Racial Frequencies in Congenital Heart Disease

By Barry J. Maron, M.D., Judith M. Applefeld, and L. Jerome Krovitz, M.D., Ph.D.

SUMMARY
A retrospective population study of racial frequencies in three congenital heart malformations, transposition of the great vessels, coarctation of the aorta, and aortic atresia with left heart hypoplasia, indicated no significant difference in incidence among Negroes and Caucasians in the Baltimore metropolitan area.

Additional Indexing Words:
Transposition of the great vessels Coarctation of the aorta Hypoplastic left heart syndrome

Since most congenital cardiovascular malformations are probably a result of complex interactions between genetic and environmental factors, racial comparisons may provide clues to the etiologic factors involved. In a retrospective study, Hernandez and associates1 suggested that the frequency of coarctation of the aorta was low in American Negroes, although others have disputed this observation.2 Because the available information regarding racial frequencies of many congenital heart defects is inadequate and often conflicting, we have assembled data on the incidence of coarctation of the aorta, as well as transposition of the great vessels, and hypoplastic left heart syndrome during a 10-year period in the Baltimore metropolitan area.

Materials and Methods
Information on all liveborn infants with transposition of the great vessels, coarctation of the aorta, and hypoplastic left heart syndrome born in Baltimore City or County, 1960–1970, was obtained by review of cardiac catheterization, surgical, and autopsy records. The majority of cases were obtained from five hospitals: Johns Hopkins, University of Maryland, Baltimore City, Sinai, and Mercy. Five infants with chromosomal anomalies (trisomy and Turner’s syndromes) were excluded.

Cases of coarctation of the aorta were included only if the diagnosis was made during the first year of life. These cases represent infantile coarctation of the aorta, regardless of whether aortic constriction was preductal or postductal. Included in the definition of hypoplastic left heart syndrome is aortic valve atresia with hypoplasia of the ascending aorta and left ventricle with or without mitral valve atresia. One case of complete interruption of the aortic arch was excluded. Data were analyzed by the chi-square method.

Results
There were no significant differences in the racial frequencies of complete transposition of the great vessels, coarctation of the aorta, or hypoplastic left heart syndrome when compared with general birth statistics for the same time period in Baltimore City and County (table 1).

Six of the seven cases of partial transposition of the great vessels (i.e. Taussig-Bing malformation and double-outlet right ventricle) occurred in Negroes; none of the 12 cases of complete transposition of the great vessels were in Caucasian infants. However, the total number of both of the anomalies was small and, therefore, the significance of this observation is difficult to interpret.

All three malformations analyzed were more frequent in males (table 2). This sex difference was greatest in hypoplastic left heart syndrome where males were affected about 2½ times more frequently than females. The sex ratio for coarctation of the aorta and complete transposition of the great vessels correspond to those previously reported.3

Discussion
Our data suggest that race has little influence on the incidence of the three congenital cardiac malformations discussed here. The findings regarding coarctation of the aorta are in agreement with those of the prospective National Institutes of Health Collaborative Perinatal Study of over 56,000 births.2 The present data are derived from over 370,000 births and permit analysis of a greater

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number of patients with congenital heart disease in a localized geographic area.

We suspect that the low incidence of coarctation of the aorta reported by Hernandez and co-workers in a retrospective study from Gainesville and Miami, Florida, may be due to sampling bias in three ways: (1) incidence figures for adult-type coarctation of the aorta may be inaccurate since these patients are often asymptomatic and the timing of diagnosis may vary from early childhood to adulthood; (2) it is a hospital-based study which does not control the factors responsible for patients choosing to come to that hospital; and (3) variations in available health care between Negro and Caucasian communities make racial comparisons difficult, particularly regarding asymptomatic or mildly symptomatic forms of congenital heart disease.

