Dietary treatment in PKU from experience to evidence
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Plasma phenylalanine in patients with phenylketonuria self managing their diet

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Dietary adherence in phenylketonuria (PKU) – measured as plasma phenylalanine concentrations (Phe) - is a major issue. In the so-called professionally steered situation, it was our practice to take all blood samples for measurement of Phe during patients’ hospital visits. Phe concentrations were interpreted by the paediatrician and the dietician phoned patients/parents with dietary advice. Self-management has been suggested to improve dietary adherence. We report Phe concentrations during our first 6 months experiences with self-management in 48 PKU patients above 1 year of age.

During this period, patients decided frequency of blood sampling, and sent samples (filter paper) to the laboratory by post. A nurse without knowledge of PKU phoned the results to patients/parents without interpretation/advice. Patient/parents decided independently regarding adjustment, but could phone the dietician for advice one day later. The clinical team received all blood test results weekly. When Phe concentrations were frequently deemed unsatisfactory, the dietician called the parent/patient. During hospital visits (Table 1), Phe concentrations, patients’ adjustments, and the protein substitute were evaluated.

Self-management was introduced following 1–2 individual meetings of patients with staff, two group sessions, and provision of written information, including advice regarding frequency of blood sampling, appropriate Phe concentrations (adapted from the British recommendations), and dietary adjustments (Table 1). As this was a change in management policy rather than a research study, there was no control group. Therefore, results of six months self-management (456 samples in 48 patients) (Wilcoxon test) were compared with those obtained over three years beforehand (1152 samples in 48 patients).

Table 1. Advised ranges for plasma Phe concentration and frequency for follow-up.

<table>
<thead>
<tr>
<th>Age (years)</th>
<th>Plasma Phe concentration (μmol/l)</th>
<th>Blood sampling</th>
<th>Clinical follow-up</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Old</td>
<td>New</td>
<td>Old</td>
</tr>
<tr>
<td>First year</td>
<td>120-240</td>
<td>120-240</td>
<td>Fortnightly</td>
</tr>
<tr>
<td>1-4</td>
<td>200-500</td>
<td>120-360</td>
<td>Monthly</td>
</tr>
<tr>
<td>5-10</td>
<td>200-500</td>
<td>120-480</td>
<td>Every 6-8 weeks</td>
</tr>
<tr>
<td>11-15</td>
<td>200-500</td>
<td>120-480</td>
<td>Every 6-8 weeks</td>
</tr>
<tr>
<td>&gt; 15</td>
<td>200-500</td>
<td>120-600</td>
<td>Every 3 months</td>
</tr>
</tbody>
</table>

Old refers to the situation steered by professionals. New refers to the situation of self-management.
Plasma phenylalanine in patients with phenylketonuria

During self-management, most patients took blood samples according to the recommended frequency. The Phe intake changed in 10 patients (mean change 2.3%, SD 5.7%). The largest differences in intake were 32% and 16% of the total Phe daily given in mg/day, observed in 2 patients with changes in growth velocity during puberty.

Parents and professionals adjusted the diet in a comparable way. The dietician was phoned by families once or twice a week, while she phoned them twice a month. In patients aged 1-4 year and 11-15 year the Phe concentrations rose (Table 2). The median proportion of samples within the advised range remained comparable, largely because of the decrease frequency of Phe concentrations below the target range.

Various explanations can be given for the increase in Phe concentrations during self-management, including the normal rise with age, increased frequency of blood sampling, a tendency for patients to attempt a dietary intake compatible with Phe concentrations just below the advised upper limit, and sampling after an overnight fast rather than later in the day during the professionally steered situation.

The question whether this rise in Phe concentrations is important is hard to answer. The mean of the medians within 1-4 years aged patients rose of 214 to 327 µmol/l but remained lower than most of the reported experiences. Younger the patients are more vulnerable to higher Phe concentrations, but the importance of variable

Table 2. Plasma phenylalanine concentration and proportion of samples with plasma phenylalanine above the recommended range in the situation steered by professionals and self-management.

<table>
<thead>
<tr>
<th>Age in years</th>
<th>Mean plasma phenylalanine concentration (µmol/l) during situation</th>
<th>Percentage samples with Phe above recommended range during situation</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Steered by Professionals</td>
<td>Self-management</td>
</tr>
<tr>
<td>1-4 (n=11)</td>
<td>214 (183-995)</td>
<td>327 (189-1007)</td>
</tr>
<tr>
<td>5-10 (n=12)</td>
<td>309 (186-1414)</td>
<td>326 (208-1282)</td>
</tr>
<tr>
<td>11-15 (n=14)</td>
<td>315 (155-433)</td>
<td>392 (277-582)</td>
</tr>
<tr>
<td>&gt; 15 (n=11)</td>
<td>587 (290-960)</td>
<td>649 (337-1266)</td>
</tr>
<tr>
<td>All ages (n=48)</td>
<td>320 (337-1266)</td>
<td>382 (189-1282)</td>
</tr>
</tbody>
</table>

Results expressed as median (range)
*(Wilcoxon)
time periods of Phe concentrations far below 360 μmol/l below 10 years of age is uncertain.\textsuperscript{4,6}

In conclusion, this is the first study on the effect of self-management on plasma Phe in PKU has shown that self-management is a viable option, but further investigation of the effects and safety is warranted.

ACKNOWLEDGEMENTS

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REFERENCES