Background and Overview

To establish the frequency of genomic imbalance in fetuses with a diagnosis of right aortic arch and normal cardiac anatomy, particularly focusing on microduplications or microdeletions which would have gone undetected by conventional karyotyping and 6 probe fish (13,18,21, X, Y, TUPLE).

Materials and Methods

Retrospective study of fetal ultrasounds between 2011 and 2016 in a tertiary referral centre in Australia. Outcomes of interest were frequency of aneuploidy, presence of extracardiac anomalies, additional postnatal findings, and need of surgery for vascular ring postnatally.

Results

26 patients were identified: Chromosomal anomalies were identified in 7/21 fetuses (33%) who had chromosomal testing. Two fetuses (10%) had 22q11.2 deletion. Molecular karyotyping identified 3 (14%) CNVs of probable clinical significance and 2 (10%) of unlikely clinical significance. 9% of patients underwent surgery for vascular ring. 29% of infants who reached the age of 2 or older underwent surgery.

Table A Case Summary

<table>
<thead>
<tr>
<th>Type of Abnormality</th>
<th>Total</th>
<th>Probable clinical significance</th>
<th>Uncertain significance</th>
<th>Unlikely significance</th>
<th>Addenbrooke’s population</th>
</tr>
</thead>
<tbody>
<tr>
<td>Right Aortic Arch with ALSA</td>
<td>14</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Right Aortic Arch with mirror image branching</td>
<td>5</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Double Aortic Arch</td>
<td>4</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Indeterminate Branching</td>
<td>3</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Total</td>
<td>26</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>2</td>
</tr>
</tbody>
</table>

Discussion

- Clinically significant chromosomal anomalies have been detected in association with this anomaly which would not have been detected with conventional karyotyping.
- Consent for invasive testing should include counseling about the possibility of findings of uncertain significance.
- Antenatal counseling on prognosis should include the chance of surgery for vascular ring and the importance of long term follow-up.

References

3. F D’Antonio, A Khalil, V Zidere, JS Carvalho. Fetuses with right aortic arch Multicentre cohort study and meta-analysis. 10.1002/uog.15805

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