Chromosome 10p13-14 and 22q11 deletion screening in 100 patients with isolated and syndromic conotruncal heart defects

R Voigt, M Maier-Weidmann, P E Lange, T Haaf

SUBJECTS AND METHODS
Each patient admitted to the German Heart Centre in Berlin for conotruncal heart malformation was carefully examined by an experienced clinician before cardiac catheterisation. One hundred patients (age range 4 days to 58 years, mean 6.1 years), 81 with isolated CTHD and 19 syndromic cases, were included in this study (table 1). Of the syndromic patients, only one presented classical DGS, one VCFS, one Down syndrome, and two had positive family history for cardiac defects. Peripheral blood lymphocyte chromosomes were analysed by fluorescence in situ hybridisation. Cosmids Sc11.1a (D22S427), 443 (D22S941/D22S942), 100c10 (COMT), and

<table>
<thead>
<tr>
<th>Table 1</th>
<th>Frequency of 22q11 deletion in 100 CTHD patients</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>Isolated conotruncal defects</td>
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<tr>
<td>---------</td>
<td>-------------------------------</td>
</tr>
<tr>
<td>Primary diagnosis</td>
<td>Number non-deleted</td>
</tr>
<tr>
<td>TA</td>
<td>4</td>
</tr>
<tr>
<td>DORV</td>
<td>8</td>
</tr>
<tr>
<td>TGA</td>
<td>22</td>
</tr>
<tr>
<td>TOF</td>
<td>28</td>
</tr>
<tr>
<td>PA</td>
<td>14</td>
</tr>
<tr>
<td>PS</td>
<td>3</td>
</tr>
<tr>
<td>Total</td>
<td>79</td>
</tr>
</tbody>
</table>

Abbreviations: CTHD; conotruncal heart defects; VCFS, velocardiofacial syndrome; DGS, DiGeorge syndrome; CTA FS, conotruncal anomaly-face syndrome; TOF, tetralogy of Fallot.
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REFERENCES


