PRENATAL DIAGNOSIS OF RIGHT AORTIC ARCH. A SPANISH MULTICENTER STUDY

Spanish Fetal Cardiology Group

NO CONFLICT OF INTEREST TO DECLARE
Introduction

- The 3-vessels trachea (3VT) view has led to a higher prenatal detection of aortic arch anomalies including right aortic arch (RAA).
- RAA with aberrant left subclavian artery and double aortic arch usually form a vascular ring and RAA with mirror image branching does not.
Introduction

- RAA may lead to mechanical compression of the airway and/or esophagus and may associate congenital heart defects and chromosomal abnormalities.
- In many series of postnatal vascular rings with RAA a high incidence of clinical symptoms are reported and surgery is usually indicated.
- But is it the same for cases detected in prenatal life? Is surgery necessary?
Methods

- Retrospective, multicenter (7 centers: 3 tertiary and 4 secondary cardiology centers) echocardiographic analysis of all cases diagnosed prenatally of RAA (1/2010-12/2011)

- The type of RAA and branching pattern were assessed. Different diagnosis were sought:
  1. RAA with anomalous left subclavian artery "U-shaped vascular loop (ALSA)
  2. Double aortic arch (DAA)
  3. RAA with mirror image branching (MIB)
  4. RAA undetermined type
Methods

- Intracardiac, extracardiac, and chromosomal anomalies were studied between different types of RAA.
- Gestational age (GA) at diagnosis, nuchal translucency (NT) and outcome data were evaluated.
- Differences between secondary and tertiary centers at defining branching pattern were tested.
- Fisher’s exact test was used.
# Results

- **N= 73 patients**

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<th>RAA</th>
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<tr>
<td><strong>GA at diagnosis</strong></td>
<td>Mean: 22.2 weeks&lt;br&gt;Std. Desviacion: 4.2 weeks</td>
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<td><strong>NT</strong></td>
<td>Mean: 1.8 mm&lt;br&gt;Std. Desviacion: 1.1</td>
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<td><strong>Diagnosis at screening Echo</strong></td>
<td>80.8 %</td>
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<td><strong>Prenatal risk factors</strong></td>
<td>Not: 89%</td>
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<td><strong>Interruptions of pregnancy</strong></td>
<td>14 (19%)</td>
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<td><strong>Autopsy study</strong></td>
<td>9/14 (64.3%)</td>
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<td><strong>Neonatal echocardiography confirmation</strong></td>
<td>93%</td>
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<td><strong>CT postnatal</strong></td>
<td>7/59 cases</td>
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<td><strong>MRI postnatal</strong></td>
<td>1/59 cases</td>
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<tr>
<td><strong>Neonatal echocardiography confirmation of ALSA (U-SHAPED )</strong></td>
<td>95 %</td>
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Types of RAA

- N= 73 patients

- DAA
- RAA+MB
- RAA+ALSA
- Undetermined

RAA larger in 100% cases

- 45%
- 10%
- 7%
- 38%
Influence of training in assessment of RAA and branching

Undetermined type or RAA with no evaluation of branching pattern was related to hospitals with fewer cases of fetal RAA/fetal studies per year.

Fisher exact test

(83.3% vs 16.7% p<0.001)
Intracardiac anomalies

- RAA was associated to intracardiac anomalies in 32 cases (44%)

- There was a significant association to other heart defects in the case of RAA + MIB (22 of 28, 78.6%, p<0.001)

- Most fetuses in the groups of ALSA (84.8%, p<0.001), undetermined type (66.7%) and double aortic arch (60% vs 40% NS) had normal hearts
Intracardiac anomalies associated to RAA

- Fallot/PA: 20%
- VSD: 17%
- DORV: 7%
- Truncus: 7%
- Complex anomalies: 49%
Chromosomal abnormalities

Fetal karyotype

34; 47%

39; 53%

- 4 (50%): 22q11 deletion +
- 1: trisomy 21
- 1: Klinefelter syndrome
- 2:
  + 46XX, Arrays: 22q 3c.32
  +46XX, t(y;22)(q12;p13)
Chromosomal abnormalities and types of RAA

- Chromosomal abnormalities were only found in RAA + MIB and in RAA with ALSA.
- No statistically significant differences between these two groups (RAA+MIB 18.75 % vs RAA+ALSA 27.7 %) were found.
- Two 22q11 deletions were found in RAA+MIB (12.5 %) and two were in RAA+ALSA (11 %).
- Of 8 fetuses with RAA + abnormal karyotype 5 (62.5%) had associated intracardiac lesions.
Extracardiac abnormalities and types of RAA

- Extracardiac abnormalities were only detected in 6 fetuses with RAA: 4 RAA + MIB (66.7%) and 2 RAA with ALSA (33.3%)
- No statistically significant differences between these two groups
- 3 of 6 fetuses had genitourinary malformations
Outcome

- Follow-up period of 1-2 years

- Only 3 of 59 patients (73 cases-14 interruptions, 5%) had symptoms due to arch anomalies

  - 1 of 5 patients with DAA (20%): stridor (first year of life)
  
  - 1 of 32 RAA + ALSA (3%): dysphagia (first year of life)
  
  - 1 of 19 RAA *MIB (5%): stridor (first year of life)
Conclusions

- The type of RAA and the branching pattern can be diagnosed antenatally.
- In our study, the most frequent type of RAA in fetal life was RAA + ALSA.
- Associated intracardiac lesions are more common in cases of RAA with mirror-image branching.
Conclusions

- Chromosomal anomalies including 22q11 deletions should be ruled out in cases of RAA +MIB associated to cardiac/extracardiac anomalies but they also have to be considered when the diagnosis is RAA +ALSA.

- Only a minority of patients with vascular rings (DAA and RAA + ALSA) detected in prenatal life had symptoms during the first year of life.