Although this study was retrospective, it is unlikely that many cases have been overlooked. Most infants born in the Baltimore metropolitan area with such severe congenital cardiac malformations have had either diagnostic cardiac catheterization, surgery, or postmortem examination within the first year of life since cyanosis is prominent in transposition of the great vessels, congestive cardiac failure common in coarctation of the aorta, and hypoplastic left heart syndrome universally fatal in infancy. Although cardiac catheterization and surgery are rarer in patients with hypoplastic left heart syndrome, accurate comparative racial incidences should be reflected in autopsy records. The frequency of infant autopsies in Baltimore is similar for Negroes (64%) and whites (51%).

A possible source of error in this study exists in the assumption that both Negro and white infants have equal opportunity for cardiac catheterization or surgery. However, since only 10 patients were

Table 1

<table>
<thead>
<tr>
<th>Malformation</th>
<th>Caucasian</th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No.</td>
<td>%</td>
<td>No.</td>
<td>%</td>
<td>Total (no.)</td>
</tr>
<tr>
<td>Complete transposition of great vessels</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Isolated</td>
<td>22</td>
<td>71</td>
<td>9</td>
<td>29</td>
<td>31</td>
</tr>
<tr>
<td>+ VSD or SIV</td>
<td>9</td>
<td>75</td>
<td>3</td>
<td>25</td>
<td>12</td>
</tr>
<tr>
<td>+ PSI + VSD</td>
<td>8</td>
<td>100</td>
<td>0</td>
<td>0</td>
<td>8</td>
</tr>
<tr>
<td>+ PSI</td>
<td>4</td>
<td>80</td>
<td>1</td>
<td>20</td>
<td>5</td>
</tr>
<tr>
<td>+ PDA</td>
<td>19</td>
<td>70</td>
<td>8</td>
<td>30</td>
<td>27</td>
</tr>
<tr>
<td>Complex*</td>
<td>10</td>
<td>59</td>
<td>7</td>
<td>41</td>
<td>17</td>
</tr>
<tr>
<td>Total (χ² = 1.193)</td>
<td>72</td>
<td>72</td>
<td>28</td>
<td>28</td>
<td>100</td>
</tr>
<tr>
<td>Partial transposition of great vessels†</td>
<td>1</td>
<td>14</td>
<td>6</td>
<td>86</td>
<td>7</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>27</td>
<td>61</td>
<td>17</td>
<td>39</td>
<td>44</td>
</tr>
</tbody>
</table>

*Associated lesions: atrioventricular communis, tricuspid atresia, coarctation of the aorta, aortic and/or mitral atresia, pulmonary atresia, total anomalous venous return, interrupted aortic arch.
†Partial transposition of the great vessels: Taussig-Bing malformation and double-outlet right ventricle.

Abbreviations: VSD = ventricular septal defect; SIV = single ventricle; PSI = infundibular pulmonary stenosis; PDA = patent ductus arteriosus.

Table 2

<table>
<thead>
<tr>
<th>Malformation</th>
<th>Total cases (no.)</th>
<th>M/F ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Transposition of great vessels</td>
<td>100</td>
<td>1.63</td>
</tr>
<tr>
<td>Hypoplastic left heart</td>
<td>44</td>
<td>2.67</td>
</tr>
<tr>
<td>Coarctation of the aorta</td>
<td>60</td>
<td>1.81</td>
</tr>
<tr>
<td>Total births§</td>
<td>372,856</td>
<td>1.06</td>
</tr>
</tbody>
</table>

encountered with a presumptive clinical diagnosis of transposition of the great vessels or coarctation of the aorta who did not undergo either catheterization, surgery, or autopsy, this would not be sufficient to significantly change the data or conclusions of the study.

Previous investigators have observed a relatively low frequency of coarctation of the aorta and of valvular and supravalvular aortic stenosis in Ceylon, Taiwan, Thailand, and Japan. In addition to race, environmental or dietary factors may be of importance in explaining the low incidence of these malformations in Far Eastern populations.

Acknowledgment

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References

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