Natural History and Outcomes of Antenatally Diagnosed Abdominal Wall Defects

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Objective

- To review the natural history and outcomes of antenatally diagnosed abdominal wall defects (AWD) at a single tertiary centre
Methods

• Review of cases of prenatally diagnosed AWD from the database of the Fetal Medicine Unit at Hospital Italiano de Buenos Aires from January 2004 to January 2010
There were 67 cases of AWD:

- 22 exomphalos
- 26 gastroschisis
- 19 classified as “others”, which included:
  - 10 cases of limb-body wall complex (LBWC)
  - 6 cases of body-stalk anomaly (BSA),
  - 2 cases of pentalogy of Cantrell
  - 1 case of OEIS Complex.
Table 1: Abdominal wall defects: maternal age and gestational age at diagnosis.

<table>
<thead>
<tr>
<th></th>
<th>Exomphalos</th>
<th>Gastrochisis</th>
<th>Others</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>LBWC</td>
</tr>
<tr>
<td>Cases (n)</td>
<td>22</td>
<td>26</td>
<td>10</td>
</tr>
<tr>
<td>Maternal age (mean +/- SD)</td>
<td>32 (*)  $(\pm 7.77)$</td>
<td>24 (*) $(\pm 4.35)$</td>
<td>27 $(\pm 5.85)$</td>
</tr>
<tr>
<td>Gestational age at diagnosis (mean +/- SD)</td>
<td>20 $(\pm 7.03)$</td>
<td>23 $(\pm 6.84)$</td>
<td>20 $(\pm 5)$</td>
</tr>
<tr>
<td>BSA</td>
<td></td>
<td></td>
<td>6</td>
</tr>
<tr>
<td>PC</td>
<td></td>
<td></td>
<td>2</td>
</tr>
<tr>
<td>OEIS</td>
<td></td>
<td></td>
<td>1</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>28.5 $(\pm 10.6)$</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>30</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>24.5 $(\pm 15)$</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>33</td>
</tr>
</tbody>
</table>

(* p=0.0001

LBWC: limb-body wall complex; BSA: body-stalk anomaly; PC: pentalogy of Cantrell
Results

Exomphalos

- Of the 22 cases of *exomphalos*, 11 cases were aneuploid, 7 euploid cases were associated with other structural anomalies, and 4 cases were isolated (Table 2)
### Table 2: Exomphalos and gastroschisis: isolated and non-isolated defects.

<table>
<thead>
<tr>
<th></th>
<th>Exomphalos</th>
<th>Gastroschisis</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Cases (n)</strong></td>
<td>22</td>
<td>26</td>
</tr>
<tr>
<td><strong>Isolated defect n (%)</strong></td>
<td>4/22 (18.2%)</td>
<td>25/26 (95%)</td>
</tr>
<tr>
<td><strong>Non-isolated defect n (%)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Normal Karyotype</td>
<td>7/22 (31.8%)</td>
<td>1/26 (5%)</td>
</tr>
<tr>
<td>Abnormal Karyotype</td>
<td>11/22 (50%)</td>
<td>0</td>
</tr>
</tbody>
</table>
Results

- Overall mortality rate in this group was 14/19 (74%), with 3 ongoing pregnancies.

- Perinatal mortality rate was:
  - 10/10 in aneuploid cases (Figure 1-2)
  - 2/5 in euploid cases with associated anomalies
  - 2/4 in the isolated subgroup (Figure 1 and Table 3).
Figure 1: Outcomes of antenatally diagnosed exomphalos.

Exomphalos
n=22

Isolated
n=4 (18.2%)

Livebirth
n=4

Neonatal death
n=2 (50%)

A&W
n=2 (50%)

Non-isolated
n=18 (81.8%)

Normal Karyotype
n=7 (39%)

IUFD
n=2 (28.5%)

Livebirth
n=3 (43%)

On going
n=2 (28.5%)

Abnormal karyotype
n=11 (61%)

IUFD
n=7 (64%)

Neonatal death
n=3 (27%)

On going
n=1 (9%)

IUFD: Intrauterine Fetal Death; A&W: Alive and well
Figure 2: Exomphalos with aneuploidy.

- Exomphalos & abnormal karyotype n=11
  - Trisomy 13 n=4
    - NND n=2
    - IUFD n=1
    - TOP n=1
  - Unbalanced traslocation n=1
    - 46,XX,t(1;5)(p22;q31)
    - IUFD n=1
  - Trisomy 18 n=6
    - IUFD n=5
    - NND n=1

TOP: Termination of pregnancy
Results

Exomphalos

• Mean gestational age at delivery of liveborns: 34 weeks (± 5.6)
Results

Gastroschisis

In the group of gastroschisis:

- 25/26 were isolated
- 1 fetus had a ventricular septal defect
- There are 3 ongoing pregnancies
- 1 TOP (MC twin pregnancy with TTTS)

(Figure 3 and Table 3)
Figure 3: Outcomes of antenatally diagnosed gastroschisis.

- **Gastroschisis**
  - **n=26**
    - **Isolated**
      - **n=25 (95%)**
        - **Livebirth**
          - **n=20 (80%)**
            - Neonatal death
              - **n=2 (10%)**
            - **A&W**
              - **n=18 (90%)**
        - **TOP**
          - **n=1 (4%)**
        - **IUFD**
          - **n=1 (4%)**
    - **Non-isolated**
      - **n=1 (5%)**
        - Normal karyotype
          - **IUFD n=1 (CIV)**
  - **On going**
    - **n=3 (12%)**

IUFD: Intrauterine Fetal Death
A&W: Alive and well
Results

Gastroschisis

- Perinatal mortality rate was 4/22 (18%):
  - 2 Intrauterine deaths
  - 2 Neonatal deaths
Results

Gastroschisis

- Mean gestational age at delivery of liveborns: 35.2 weeks (± 4.2)
Results

Others

- In the group of “Others”, only the case with OEIS complex survived.
- There were:
  - 2 TOP
  - 10 IUD
  - 6 neonatal deaths
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<tbody>
<tr>
<td><strong>Cases (n)</strong></td>
<td>22</td>
<td>26</td>
<td>10</td>
</tr>
<tr>
<td><strong>TOP (n)</strong></td>
<td>0</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td><strong>IUFD (n)</strong></td>
<td>9 (*)</td>
<td>2</td>
<td>6</td>
</tr>
<tr>
<td><strong>Neonatal death (n)</strong></td>
<td>5(*)</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td><strong>Total Death</strong></td>
<td>14/19(*)</td>
<td>5/23(*)</td>
<td>10</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
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<th>BSA</th>
<th>PC</th>
<th>OEIS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cases</td>
<td>10</td>
<td>6</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>TOP</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>IUFD</td>
<td>6</td>
<td>3</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Neonatal death</td>
<td>3</td>
<td>2</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Total Death</td>
<td>10 (100%)</td>
<td>6 (100%)</td>
<td>2 (50%)</td>
<td>0</td>
</tr>
</tbody>
</table>

(*) 3 cases ongoing  (**) One infant hospitalized in intensive care since delivery died at 20 month.
Conclusions

• Among the prenatally diagnosed AWD, only gastroschisis had a favorable outcome (78% of survival rate)

• For non-gastroschisis AWD, prognosis is poor (perinatal mortality rate 84%), especially those associated with other structural anomalies or aneuploides
Conclusions

- Spontaneous or iatrogenic *prematurity* is another feature that affects most cases of viable AWD.
- Therefore, prenatal diagnosis of an AWD needs a comprehensive assessment for a specific diagnosis, and those fetuses with chances of neonatal correction should be managed and delivered in tertiary care centers.