Correspondence

Journal of Medical Genetics, 1981, 18, 158–159

Birth frequency of bilateral renal agenesis

Sir,

In a family study of renal agenesis (J Med Genet 1979; 16: 176–88), we reported an estimate of the birth frequency in 1974 of bilateral renal agenesis, based on death and stillbirth certificates supplied by OPCS, which gave Potter's syndrome, or a not fully specified renal anomaly or agenesis, as an underlying or one of multiple causes of death. For the deaths from 'multiple causes', we had only a 25% sample of certificates and we had no information on stillbirths because of 'multiple causes'; but for deaths and stillbirths where the underlying cause was renal agenesis, we had a 100% sample. The diagnosis was confirmed from necropsy reports in 50 cases. In four instances where a necropsy had been performed, but no report was available, we included the case because the death certificate or the paediatrician stated the cause of death to be renal agenesis. In two instances where no necropsy had been performed, but the death certificate stated Potter's syndrome and congenital abnormalities, we scored each case as a § only. The estimated birth frequency was 0·122 per 1000 total births.

We now report data, applying the same criteria, for the three subsequent years. These are shown in the table together with the data for 1974. In 1977, as in 1974, only a 25% sample of deaths from multiple causes was available and so the number was multiplied by 4.

The birth frequency per thousand total births for each of the four years is: 1974, 0·122; 1975, 0·096; 1976, 0·095; 1977, 0·120. The figures for 1974 and 1975 do not include stillbirths from multiple causes. If, say, an allowance of three further cases is made for this deficit, the birth frequency estimate for 1974 and 1975 would be 0·127 and 0·101, respectively. Inevitably these figures will be somewhat of an underestimate.

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Familial partial 14 trisomy

Sir,

A familial partial 14 trisomy was reported in the February 1979 issue of the Journal. On the basis of banding there were four affected subjects, 47, + der(14), t(9;14)(p24;q21), and three maternal translocation carriers t(9;14)(p24;q21). The chromosomal rearrangement includes triplication of the locus for the enzyme nucleoside phosphorylase (NP). George and Francke have found that NP (purine-nucleoside:orthophosphate ribosyltransferase, EC 2.4.2.1) activity expressed in red blood cells is proportional to the number of alleles present. They assayed erythrocytes from a series of normal controls and subjects with partial 14 duplications to localise this enzyme. Three patients with partial 14 trisomy had NP activities of 21·9, 21·8, and 18·9 units of activity/g haemoglobin and were thought to demonstrate the activity of a triple gene dose. Sixty-one normal persons had a mean activity of 12·7 units, range 7·9 to 18·0. With this assay procedure, our patient DT had 21·7 units of NP activity/g haemoglobin (mean of eight normal people 15·5, range 13·0 to 17·9). These results support the reported karyotypes, the mapping of NP to region 14q13,3

<table>
<thead>
<tr>
<th>Year of birth</th>
<th>Confirmed at necropsy</th>
<th>Stillbirths</th>
<th>Estimate where necropsy not performed or report not available</th>
<th>Estimated total</th>
<th>Total births</th>
<th>Estimated birth frequency per thousand</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>Deaths</td>
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<tr>
<td></td>
<td>Underlying M F</td>
<td>Multiple M F</td>
<td>Underlying M F</td>
<td>Multiple M F</td>
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<tr>
<td>1975</td>
<td>24 10</td>
<td>13 3</td>
<td>1 2</td>
<td>1</td>
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<tr>
<td>1976</td>
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<td>3 0</td>
<td>5 1</td>
<td>2</td>
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<td>2.5</td>
</tr>
<tr>
<td>1977</td>
<td>18 11</td>
<td>3 3</td>
<td>4 0</td>
<td>1</td>
<td>Not available</td>
<td>2</td>
</tr>
</tbody>
</table>

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Birth frequency of bilateral renal agenesis.

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