Psychosocial factors and metabolic control in PKU patients followed-up at CHUPorto-Portugal before and after full subsidy of dietary products used in the nutritional treatment

Francisca Ferreira Almeida Morais¹, Ricardo Costa¹,⁵, Esmeralda Martins²,³,⁴, Carla Carmona²,³,⁴, Manuela Ferreira Almeida²,³,⁴
¹ Faculdade de Ciências da Saúde, University of Beira Interior, Covilhã, Portugal
² Centro Genética Médica, Centro Hospitalar Universitário Porto, Porto, Portugal
³ Reference Center for Hereditary Diseases Metabolism, CHUP, Porto, Portugal
⁴ Unit for Multidisciplinary Research in Biomedicine, Porto, Portugal
⁵ Pediatric Department, Centro Hospitalar Universitário Cova da Beira, Covilhã, Portugal

Introduction

A natural protein and phenylalanine (Phe)-restricted diet, supplemented with protein substitutes and special low protein foods (SLPF) remains the mainstay treatment of phenylketonuria (PKU). High Phe levels are associated with neurotoxic effects that may interfere with neurocognitive development and socio-affective outcomes. To facilitate diet compliance, SLPF were fully reimbursed in Portugal since June 2005.

Our aim was to compare PKU patients before and after the full reimbursement of SLPF taking into account their metabolic control, cognitive and psychosocial outcomes.

Methods

❖ A retrospective observational study was conducted on 52 PKU patients considering two age groups: Group A - <12 years and group B - ≥12 years at 01/01/2006, in two periods (01/01/1997-31/12/2005, before reimbursement; 01/01/2006-31/12/2014 after reimbursement).
❖ The inclusion criteria were: PKU patients early diagnosed at the neonatal screening, followed-up at Centro Hospitalar Universitário do Porto (CHUPorto), with blood Phe levels at screening ≥6 mg/dL and following a restricted Phe diet supplemented with SLPF.

The exclusion criteria were: PKU patients with blood Phe levels at screening <6 mg/dL, born after 2006, who do not follow a Phe-restricted diet since diagnosis, without consumption of SLPF, with coexistence of other pathologies that interfere with physical and neurocognitive development and late diagnosed.
❖ Gender, age, PKU classification, quality of dietetic control (QDC): [average of annual medians Phe levels (good QDC: <12 years – ≤ 6 mg/dL; ≥12 years - ≤ 8 mg/dL)], global quotient development (QD)/intelligence quotient (IQ), school curriculum and comorbidities were evaluated.
**Results**

<table>
<thead>
<tr>
<th>GROUP</th>
<th>Mean Age</th>
<th>N</th>
</tr>
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<tbody>
<tr>
<td>Group A &lt;12 years</td>
<td>5.88</td>
<td>26</td>
</tr>
<tr>
<td>Group B ≥12 years</td>
<td>17.62</td>
<td>26</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>52</td>
</tr>
</tbody>
</table>

*Table 1. Mean age by age group*

**Figure 1. Gender distribution by age group**

**Figure 2. PKU classification by age group**
Figure 5. Metabolic control by period

Figure 6. Distribution of global IQ values by period
Figure 3. Distribution of curriculum type by age group in each period

Figure 4. Distribution of comorbidities by age group in each period
Summary

✓ Full subsidy improved patients dietary compliance, as evidenced by the improvement in metabolic control parameters in both age groups. This effect was higher in group B, who had worse metabolic control prior to the reimbursement.

✓ The IQ of the group A (<12y) improved after the reimbursement. This could be a result of the better dietary compliance, with a long-term effect on the IQ of these younger patients.

✓ Regarding the school curriculum and comorbidities, we observed that the full subsidy did not have a significant effect on any of these parameters, and they remained similar. A more comprehensive study is recommended to allow conclusions regarding a more extended period of time, which allows to assess particular changes in these variables.

✓ Full reimbursement of SLPF improved the dietary compliance resulting on a better metabolic control and allowing a better neurocognitive and socioaffective development. Studies taking into account others variables that may interfere with these outcomes should be done.