Selective Screening of Phenylketonuria, Tyrosinemia and Maple Syrup Urine Disease in Southern Iran

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Abstract

Inborn errors of amino-acids metabolism and other inherited Mendelian disorders are common in the Middle East. The number of diagnosed inborn errors of amino acid metabolism is growing constantly on account of and availability and improved of analytical techniques. The aim of this work was to determine a rough estimate of the incidence rates of phenylketonuria (PKU), tyrosinemia, and maple syrup urine disease (MSUD) in Fars Province, South of Iran. Using a high performance liquid chromatography, 1044 patients with signs and symptoms suggestive of PKU, tyrosinemia and MSUD were investigated between 1996 and 2001, for the presence of the disorders. Of 1044 patients, 43 cases (4.1%) with PKU, 15 (1.4%) with tyrosinemia and 6 (0.6%) with MSUD were diagnosed. The incidence rates of PKU, tyrosinemia and MSUD in our region is higher than the rates reported from Europe presumably because of the relatively higher rates of consanguinity.


Keywords • Selective screening • inborn errors of amino acid metabolism • phenylketonuria • tyrosinemia • maple syrup urine disease

Introduction

There are reports suggesting that the inborn errors of metabolism such as phenylketonuria (PKU), tyrosinemia and maple syrup urine disease (MSUD) are common in the Middle East.1,4 It is well known that early diagnosis and institution of appropriate treatment hads to drama tie unprovent improves outcome in such patients. However, in many developing countries neonates are not routinely screened for these disorders. In Iran, due to the lack of mandatory screening of newborns, there is no published data available regarding the incidence inborn errors of amino acid metabolism. The current work was conducted to determine the epidemiology of PKU, tyrosinemia and MSUD in, southern Iran, between January 1996 and December 2001. One thousand and forty-four children presenting with signs and symptoms suggestive of PKU, tyrosinemia and MSUD were investigated. The spot blood samples were collected. The age of the newborn at collection time was recorded. The
high performance liquid chromatography (HPLC) technique, was based on the method proposed by Terrlink, et al., with pre-column derivatization of amino acids by o-phthalaldehyde (OPA), separation of the derivatives by reversed phase chromatography and quantitation of amino acids in the blood spot by fluorescence detection. Results from this selective screening study (Table 1) revealed an alarmingly high incidence of PKU, tyrosinemia and MSUD prevails in our region. Of 43 cases with PKU, 15 with tyrosinemia and 6 with MSUD detected in this selective screening study, 34, 11 and 4 children had consanguineous parents, respectively. The consanguineous marriages are quite common in this region. The high incidence of inborn errors of metabolism with high rate of consanguineous marriage has already been reported in the Middle East. Based on the obtained results, it seems that a mass screening programme for early diagnosis of inborn error of amino acid metabolism should be instituted in this region.

Acknowledgement

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References


Table 1: The incidence of PKU, tyrosinemia and MSUD from January 1996 to December 2001 in Fars Province, South of Iran

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Numbers of cases diagnosed</th>
<th>Numbers of cases born and diagnosed from 1996-2001</th>
<th>Incidence (95% CI) per 100,000 births</th>
</tr>
</thead>
<tbody>
<tr>
<td>PKU</td>
<td>43</td>
<td>29</td>
<td>27.2 (17.3–37.1)</td>
</tr>
<tr>
<td>Tyrosinemia</td>
<td>15</td>
<td>10</td>
<td>9.4 (3.8–15.2)</td>
</tr>
<tr>
<td>MSUD</td>
<td>6</td>
<td>5</td>
<td>4.7 (0.6–8.8)</td>
</tr>
</tbody>
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