Frequency of the G6PD nt 1311 C/T polymorphism in English and Iranian populations: relevance to studies of X chromosome inactivation

Y Mortazavi, R Chopra, E C Gordon-Smith, T R Rutherford

Abstract

X chromosome inactivation is widely studied using DNA sequence polymorphisms and DNA methylation as a surrogate measure of inactivation, but the correlation of methylation with inactivation is not perfect. Thus, it may be better to study sequence polymorphisms expressed in the mRNA. A recent paper reported use of a silent C/T polymorphism at nt 1311 of the G6PD cDNA, and this polymorphism was reported to have a frequency of 40% in all ethnic groups. We have screened 218 English and 50 Iranian subjects by PCR and restriction digestion; 53/218 (24%) British and 22/50 (44%) Iranian subjects were heterozygous. Thus, X inactivation studies using this polymorphism may be useful in some populations, including Iran, but much less so in the UK.

Keywords: G6PD; nt 1311 C/T polymorphism; X chromosome inactivation

Table 1 T allele gene frequencies

<table>
<thead>
<tr>
<th>Ref</th>
<th>Population</th>
<th>No of chromosomes studied</th>
<th>T allele gene frequency</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>6</td>
<td>Oriental</td>
<td>59</td>
<td>0.051</td>
<td>p&lt;0.05</td>
</tr>
<tr>
<td>6</td>
<td>Central/South American</td>
<td>30</td>
<td>0.100</td>
<td>NS</td>
</tr>
<tr>
<td>6</td>
<td>White</td>
<td>68</td>
<td>0.132</td>
<td>NS</td>
</tr>
<tr>
<td>6</td>
<td>Sicilian</td>
<td>18</td>
<td>0.167</td>
<td>NS</td>
</tr>
<tr>
<td>6</td>
<td>White Jewish</td>
<td>41</td>
<td>0.222</td>
<td>NS</td>
</tr>
<tr>
<td>6</td>
<td>African</td>
<td>20</td>
<td>0.25</td>
<td>NS</td>
</tr>
<tr>
<td>6</td>
<td>Indian</td>
<td>20</td>
<td>0.45</td>
<td>p&lt;0.05</td>
</tr>
<tr>
<td>5</td>
<td>Mixed “Middle Eastern”</td>
<td>36</td>
<td>0.14</td>
<td></td>
</tr>
<tr>
<td>This</td>
<td>English</td>
<td>436</td>
<td>0.137</td>
<td></td>
</tr>
<tr>
<td>paper</td>
<td>Iranian</td>
<td>100</td>
<td>0.22</td>
<td>p&lt;0.05</td>
</tr>
</tbody>
</table>
useful in populations like that in the UK, owing to the low frequency of heterozygotes.

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