema
Detected pulmonary lesions

CDH - 13
CLE - 1
PH - 3
BPS - 2
CPAM -13
CPE - 17

CPE= congenital pleural effusion; CPAM= congenital pulmonary airway malformation; CDH= congenital diaphragmatic hernia; CLE= congenital lobar emphysema ; PH= pulmonary hypoplasia; BPS= Broncho-pulmonary sequestration;
Detected pulmonary lesions – cont.

<table>
<thead>
<tr>
<th></th>
<th>Total</th>
<th>Single lesion</th>
<th>Multiple lesions</th>
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</thead>
<tbody>
<tr>
<td>CPE</td>
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<td>6</td>
<td>11</td>
</tr>
<tr>
<td>CPAM</td>
<td>13</td>
<td>13</td>
<td>0</td>
</tr>
<tr>
<td>CDH</td>
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<td>7</td>
<td>6</td>
</tr>
<tr>
<td>PH</td>
<td>3</td>
<td>0</td>
<td>3</td>
</tr>
<tr>
<td>BPS</td>
<td>2</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>CLE</td>
<td>1</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Total</td>
<td>49</td>
<td>29</td>
<td>20</td>
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</tbody>
</table>

CPE= congenital pleural effusion; CPAM= congenital pulmonary airway malformation; CDH= congenital diaphragmatic hernia; PH= pulmonary hypoplasia; BPS= Broncho-pulmonary sequestration; CLE= congenital lobar emphysema
Comparison of detection rates

Detection rates did not change (1.32/1000 in 2007-2017 vs 1.58/1000 in 2001-2007, p=0.64)

2001-2007 – 12,016 tests, 19 CTMs
2007-2017 – 22,700 tests, 30 CTMs
Results - part 2 (early diagnosis)

• 31,261 early prenatal US → 31 CTMs
• Median GA 15.2wks (11.6–16.0)
• CPE (15 fetuses) → CDH (10 fetuses) → PH (3 fetuses) → CPAM (2 fetuses) → BPS (1 fetus)
• 15 additional lesions; 4 hydrops
Rates of early detection
Detected lesions, additional lesions and follow up

<table>
<thead>
<tr>
<th>Lesion</th>
<th>GA</th>
<th>Additional lesions</th>
<th>Follow up</th>
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<tbody>
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<td>CPE</td>
<td>12+4</td>
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<td>TOP</td>
</tr>
<tr>
<td>CPE</td>
<td>14+6</td>
<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>CPE</td>
<td>15+3</td>
<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>CPE</td>
<td>12+4</td>
<td>multiple</td>
<td>missed</td>
</tr>
<tr>
<td>CPE</td>
<td>15+0</td>
<td>multiple</td>
<td>missed</td>
</tr>
<tr>
<td>CPE</td>
<td>13+2</td>
<td>multiple</td>
<td>unknown</td>
</tr>
<tr>
<td>CPE</td>
<td>14+0</td>
<td>multiple</td>
<td>unknown</td>
</tr>
<tr>
<td>CPE</td>
<td>14+6</td>
<td>multiple</td>
<td>resolved</td>
</tr>
<tr>
<td>CPE</td>
<td>14+4</td>
<td>hydrops</td>
<td>TOP</td>
</tr>
<tr>
<td>CPE</td>
<td>15+2</td>
<td>hydrops</td>
<td>unknown</td>
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<tr>
<td>CPE</td>
<td>15+2</td>
<td>hydrops</td>
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</tr>
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<td>14+2</td>
<td>none</td>
<td>unknown</td>
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<td>CPE</td>
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</tr>
<tr>
<td>CPE</td>
<td>16+0</td>
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</tbody>
</table>

CPE= congenital pleural effusion; GA= gestational age; TOP=termination of pregnancy
CPE - summary

• 15 cases of CPE; 6 unfavorable outcome
• 4 TOP - trisomy 18, single umbilical artery, axillary cyst and VSD, cystic hygroma, amputation of hand and cardiomyopathy
• 8 multiple lesions – 3 TOP, 2 missed, 2 unknown, 1 resolved (initial echogenic bowel & hydrocephalus)
• 3 hydrops – 1 TOP, 1 unknown, 1 resolved (pericardial effusion in a twin)
• 4 isolated CPE → 3 resolved, 1 unknown (parvo virus)
Detected lesions, additional lesions and follow up – cont.

<table>
<thead>
<tr>
<th>Lesion</th>
<th>GA</th>
<th>Additional lesions</th>
<th>Follow up</th>
</tr>
</thead>
<tbody>
<tr>
<td>CDH</td>
<td>11+4</td>
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<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
<td>15+1</td>
<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
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<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
<td>15+6</td>
<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
<td>12+0</td>
<td>hydrops</td>
<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
<td>15+1</td>
<td>none</td>
<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
<td>15+1</td>
<td>none</td>
<td>TOP</td>
</tr>
<tr>
<td>CDH</td>
<td>16+0</td>
<td>none</td>
<td>TOP</td>
</tr>
<tr>
<td>PH</td>
<td>15+1</td>
<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>PH</td>
<td>15+5</td>
<td>multiple</td>
<td>TOP</td>
</tr>
<tr>
<td>PH</td>
<td>16+0</td>
<td>multiple</td>
<td>TOP</td>
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<tr>
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<td>asymptomatic</td>
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<tr>
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<td>15+6</td>
<td>none</td>
<td>unknown</td>
</tr>
<tr>
<td>BPS</td>
<td>15+4</td>
<td>none</td>
<td>Resolved</td>
</tr>
</tbody>
</table>

CDH= congenital diaphragmatic hernia; PH= pulmonary hypoplasia; CPAM= congenital pulmonary airway malformation; BPS= broncho-pulmonary sequestration; GA= gestational age; TOP= termination of pregnancy
CDH detected in early ultrasound (US) examination

Upper images – fetus no. 1 – transverse (right) and sagittal (left) views of left CDH, week 15. Arrows point to the intestine encircling the heart.

Lower image – fetus no. 2 – right CDH, week 16. The liver is in the thoracic cavity.
Early prenatal ultrasound diagnosis of congenital thoracic malformations

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\textbf{ABSTRACT}

\textbf{Objectives:} To evaluate and describe the spectrum and rate of congenital thoracic malformations (CTMs) diagnosed by early prenatal sonography (gestational age (GA) less than 16 weeks).

\textbf{Methods:} A retrospective, cross-sectional analysis of prenatal ultrasound screening tests in a community-based clinic.

\textbf{Results:} In 2001–2017, 31 261 prenatal ultrasound tests detected 31 CTMs at a gestational age of 15.2 (range, 11.6–16.0) weeks. The most common malformation was congenital pleural effusion (CPE) (15 fetuses, 0.48/1000), followed by congenital diaphragmatic hernia (CDH) (10 fetuses, 0.32/1000). Pulmonary hypoplasia (PH), congenital pulmonary airway malformation and bronchopulmonary sequestration appeared in much smaller proportions (three, two and one fetuses, respectively). Most CTMs were associated with additional fetal lesions (15 fetuses, 48%). All early CDH (10 fetuses) and PH (three fetuses) and 6/15 with CPE had termination of pregnancy or missed abortions.

\textbf{Conclusions:} Prenatal ultrasound before 16 GA was able to detect CTMs in 0.99/1000 of screening ultrasound (US) performed. Most CTMs tended to appear with multiple lesions and were associated with unfavorable outcomes. Earlier prenatal diagnosis may enable early termination of pregnancy in fetuses with lethal malformations.

\textbf{Introduction}

Early first-trimester ultrasound (US) is regularly used to assess the risk of congenital anomalies. However, the detection of congenital thoracic malformations (CTMs) that could be detected by earlier US is underreported.
Limitations & strengths

• Limitations –
  ✓ Retrospective nature
  ✓ Information about outcome could not be obtained in all pregnancies
  ✓ A private clinic; high rate of TOP may not be applicable to other populations

• Strength –
  ▪ A reliable single center, large database performed by the same highly-experienced sonographer
Summary & conclusions

- CDH and CPE tend to appear with multiple lesions, warrant further attention
- Detection rates stable - last decade vs. previous years →→ ↑ studies performed, not a true higher incidence
- Prenatal US <16 GA detected 0.99/1000 CTMs - multiple lesions; unfavorable outcomes
- Earlier detection of severe malformations - important for appropriate counseling
- Further studies, with longitudinal follow-up and outcome are warranted
Thanks:
✓ Dr. Mordechai Pollak
✓ Prof. Moshe Bronshtein
✓ Dr. Ido Solt
✓ Prof. Lea Bentur